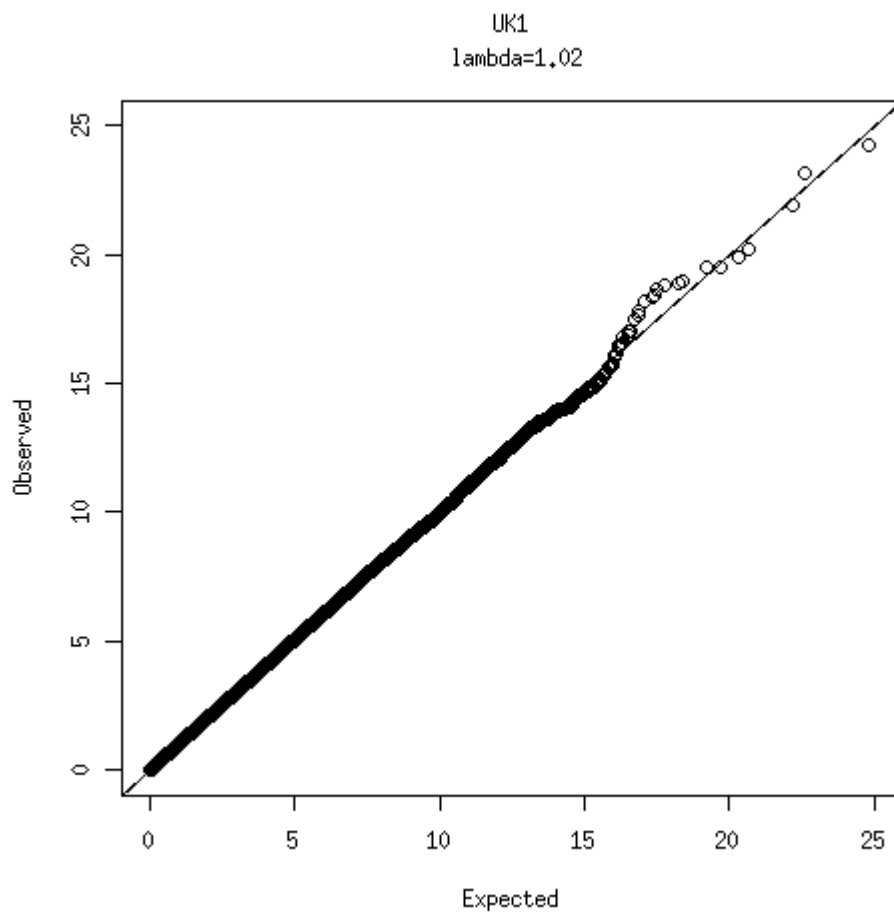


|                 | UK1                               | UK2                                   | Scotland1                             | Scotland2                             | VQ58                                  |
|-----------------|-----------------------------------|---------------------------------------|---------------------------------------|---------------------------------------|---------------------------------------|
| <b>pre-QC</b>   | <b>940 cases<br/>965 controls</b> | <b>2,873 cases<br/>2,871 controls</b> | <b>1,012 cases<br/>1,012 controls</b> | <b>2,057 cases<br/>2,111 controls</b> | <b>1,800 cases<br/>2,690 controls</b> |
| Call rate       | 15                                | 30                                    | 15                                    | 22                                    | 0                                     |
| Ethnicity       | 54                                | 6                                     | 9                                     | 7                                     | 0                                     |
| Relatedness     | 26                                | 197*                                  | 9                                     | 30*                                   | 9                                     |
| Sex discrepancy | 3                                 | 45                                    | 15                                    | 22                                    | 1                                     |
| Other           | 17                                | 9                                     | 5                                     | 5                                     | 0                                     |
| <b>post-QC</b>  | <b>890 cases<br/>900 controls</b> | <b>2,659 cases<br/>2,798 controls</b> | <b>973 cases<br/>998 controls</b>     | <b>2,007 cases<br/>2,075 controls</b> | <b>1,794 cases<br/>2,686 controls</b> |

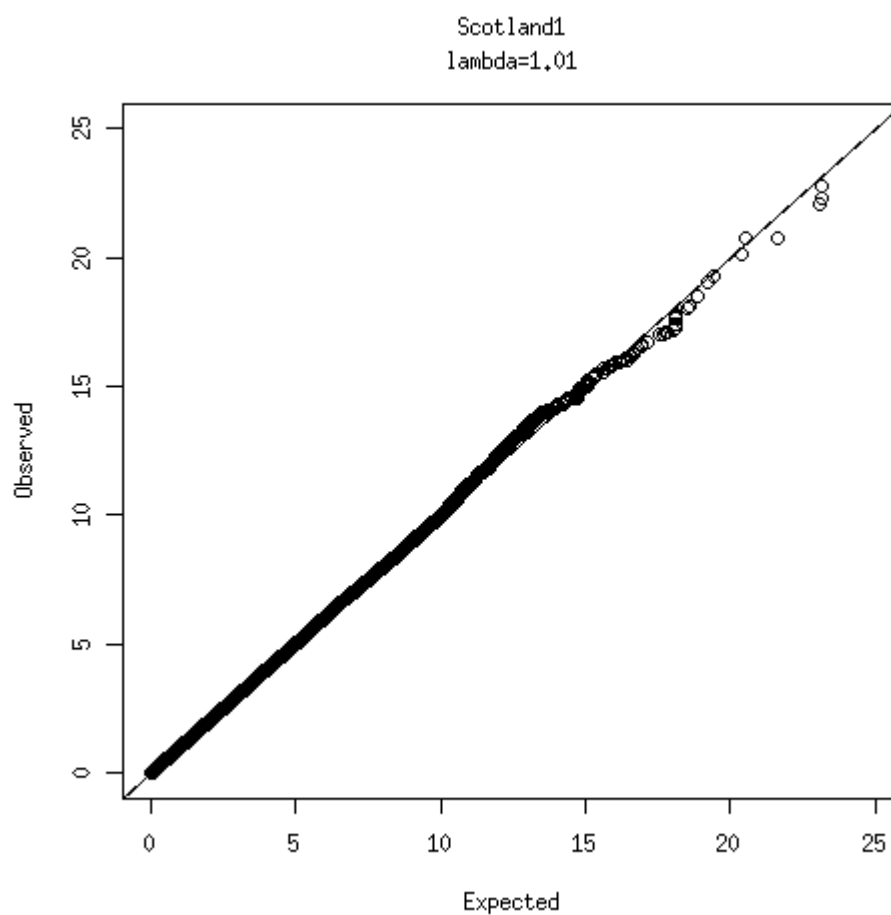
**Supplementary Figure 1: Details of the quality control filters applied to each GWAS.** Samples were excluded - call rate (<95%), Ethnicity (principle components analysis or self-reported not to be of European descent), relatedness (duplicates or related within or between each case-control series), sex discrepancy, other (cases found to carry a high-risk CRC mutation, controls with a 1<sup>st</sup> degree relative with CRC, low concordance of genotypes in duplicates, subjects withdrawn from the study). \*samples preferentially removed from these data-sets over GWAS datasets.

**Supplementary Figure 2: Quantile-Quantile (Q-Q) plots of observed and expected  $\chi^2$  values of association between SNP genotype and colorectal cancer risk. (a) UK1, (b) Scotland1, (c) UK2, (d) Scotland2 and (e) VQ58**

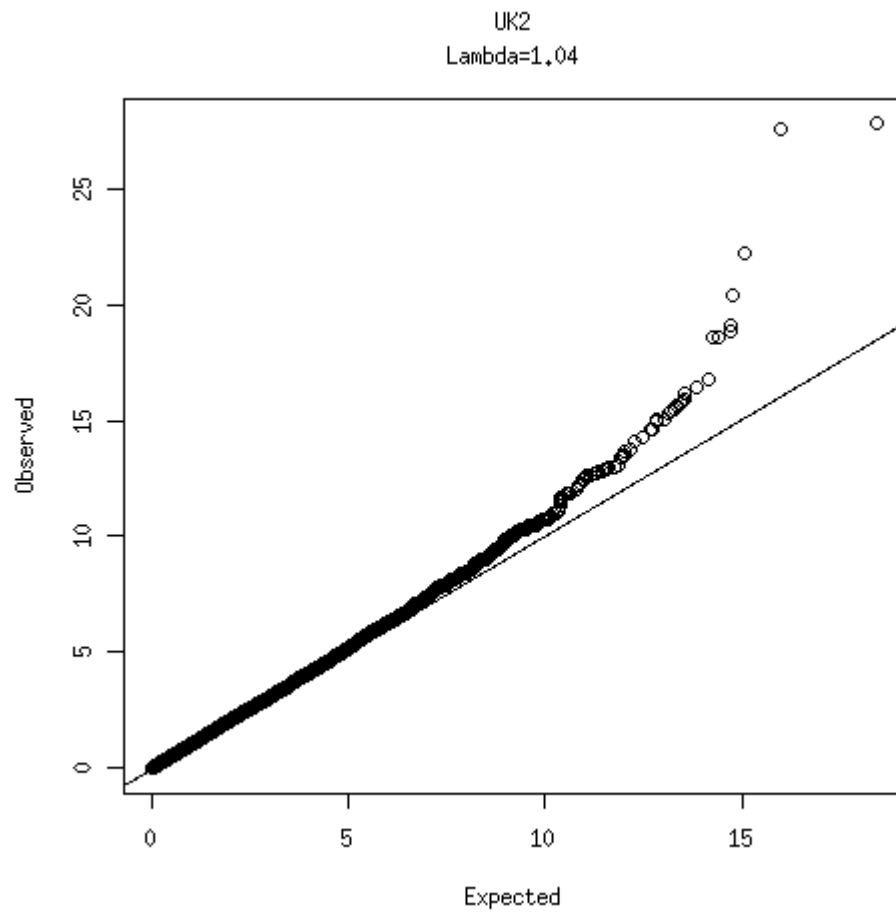
(a)



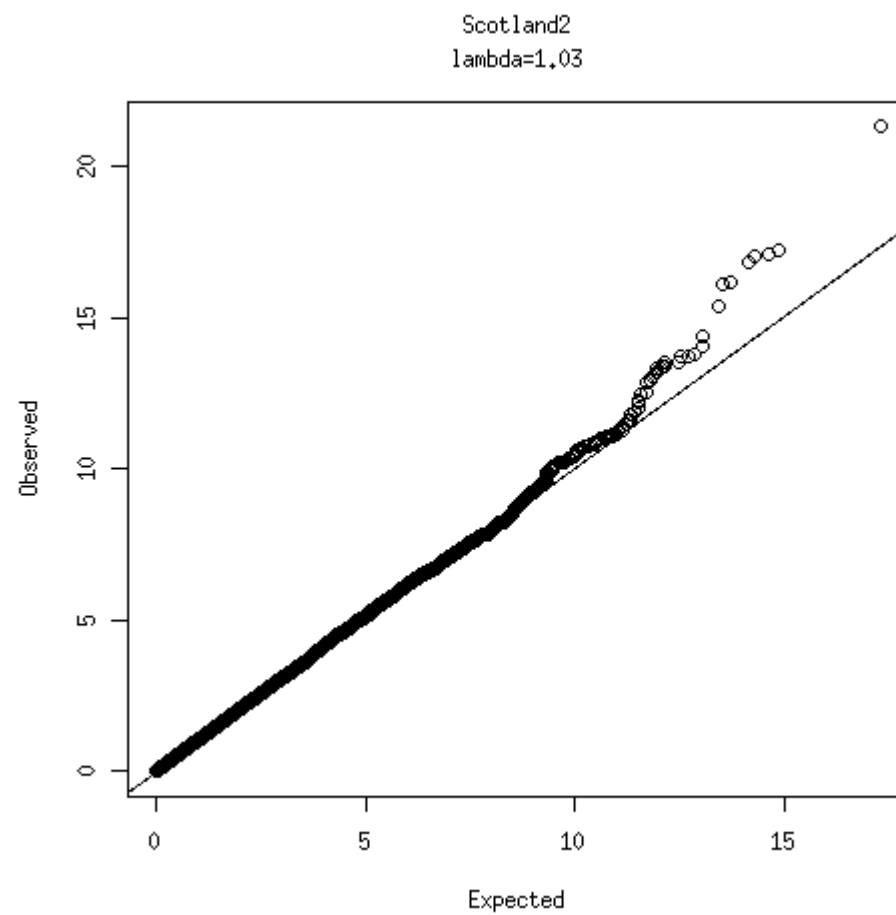
(b)



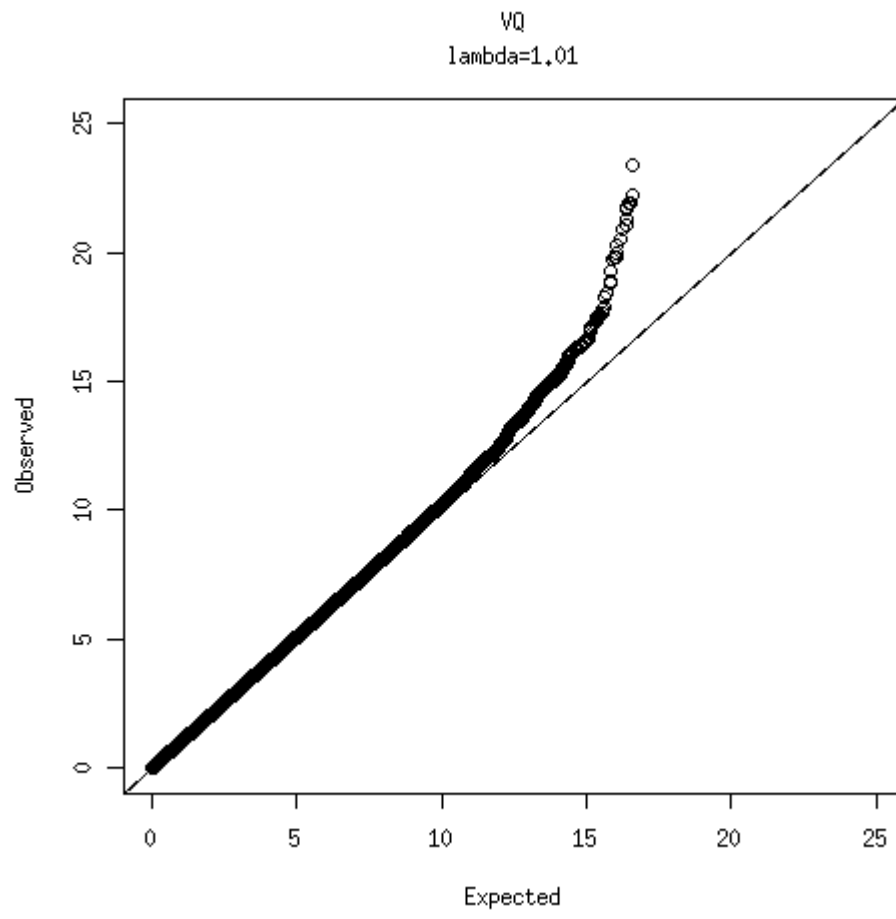
(c)



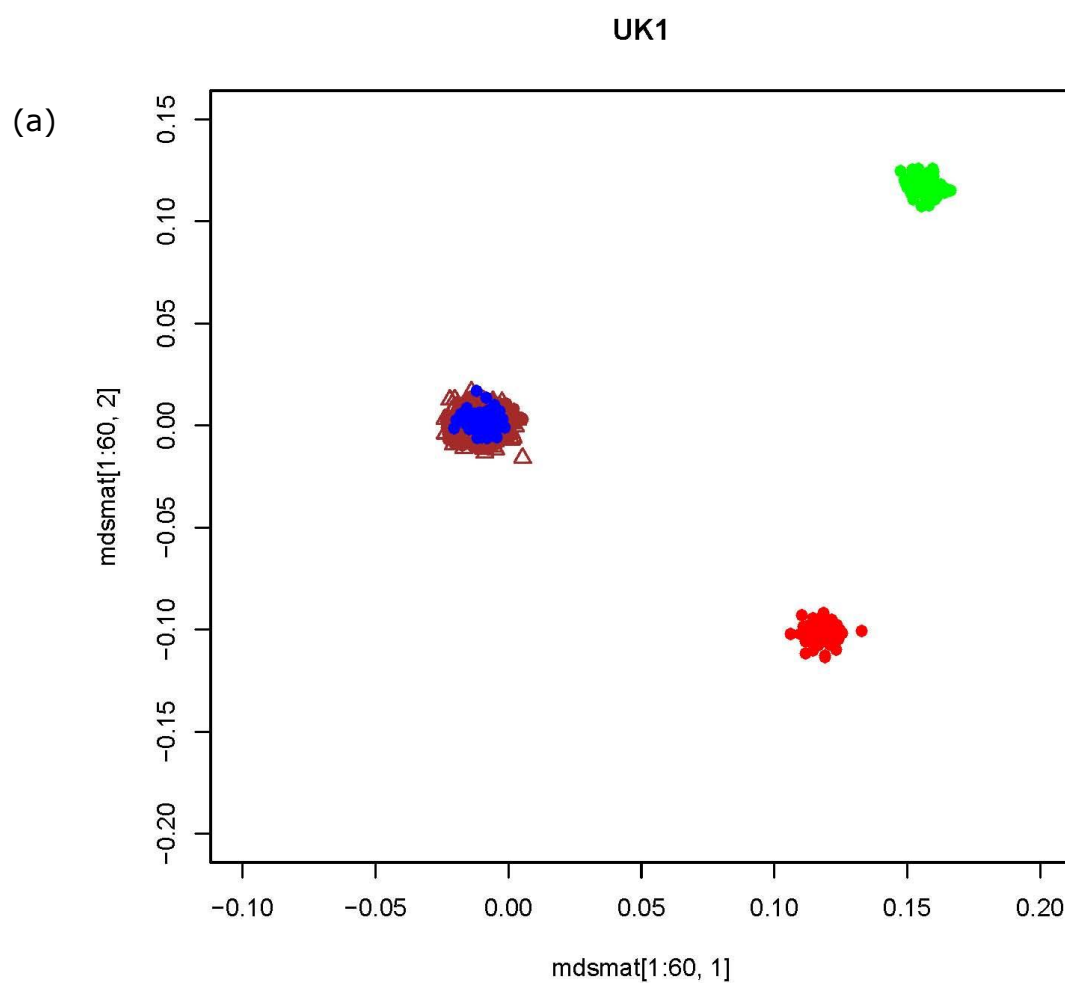
(d)



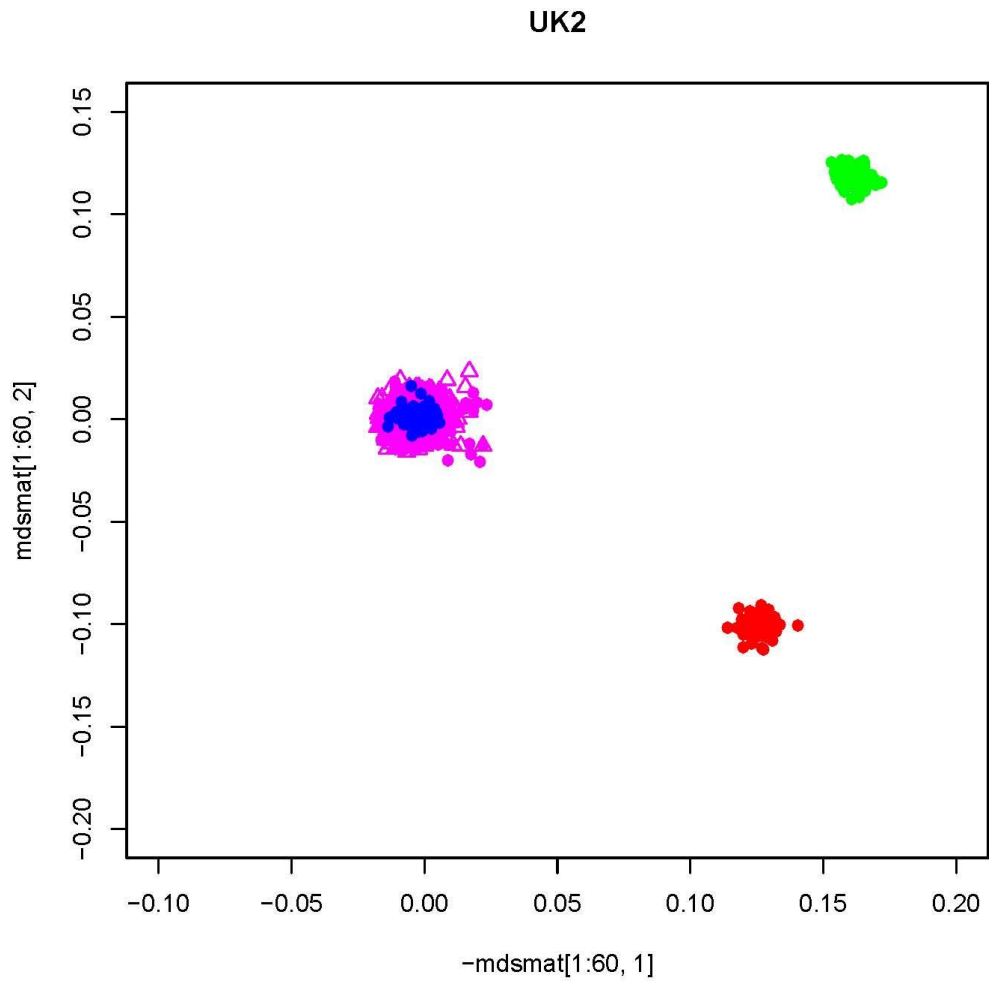
(e)



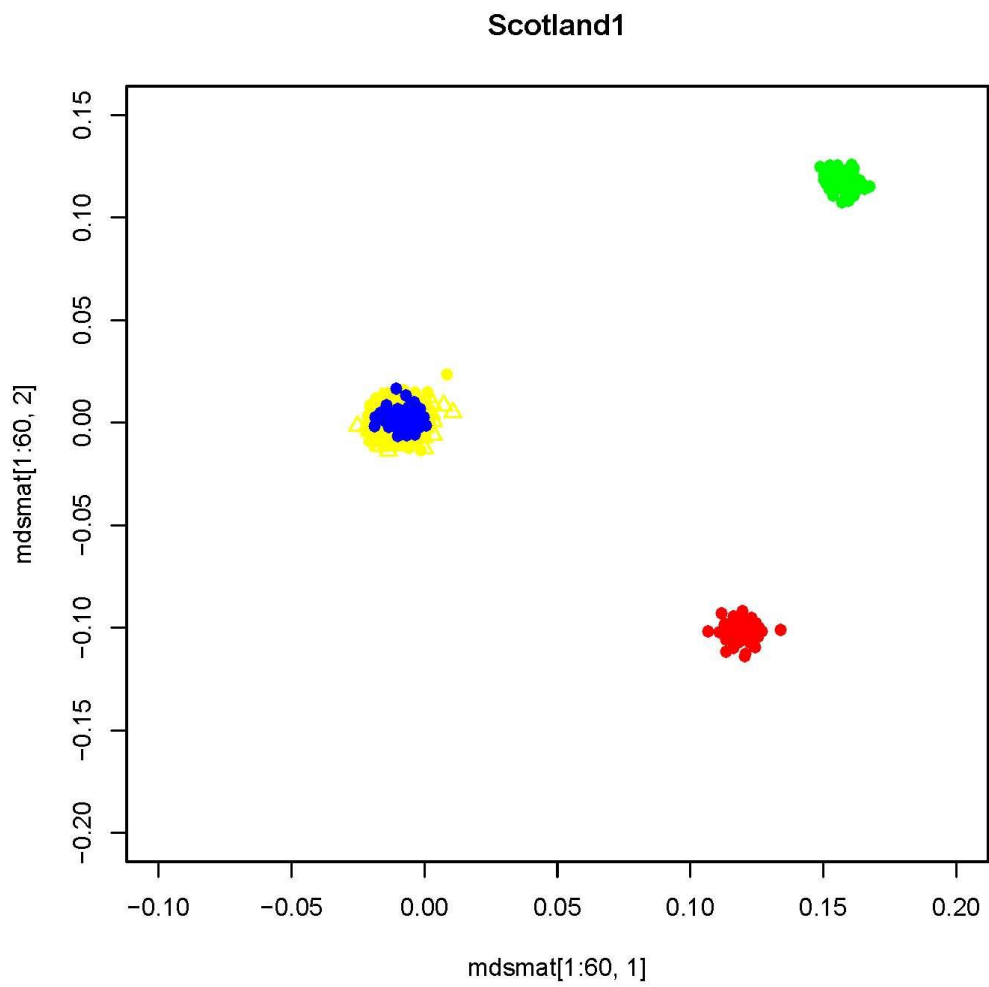
**Supplementary Figure 3: Identification of individuals in the GWAS of non-European ancestry in cases and controls.** The first two principal components of the analysis are plotted, before exclusions, for the (a) UK1, (b) UK2, (c) Scotland1, (d) Scotland2 and (e) VQ58 cases and controls. HapMap CEU individuals are plotted in blue; CHB+JPT individuals are plotted in green; YRI individuals are plotted in red; GWAS cases are plotted as circles and controls as triangles.



(b)

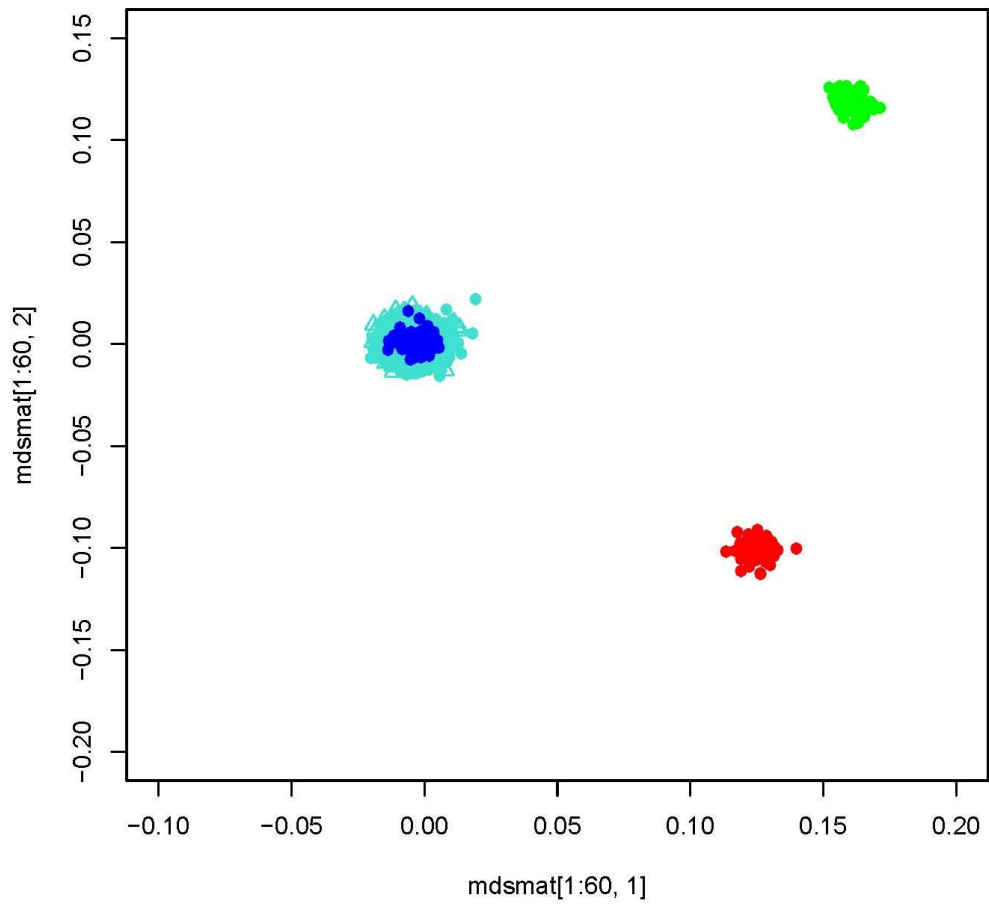


(c)



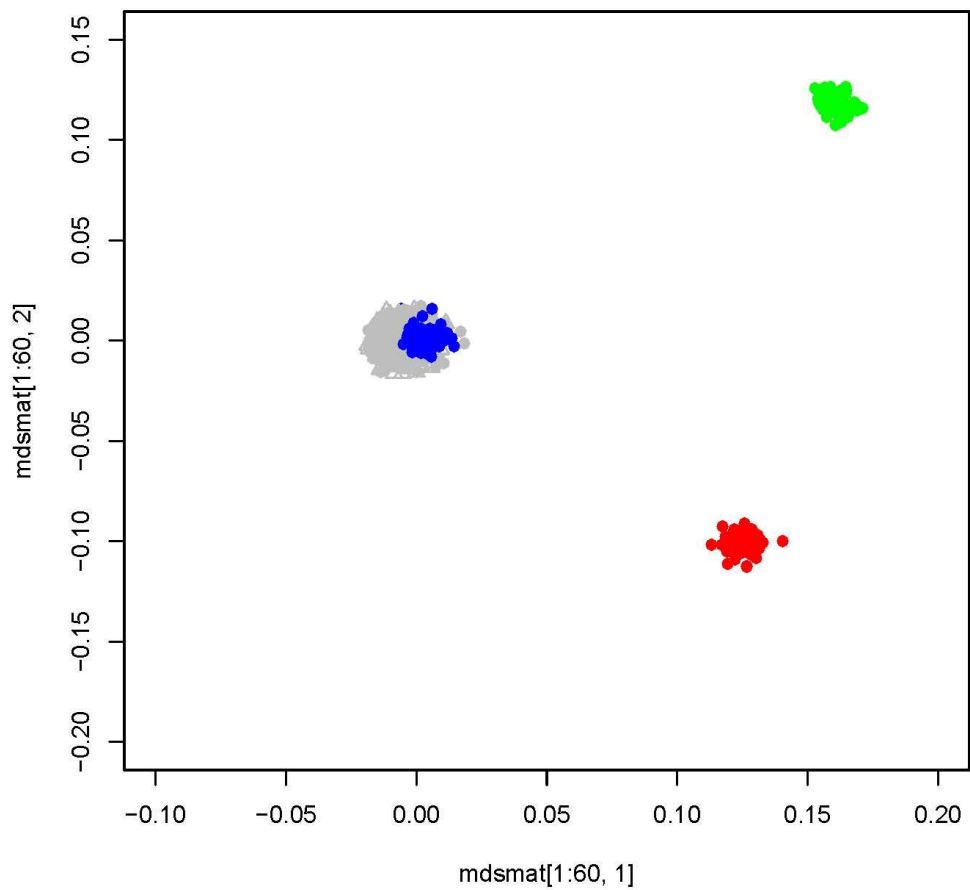
(d)

Scotland2

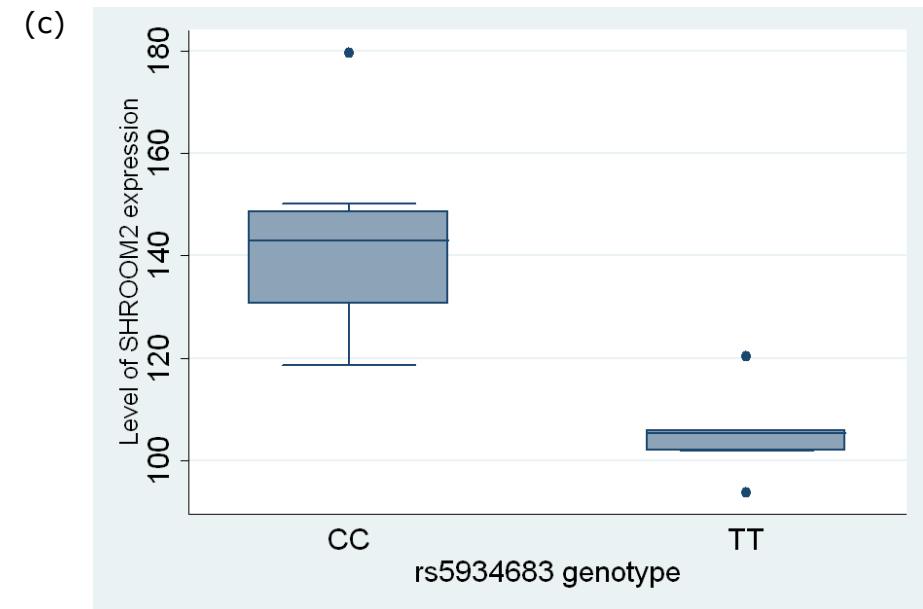
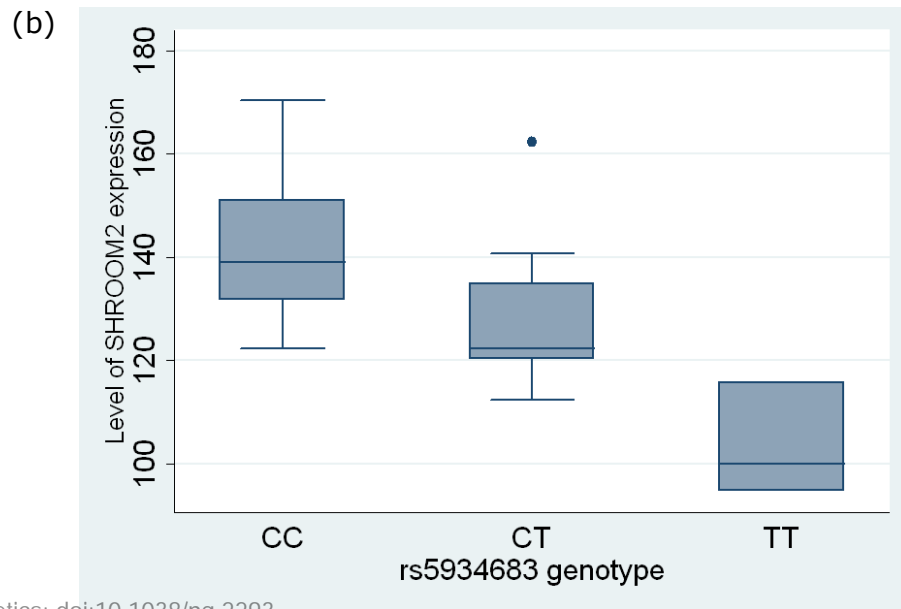
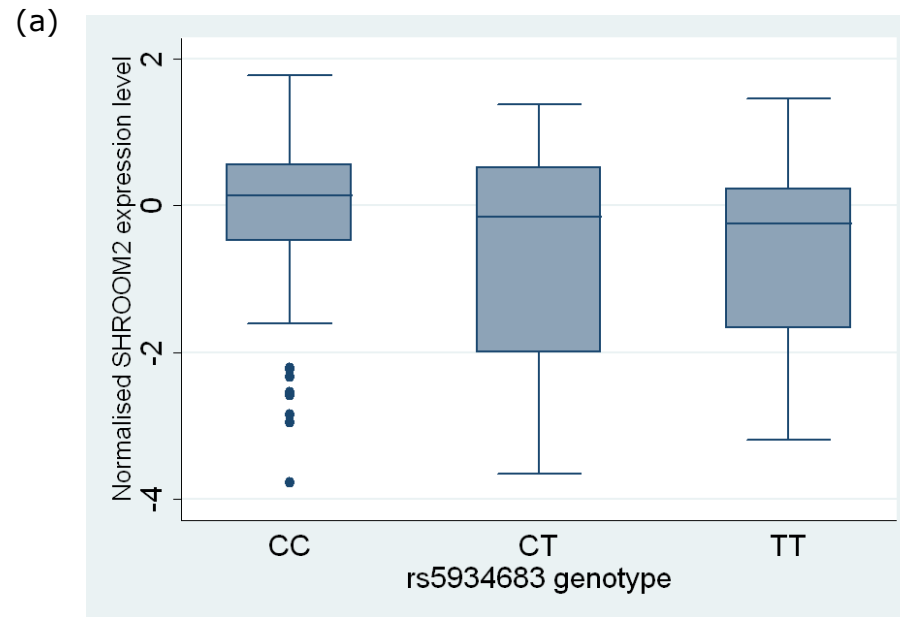


(e)

VQ58



**Supplementary Figure 4 : Relationship between rs5934683 genotype and *SHROOM2* expression.** Differences in the distribution of expression by SNP (a) 154 CRCs from The Cancer Genome Atlas (TCGA) project, (b-c) 42 colon tissue samples, (b) females and (c) males.





**Supplementary Table 1: Summary of the sample sets used in the study.**

| Series                  | Study setting  | Study centre                      | Sampling   | Genotyping platform  | No. cases    | No. controls |
|-------------------------|--|-----------------------------------|--|--|--------------|--------------|
| <b>GWAS studies</b>     |  |                                   |  |  |              |              |
| UK1                     | CORGI (Colorectal Tumour Gene Identification Consortium)   | Oxford University                 | Cases enriched for family history of CRC ascertained through clinical genetics centres in the UK. Spouse controls with no personal family history of CRC.  | Illumina HumanHap550   | 940          | 965          |
| UK2                     | NSCCG (National study of Colorectal Cancer). <a href="http://www.icr.ac.uk/research/research_divisions/Genetics_and_Epidemiology/index.shtml">http://www.icr.ac.uk/research/research_divisions/Genetics_and_Epidemiology/index.shtml</a>   | Institute of Cancer Research      | Population-based UK study. Spouse controls from NSCCG and GELCAPS (Genetic Lung Cancer Predisposition Study).  | Illumina iSelect and Goldengate  | 2,873        | 2,871        |
| SCOTLAND1               | COGS (Colorectal Cancer Genetics Susceptibility Study)   | Edinburgh University              | Population-based incident case series aged <55 at diagnosis; Scotland. Population-based controls frequency matched by area of residence within Scotland  | Illumina HumanHap300<br>Illumina HumanHap240S                                    | 1,012        | 1,012        |
| SCOTLAND2               | SOCCS (Scottish Colorectal Cancer Study)   | Edinburgh University              | Population-based incident case series; Scotland. Population-based controls frequency matched by area of residence within Scotland.   | Illumina iSelect and Goldengate  | 2,057        | 2,111        |
| VQ58                    | Cases: VICTOR, post treatment stage of a phase III, randomised controlled trial of rofecoxib (VIOXX) in CRC patients after potentially curative therapy. QUASAR2, multicentre study if capecitabine +/- bevacizumab as adjuvant CRC treatment. <a href="http://www.octo-oxford.org.uk/alltrials">http://www.octo-oxford.org.uk/alltrials</a> . Controls: 58BC (UK 1958 Birth Cohort) <a href="http://www.b58cgene.sgul.ac.uk">http://www.b58cgene.sgul.ac.uk</a> | Oxford University                 | Cases recruited as a clinical-based series and controls as population-based series.  | Illumina HumanHap300<br>Illumina HumanHap370<br>Illumina Human 1.2MDuo Custom_V1 | 1,800        | 2,690        |
| <b>TOTAL</b>            | <b>GWAS Sets</b>   |                                   |  |  | <b>8,682</b> | <b>9,649</b> |
| <b>Non-GWAS studies</b> |  |                                   |  |  |              |              |
| COINBNS                 | COIN, COIN-B <a href="http://public.ukcrn.org.uk">http://public.ukcrn.org.uk</a><br>NBS UK National Blood Service Blood Donor samples) <a href="http://www.wtccc.org.uk">http://www.wtccc.org.uk</a>   | University of Cardiff             | Multicentre study of cetuximab and other therapies in metastatic CRC. Controls were unselected UK blood donors   | Illumina Goldengate<br>Illumina Human 1.2MDuo Custom_V1                          | 2,183        | 2,501        |
| CCFR1                   | CCFR (Colon Cancer Family Registry) <a href="http://epi.grants.cancer.gov/CFR">http://epi.grants.cancer.gov/CFR</a>  | University of Southern California | Recently diagnosed CRC cases reported to population complete cancer registries in the USA (Seattle Familial Colorectal Cancer Registry), Canada (Ontario Familial Cancer Registry) and Australia (Australasian Colorectal Cancer Family Study). Population based controls. | Illumina HumanHap 550  | 1,290        | 1,055        |

|              |   |   |   |   |               |               |
|--------------|---|---|---|---|---------------|---------------|
| UK3          | NSCCG (National study of Colorectal Cancer).<br><a href="http://www.icr.ac.uk/research/research_divisions/Genetics_and_Epidemiology/index.shtml">http://www.icr.ac.uk/research/research_divisions/Genetics_and_Epidemiology/index.shtml</a> | Institute of Cancer Research                            | Population-based UK study. Spouse controls from NSCCG and GELCAPS (Genetic Lung Cancer Predisposition Study).   | KASPar  | 7,862         | 4,431         |
| UK4          | CORGI (Colorectal Tumour Gene Identification Consortium)  | Oxford University                                       | Cases enriched for family history of CRC ascertained through clinical genetics centres in the UK. Spouse controls with no personal family history of CRC. | KASPar  | 589           | 1,087         |
| SCOTLAND3    | SOCCS3 Study  | Edinburgh University                                    | CRC cases arising within the Lothian region of Scotland. Cancer free population controls from Lothian born 1921 and 1936 (LBC1921 and LBC1936).           | Taqman  | 736           | 1,522         |
| CAMBRIDGE    | UKSEARCH (Studies of Epidemiology and Risk Factors in Cancer Heredity)<br><a href="http://www.srl.cam.ac.uk/search">http://www.srl.cam.ac.uk/search</a>   | Cambridge University                                    | Population-based case-control study   | Kaspar  | 2,248         | 2,288         |
| CROATIA      | Croatian Colorectal Cancer Genetic Study  | University Hospital Centre 'Sestre milosrdnice', Zagreb | Hospital based recruited from University Hospital in Zagreb, Croatia. Urban Croatian population from Split, Croatia.                                      | Taqman  | 375           | 1,051         |
| HELSINKI     | FCCPS (Finnish Colorectal Cancer Predisposition Study)<br><a href="http://research.med.helsinki.fi/gsb/aaltonen">http://research.med.helsinki.fi/gsb/aaltonen</a>   | University of Helsinki, Finland                         | Population-based study, south-eastern Finland   | KASPar  | 1,013         | 827           |
| SWEDEN       | Swedish Low Risk Colorectal Cancer Study  | Karolinska Institute                                    | Cases ascertained from 14 different surgical clinics in Sweden between 2004 and 2006. Controls were blood donors or healthy volunteers from Uppsala.      | Taqman  | 3,217         | 2,895         |
| JAPAN        | Biobank Japan project   | The University of Tokyo                                 | Cases and controls were ascertained from 66 hospitals in Japan. Controls included subjects with non-cancer diseases or healthy volunteers.                | Illumina Human 10Quad, Illumina HumanHap550v3 | 1,583         | 1,898         |
| <b>TOTAL</b> | <b>Non-GWAS Studies</b>   |   | <b>Various populations</b>  |   | <b>21,096</b> | <b>19,555</b> |
| <b>TOTAL</b> | <b>COMBINED STUDIES</b>   |   |   |   | <b>29,778</b> | <b>29,204</b> |

**Supplementary Table 2: Summary results for the SNPS: rs1321311 (6p21), rs3824999 (11q13.4) and rs5934683 (Xp22.2) associated with CRC risk.** <sup>a</sup>Risk allele frequency (RAF), genotype counts are given in brackets. <sup>b</sup>Odds ratio. <sup>c</sup>95% Confidence Interval.

**rs1321311 (6p21.31)**

| STUDY           | Cases                           | Controls             | OR <sup>b</sup> | 95% CI <sup>c</sup> | P-value                      |
|-----------------|---------------------------------|----------------------|-----------------|---------------------|------------------------------|
|                 | RAF <sup>a</sup> (AA / AC / CC) | RAF (AA / AC / CC)   |                 |                     |                              |
| UK1             | 0.26 (53/363/473)               | 0.23 (49/317/534)    | 1.20            | 1.03-1.40           | 2.00x10 <sup>-2</sup>        |
| UK2             | 0.25 (169/970/1519)             | 0.23 (149/996/1652)  | 1.08            | 0.99-1.18           | 7.23x10 <sup>-2</sup>        |
| SCOTLAND1       | 0.25 (65/356/552)               | 0.22 (53/331/614)    | 1.18            | 1.02-1.37           | 2.45x10 <sup>-2</sup>        |
| SCOTLAND2       | 0.24 (109/741/1157)             | 0.23 (110/736/1229)  | 1.05            | 0.95-1.16           | 3.60x10 <sup>-1</sup>        |
| VQ58            | 0.24 (108/634/1052)             | 0.23 (141/955/1588)  | 1.04            | 0.94-1.14           | 4.80x10 <sup>-1</sup>        |
| COINNBS         | 0.25 (135/810/1201)             | 0.23 (128/892/1481)  | 1.13            | 1.03-1.24           | 1.26x10 <sup>-2</sup>        |
|                 |                                 |                      | <b>1.09</b>     | <b>1.05-1.14</b>    | <b>4.79 x10<sup>-5</sup></b> |
| CCFR1           | 0.27 (89/449/644)               | 0.24 (65/352/575)    | 1.12            | 0.98-1.28           | 9.83x10 <sup>-2</sup>        |
| UK3             | 0.24 (449/2836/4368)            | 0.23 (227/1531/2541) | 1.08            | 1.01-1.14           | 2.26x10 <sup>-2</sup>        |
| UK4             | 0.23 (29/217/343)               | 0.24 (35/264/397)    | 0.96            | 0.8-1.16            | 6.94x10 <sup>-1</sup>        |
| SCOTLAND3       | 0.25 (49/267/404)               | 0.23 (82/535/905)    | 1.14            | 0.98-1.31           | 8.17x10 <sup>-2</sup>        |
| CAMBRIDGE       | 0.25 (143/826/1279)             | 0.23 (116/789/1338)  | 1.11            | 1.01-1.23           | 2.86x10 <sup>-2</sup>        |
| CROATIA         | 0.27 (25/156/194)               | 0.25 (67/374/594)    | 1.16            | 0.96-1.41           | 1.16x10 <sup>-1</sup>        |
| HELSINKI        | 0.21 (44/334/621)               | 0.17 (20/245/550)    | 1.27            | 1.07-1.50           | 5.63x10 <sup>-3</sup>        |
| SWEDEN          | 0.22 (132/1092/1900)            | 0.21 (128/872/1705)  | 1.05            | 0.96-1.15           | 2.61x10 <sup>-1</sup>        |
|                 |                                 |                      | <b>1.09</b>     | <b>1.06-1.13</b>    | <b>1.12x10<sup>-9</sup></b>  |
| JAPAN           | 0.14 (26/390/1167)              | 0.12 (38/380/1479)   | 1.18            | 1.03-1.36           | 1.71x10 <sup>-2</sup>        |
|                 |                                 |                      |                 |                     |                              |
| <b>Combined</b> |                                 |                      | <b>1.10</b>     | <b>1.07-1.13</b>    | <b>1.14x10<sup>-10</sup></b> |

**rs3824999 (11q13.4)**

| STUDY           | Cases                           | Controls              | OR <sup>d</sup> | 95% CI <sup>c</sup> | P-value                      |
|-----------------|---------------------------------|-----------------------|-----------------|---------------------|------------------------------|
|                 | RAF <sup>a</sup> (AA / AC / CC) | RAF (AA / AC / CC)    |                 |                     |                              |
| UK1             | 0.53 (194/443/253)              | 0.50 (231/438/231)    | 1.14            | 1.00-1.30           | 4.90x10 <sup>-2</sup>        |
| UK2             | 0.52 (624/1310/724)             | 0.50 (699/1423/676)   | 1.10            | 1.02-1.18           | 1.66x10 <sup>-2</sup>        |
| SCOTLAND1       | 0.53 (216/489/268)              | 0.50 (244/516/238)    | 1.13            | 1.00-1.28           | 5.94x10 <sup>-2</sup>        |
| SCOTLAND2       | 0.50 (505/998/504)              | 0.51 (486/1045/544)   | 0.94            | 0.87-1.03           | 1.99x10 <sup>-1</sup>        |
| VQ58            | 0.53 (376/920/498)              | 0.49 (704/1318/653)   | 1.19            | 1.09-1.30           | 5.48x10 <sup>-5</sup>        |
| COINNBS         | 0.52 (519/1063/601)             | 0.50 (627/1241/633)   | 1.07            | 0.99-1.16           | 9.21x10 <sup>-2</sup>        |
|                 |                                 |                       | <b>1.08</b>     | <b>1.05-1.13</b>    | <b>1.77 x10<sup>-5</sup></b> |
| CCFR1           | 0.54 (241/606/326)              | 0.50 (242/510/247)    | 1.15            | 1.02-1.30           | 2.45x10 <sup>-2</sup>        |
| UK3             | 0.51 (1789/3857/2005)           | 0.51 (1061/2211/1117) | 1.03            | 0.98-1.09           | 2.46x10 <sup>-1</sup>        |
| UK4             | 0.52 (133/288/156)              | 0.50 (259/540/252)    | 1.10            | 0.95-1.27           | 2.00x10 <sup>-1</sup>        |
| SCOTLAND3       | 0.52 (162/373/195)              | 0.49 (385/774/361)    | 1.13            | 1.00-1.29           | 5.32x10 <sup>-2</sup>        |
| CAMBRIDGE       | 0.52 (499/1121/584)             | 0.51 (523/1074/560)   | 1.04            | 0.96-1.14           | 3.15x10 <sup>-1</sup>        |
| CROATIA         | 0.53 (80/188/101)               | 0.52 (239/536/276)    | 1.05            | 0.88-1.24           | 6.08x10 <sup>-1</sup>        |
| HELSINKI        | 0.48 (261/460/228)              | 0.48 (235/395/196)    | 1.02            | 0.90-1.17           | 7.16x10 <sup>-1</sup>        |
| SWEDEN          | 0.53 (741/1546/904)             | 0.49 (713/1509/673)   | 1.14            | 1.06-1.22           | 3.41x10 <sup>-4</sup>        |
|                 |                                 |                       | <b>1.08</b>     | <b>1.05-1.10</b>    | <b>1.65x10<sup>-9</sup></b>  |
| JAPAN           | 0.45 (490/761/331)              | 0.43 (630/908/360)    | 1.09            | 0.99-1.19           | 8.46x10 <sup>-2</sup>        |
|                 |                                 |                       |                 |                     |                              |
| <b>Combined</b> |                                 |                       | <b>1.08</b>     | <b>1.05-1.10</b>    | <b>3.65x10<sup>-10</sup></b> |

**rs5934683 (Xp22.2)**

| STUDY           | Cases                           |                      | Controls           |                      | OR <sup>b</sup> | 95% CI <sup>c</sup> | P-value                      |
|-----------------|---------------------------------|----------------------|--------------------|----------------------|-----------------|---------------------|------------------------------|
|                 | Male                            | Female               | Male               | Female               |                 |                     |                              |
|                 | RAF <sup>a</sup> (TT / CT / CC) | RAF (TT / CT / CC)   | RAF (TT / CT / CC) | RAF (TT / CT / CC)   |                 |                     |                              |
| UK1             | 0.37 (153/-/258)                | 0.38 (65/235/180)    | 0.30 (123/-/284)   | 0.31 (50/205/237)    | 1.23            | 1.10-1.38           | 3.32x10 <sup>-4</sup>        |
| UK2             | 0.36 (398/-/708)                | 0.35 (180/723/648)   | 0.35 (393/-/733)   | 0.33 (198/702/770)   | 1.05            | 0.99-1.13           | 1.23x10 <sup>-2</sup>        |
| SCOTLAND1       | 0.36 (177/-/317)                | 0.35 (57/226/196)    | 0.35 (181/-/331)   | 0.34 (55/219/212)    | 1.03            | 0.93-1.15           | 5.75x10 <sup>-1</sup>        |
| SCOTLAND2       | 0.38 (459/-/754)                | 0.35 (106/347/341)   | 0.34 (412/-/808)   | 0.35 (109/382/364)   | 1.07            | 1.00-1.15           | 6.44x10 <sup>-2</sup>        |
| VQ58            | 0.34 (299/-/592)                | 0.35 (64/245/223)    | 0.31 (427/-/958)   | 0.33 (132/586/581)   | 1.07            | 0.99-1.16           | 8.31x10 <sup>-2</sup>        |
|                 |                                 |                      |                    |                      | <b>1.08</b>     | <b>1.04-1.12</b>    | <b>8.19x10<sup>-5</sup></b>  |
| CCFR1           | 0.36 (218/-/392)                | 0.37 (77/267/220)    | 0.33 (158/-/320)   | 0.33 (49/245/227)    | 1.11            | 1.00-1.23           | 4.66x10 <sup>-2</sup>        |
| UK3             | 0.36 (1763/-/3097)              | 0.34 (318/1216/1178) | 0.31 (496/-/1089)  | 0.33 (297/1199/1243) | 1.11            | 1.06-1.16           | 1.87x10 <sup>-5</sup>        |
| UK4             | 0.39 (106/-/168)                | 0.34 (36/132/129)    | 0.35 (158/-/291)   | 0.32 (69/206/269)    | 1.10            | 0.97-1.25           | 1.38x10 <sup>-1</sup>        |
| SCOTLAND3       | 0.33 (138/-/278)                | 0.33 (34/129/133)    | 0.33 (233/-/477)   | 0.35 (87/383/327)    | 0.98            | 0.88-1.09           | 7.01x10 <sup>-1</sup>        |
| CAMBRIDGE       | 0.36 (443/-/804)                | 0.34 (104/423/395)   | 0.34 (304/-/591)   | 0.35 (160/572/535)   | 1.01            | 0.94-1.08           | 8.23x10 <sup>-1</sup>        |
| CROATIA         | 0.39 (68/-/106)                 | 0.36 (18/62/58)      | 0.41 (118/-/172)   | 0.34 (67/203/231)    | 1.04            | 0.89-1.21           | 6.48x10 <sup>-1</sup>        |
| HELSINKI        | 0.43 (182/-/246)                | 0.41 (70/197/144)    | 0.38 (159/-/257)   | 0.40 (86/111/158)    | 1.08            | 0.96-1.20           | 2.02x10 <sup>-1</sup>        |
| SWEDEN          | 0.34 (573/-/1099)               | 0.35 (204/601/632)   | 0.32 (423/-/894)   | 0.33 (144/632/624)   | 1.07            | 1.00-1.13           | 4.67x10 <sup>-2</sup>        |
|                 |                                 |                      |                    |                      | <b>1.07</b>     | <b>1.05-1.10</b>    | <b>7.46x10<sup>-10</sup></b> |
| JAPAN           | 0.88 (850/-/1111)               | 0.87 (470/131/12)    | 0.87 (1041/-/150)  | 0.87 (516/160/6)     | 1.04            | 0.93-1.16           | 5.38x10 <sup>-1</sup>        |
|                 |                                 |                      |                    |                      |                 |                     |                              |
| <b>Combined</b> |                                 |                      |                    |                      | <b>1.07</b>     | <b>1.04-1.10</b>    | <b>7.30x10<sup>-10</sup></b> |
|                 |                                 |                      |                    |                      |                 |                     |                              |

**Supplementary Table 3: Relationship between rs1321311 (6p21), rs3824999 (11q13.4), and rs5934683 (Xp22.2) genotypes and sex, age at diagnosis of CRC, tumor site (rectal [ICD9:154], colonic [ICD9:153]), stage, family history of CRC (presence of 1+ first degree relative) and MSI status. \*OR>1.0 indicative of over-representation of rectal disease.**

|                       | rs1321311        |         |                     | rs3824999        |         |                     | rs5934683        |         |                     |
|-----------------------|------------------|---------|---------------------|------------------|---------|---------------------|------------------|---------|---------------------|
|                       | OR (95% CI)      | p-value | Sample size         | OR (95% CI)      | p-value | Sample size         | OR (95% CI)      | p-value | Sample size         |
| <b>Sex</b>            | 0.99 (0.95-1.03) | 0.61    | 23,280 <sup>a</sup> | 1.04 (1.00-1.08) | 0.03    | 24,313 <sup>a</sup> | -                | -       | -                   |
| <b>Age</b>            | 0.97 (0.92-1.02) | 0.19    | 22,924 <sup>b</sup> | 0.99 (0.95-1.03) | 0.75    | 22,956 <sup>b</sup> | 1.00 (0.99-1.01) | 0.68    | 20,667 <sup>c</sup> |
| <b>Site*</b>          | 1.02 (0.98-1.08) | 0.26    | 19,843 <sup>d</sup> | 0.97 (0.93-1.01) | 0.12    | 19,889 <sup>d</sup> | 1.00 (0.98-1.01) | 0.70    | 17,589 <sup>e</sup> |
| <b>Family History</b> | 0.97 (0.89-1.04) | 0.39    | 11,867 <sup>f</sup> | 0.97 (0.90-1.03) | 0.32    | 11,909 <sup>f</sup> | 1.00 (0.97-1.02) | 0.81    | 11,782 <sup>f</sup> |
| <b>MSI</b>            | 0.96 (0.81-1.14) | 0.67    | 4,453 <sup>g</sup>  | 1.06 (0.92-1.23) | 0.41    | 4,462 <sup>g</sup>  | 0.99 (0.98-1.04) | 0.66    | 3,060 <sup>h</sup>  |
| <b>Stage</b>          | 0.96 (0.88-1.05) | 0.37    | 5,681 <sup>i</sup>  | 1.02 (0.95-1.10) | 0.55    | 5,726 <sup>i</sup>  | 1.00 (0.98-1.04) | 0.66    | 5,679 <sup>i</sup>  |

a: UK1, SCOTLAND 1, SCOTLAND 2, VQ58, CCFR1, UK2, UK3, SCOTLAND 3 , COINNBS, SWEDEN, UK4, HELSINKI

b: UK1, SCOTLAND 1, SCOTLAND 2, VQ58, UK2, UK3, SCOTLAND 3 , COINNBS, SWEDEN, UK4, HELSINKI

c: UK1, SCOTLAND 1, SCOTLAND 2, VQ58, UK2, UK3, SCOTLAND 3 , SWEDEN, UK4, HELSINKI

d: SCOTLAND 2, VQ58, UK2, UK3, SCOTLAND 3 , COINNBS, SWEDEN, HELSINKI

e: SCOTLAND 2, VQ58, UK2, UK3, SCOTLAND 3 , SWEDEN, HELSINKI

f: UK2, UK3, SCOTLAND 3, HELSINKI

g: UK1, UK2, UK3, COINNBS

h: UK1, UK2, UK3

i: UK1, SCOTLAND 2, SCOTLAND 3, SWEDEN, HELSINKI

**Supplementary Table 4: Details of transcription factor binding sites (TFBSs) and regulatory regions as predicted by Transfac Matrix Database (v7), ENCODE CHIP-Seq and ENCODE UW DNaseI Hypersensitivity (HS) data.** "Score" refers to the confidence value assigned to each predicted binding region by the three different programs; regions of DNaseI HS are highlighted. For comparison, the observed and imputed SNPs and associated SNP-Test *P*-values are shown. The genotyped SNP with the most significant association is highlighted in blue; imputed SNPs showing a more significant association are highlighted in red with the SNP showing the most significant association highlighted in purple. Also shown are gene regions with exons highlighted in grey.

### 6p21 (rs1321311)

| Gene region | rsID       | SNP type  | P-value | TF type | Source          | Score | Chrom start | Chrom end |
|-------------|------------|-----------|---------|---------|-----------------|-------|-------------|-----------|
|             | rs10214849 | Genotyped | 0.632   |         |                 |       | 36709423    |           |
|             | rs9296193  | Imputed   | 0.667   |         |                 |       | 36709434    |           |
| DNaseI HS   |            |           |         |         | ENCODE          | 1000  | 36709820    | 36710050  |
|             | rs6937430  | Imputed   | 0.629   |         |                 |       | 36709875    |           |
| DNaseI HS   |            |           |         |         | ENCODE          | 173   | 36710260    | 36710430  |
| DNaseI HS   |            |           |         |         | ENCODE          | 95    | 36710560    | 36710770  |
|             | rs4713989  | Genotyped | 0.645   |         |                 |       | 36712176    |           |
| DNaseI HS   |            |           |         |         | ENCODE          | 153   | 36712200    | 36712510  |
|             |            |           |         | EBF     | ENCODE ChIP-seq | 799   | 36712201    | 36712641  |
|             |            |           |         | CEBPB   | ENCODE ChIP-seq | 388   | 36714395    | 36714711  |
| DNaseI HS   |            |           |         |         | ENCODE          | 143   | 36715220    | 36715510  |
|             |            |           |         | RXRA    | ENCODE ChIP-seq | 178   | 36717292    | 36717668  |
|             |            |           |         | p300    | ENCODE ChIP-seq | 303   | 36717324    | 36717624  |
|             |            |           |         | HNF4A   | ENCODE ChIP-seq | 403   | 36717355    | 36717660  |
|             |            |           |         | BAF155  | ENCODE ChIP-seq | 1000  | 36718400    | 36719560  |
|             |            |           |         | BAF170  | ENCODE ChIP-seq | 861   | 36718451    | 36719233  |
|             |            |           |         | FOSL2   | ENCODE ChIP-seq | 240   | 36718572    | 36719302  |
| DNaseI HS   |            |           |         |         | ENCODE          | 394   | 36718580    | 36718770  |
|             |            |           |         | HEY1    | ENCODE ChIP-seq | 37    | 36718705    | 36719185  |
|             |            |           |         | Ini1    | ENCODE ChIP-seq | 259   | 36718791    | 36719580  |
|             |            |           |         | GR      | ENCODE ChIP-seq | 170   | 36718796    | 36719242  |
|             |            |           |         | JunD    | ENCODE ChIP-seq | 119   | 36718884    | 36719160  |
| DNaseI HS   |            |           |         |         | ENCODE          | 768   | 36718900    | 36719370  |
|             |            |           |         | NRSF    | ENCODE ChIP-seq | 206   | 36718923    | 36719167  |
|             | rs9368944  | Genotyped | 0.567   |         |                 |       | 36719846    |           |
|             |            |           |         | GR      | ENCODE ChIP-seq | 258   | 36720272    | 36720793  |
|             |            |           |         | Ini1    | ENCODE ChIP-seq | 189   | 36720328    | 36720885  |
|             |            |           |         | BAF155  | ENCODE ChIP-seq | 469   | 36720355    | 36720906  |
|             |            |           |         | JunD    | ENCODE ChIP-seq | 312   | 36720381    | 36720626  |
| DNaseI HS   |            |           |         |         | ENCODE          | 1000  | 36720400    | 36720610  |
|             |            |           |         | CEBPB   | ENCODE ChIP-seq | 310   | 36720626    | 36720906  |
| DNaseI HS   |            |           |         |         | ENCODE          | 401   | 36720700    | 36720950  |
|             |            |           |         | PU.1    | ENCODE ChIP-seq | 1000  | 36720908    | 36721324  |
| DNaseI HS   |            |           |         |         | ENCODE          | 110   | 36721060    | 36721250  |
|             |            |           |         | Pbx1a   | TransFac        | 1.89  | 36725309    | 36725318  |
|             |            |           |         | NFKB    | ENCODE ChIP-seq | 126   | 36725374    | 36725592  |
|             |            |           |         | NRSF    | ENCODE ChIP-seq | 174   | 36725389    | 36725761  |
|             |            |           |         | EBF     | ENCODE ChIP-seq | 241   | 36725392    | 36725813  |
| DNaseI HS   |            |           |         |         | ENCODE          | 97    | 36725440    | 36725650  |



|           |                   |                  |                 |          |                 |      |                 |          |
|-----------|-------------------|------------------|-----------------|----------|-----------------|------|-----------------|----------|
|           |                   |                  |                 | BAF155   | ENCODE ChIP-seq | 628  | 36725508        | 36725699 |
|           |                   |                  |                 | MZF-1    | TransFac        | 1.69 | 36725591        | 36725599 |
|           | <b>rs12207916</b> | <b>Imputed</b>   | <b>5.16E-03</b> |          |                 |      | <b>36725630</b> |          |
| DNaseI HS |                   |                  |                 | CEBPB    | ENCODE ChIP-seq | 108  | 36725640        | 36725912 |
|           |                   |                  |                 |          | ENCODE          | 159  | 36725660        | 36725930 |
|           |                   |                  |                 | c-Jun    | ENCODE ChIP-seq | 426  | 36725832        | 36725949 |
|           | rs6457932         | Imputed          | 0.349           |          |                 |      | 36726118        |          |
|           | rs6457933         | Imputed          | 0.108           |          |                 |      | 36726246        |          |
|           |                   |                  |                 | IRF4     | ENCODE ChIP-seq | 497  | 36726514        | 36727028 |
|           |                   |                  |                 | PU.1     | ENCODE ChIP-seq | 1000 | 36726517        | 36727130 |
|           |                   |                  |                 | SP1      | ENCODE ChIP-seq | 208  | 36726586        | 36727062 |
|           |                   |                  |                 | BCL11A   | ENCODE ChIP-seq | 784  | 36726596        | 36727038 |
|           |                   |                  |                 | NFKB     | ENCODE ChIP-seq | 46   | 36726612        | 36727087 |
|           |                   |                  |                 | TCF12    | ENCODE ChIP-seq | 427  | 36726630        | 36726960 |
| DNaseI HS |                   |                  |                 |          | ENCODE          | 753  | 36726660        | 36727130 |
|           |                   |                  |                 | PAX5-N19 | ENCODE ChIP-seq | 241  | 36726670        | 36727012 |
|           | rs1321313         | Genotyped        | 0.013           |          |                 |      | 36726799        |          |
|           | rs9349003         | Imputed          | 0.340           |          |                 |      | 36727368        |          |
|           | <b>rs7742159</b>  | <b>Imputed</b>   | <b>5.39E-03</b> |          |                 |      | <b>36727430</b> |          |
|           |                   |                  |                 | EBF      | ENCODE ChIP-seq | 118  | 36727550        | 36727878 |
| DNaseI HS |                   |                  |                 | CEBPB    | ENCODE ChIP-seq | 166  | 36727704        | 36727910 |
|           |                   |                  |                 |          | ENCODE          | 102  | 36727720        | 36727890 |
|           | rs9366912         | Imputed          | 0.407           |          |                 |      | 36727861        |          |
|           | rs12524435        | Imputed          | 0.339           |          |                 |      | 36728126        |          |
|           |                   |                  |                 | CEBPB    | ENCODE ChIP-seq | 341  | 36728145        | 36728376 |
|           |                   |                  |                 | RXRA     | ENCODE ChIP-seq | 114  | 36728199        | 36728477 |
|           | rs10947616        | Imputed          | 0.338           |          |                 |      | 36728477        |          |
|           | <b>rs9470358</b>  | <b>Imputed</b>   | <b>4.60E-03</b> |          |                 |      | <b>36728480</b> |          |
|           | rs4713993         | Imputed          | 0.338           |          |                 |      | 36728720        |          |
|           | rs7382183         | Imputed          | 0.338           |          |                 |      | 36728853        |          |
|           | rs6915170         | Imputed          | 0.338           |          |                 |      | 36729017        |          |
|           | rs4568449         | Imputed          | 0.108           |          |                 |      | 36729121        |          |
|           | rs4464789         | Genotyped        | 0.329           |          |                 |      | 36729156        |          |
|           | rs10456441        | Imputed          | 0.338           |          |                 |      | 36729337        |          |
|           | rs10456442        | Imputed          | 0.338           |          |                 |      | 36729389        |          |
|           | rs10456443        | Imputed          | 0.338           |          |                 |      | 36729425        |          |
|           | rs4713994         | Imputed          | 0.409           |          |                 |      | 36729511        |          |
|           | rs12210094        | Imputed          | 0.337           |          |                 |      | 36730291        |          |
|           | rs12528913        | Imputed          | 0.326           |          |                 |      | 36730527        |          |
|           | <b>rs9918353</b>  | <b>Imputed</b>   | <b>5.41E-03</b> |          |                 |      | <b>36730655</b> |          |
| DNaseI HS |                   |                  |                 |          | ENCODE          | 417  | 36730800        | 36731230 |
|           | rs1321312         | Imputed          | 0.325           |          |                 |      | 36730852        |          |
|           | <b>rs1321311</b>  | <b>Genotyped</b> | <b>6.25E-03</b> |          |                 |      | <b>36730878</b> |          |
|           | rs12530170        | Genotyped        | 0.824           |          |                 |      | 36731061        |          |
|           |                   |                  |                 | NRSF     | ENCODE ChIP-seq | 95   | 36731099        | 36731443 |
|           | rs1321310         | Imputed          | 7.42E-03        |          |                 |      | 36731102        |          |
|           | rs4331968         | Imputed          | 8.08E-03        |          |                 |      | 36731221        |          |
| DNaseI HS |                   |                  |                 |          | ENCODE          | 357  | 36731260        | 36731450 |
|           | rs9470361         | Imputed          | 7.73E-03        |          |                 |      | 36731357        |          |
|           | rs7774130         | Imputed          | 8.06E-03        |          |                 |      | 36731734        |          |
|           |                   |                  |                 | EBF      | ENCODE ChIP-seq | 110  | 36731903        | 36732220 |
|           | rs7740181         | Imputed          | 0.117           |          |                 |      | 36732089        |          |
|           |                   |                  |                 | c-Jun    | ENCODE ChIP-seq | 514  | 36732176        | 36732506 |
|           |                   |                  |                 | c-Fos    | ENCODE ChIP-seq | 305  | 36732201        | 36732497 |

|           |            |            |           |          |                 |                 |          |          |          |
|-----------|------------|------------|-----------|----------|-----------------|-----------------|----------|----------|----------|
| DNaseI HS |            |            |           |          | ENCODE          | 1000            | 36732300 | 36732490 |          |
|           |            |            |           | USF-1    | ENCODE ChIP-seq | 817             | 36732353 | 36732453 |          |
|           | rs12203953 | Imputed    | 0.350     |          |                 |                 | 36732400 |          |          |
|           | rs9470362  | Imputed    | 0.123     |          |                 |                 | 36732627 |          |          |
|           | rs9470363  | Imputed    | 0.124     |          |                 |                 | 36732691 |          |          |
|           | rs6907437  | Imputed    | 0.128     |          |                 |                 | 36732999 |          |          |
| DNaseI HS |            |            |           |          | ENCODE          | 182             | 36733120 | 36733310 |          |
|           |            | rs6907793  | Imputed   | 0.131    |                 |                 | 36733184 |          |          |
|           |            | rs6930671  | Imputed   | 0.132    |                 |                 | 36733250 |          |          |
|           |            | rs11969445 | Imputed   | 0.130    |                 |                 | 36733360 |          |          |
|           | rs9470366  | Imputed    | 0.031     |          |                 | 36733540        |          |          |          |
| DNaseI HS |            |            |           |          | ENCODE          | 1000            | 36733840 | 36734250 |          |
|           |            |            |           | PAX5-N19 | ENCODE ChIP-seq | 121             | 36733879 | 36734419 |          |
|           |            |            |           | BATF     | ENCODE ChIP-seq | 163             | 36733913 | 36734308 |          |
|           |            |            |           | EBF      | ENCODE ChIP-seq | 493             | 36733933 | 36734256 |          |
|           |            |            |           | NFKB     | ENCODE ChIP-seq | 120             | 36733980 | 36734419 |          |
|           |            |            |           | PAX5-C20 | ENCODE ChIP-seq | 251             | 36734000 | 36734333 |          |
|           |            | rs6936598  | Imputed   | 0.130    |                 |                 | 36734074 |          |          |
|           |            |            |           | Arnt     | TransFac        | 2.76            | 36734095 | 36734115 |          |
|           |            |            |           | c-Myc    | TransFac        | 2.13            | 36734095 | 36734115 |          |
|           |            |            |           | SREBP-1a | TransFac        | 2.67            | 36734099 | 36734110 |          |
|           |            | rs6936993  | Imputed   | 0.132    |                 |                 | 36734300 |          |          |
|           | rs9470367  | Imputed    | 0.326     |          |                 | 36734910        |          |          |          |
|           | rs7756236  | Imputed    | 7.85E-03  |          |                 | 36735031        |          |          |          |
| DNaseI HS |            |            |           |          | ENCODE          | 1000            | 36735140 | 36735390 |          |
|           |            |            |           | BCL3     | ENCODE ChIP-seq | 119             | 36735264 | 36735564 |          |
|           |            |            |           | NFKB     | ENCODE ChIP-seq | 1000            | 36735422 | 36735425 |          |
|           |            | rs6920453  | Imputed   | 0.319    |                 |                 | 36735461 |          |          |
|           |            | rs9462207  | Imputed   | 7.85E-03 |                 |                 | 36735577 |          |          |
|           |            | rs9368950  | Genotyped | 0.347    |                 |                 | 36735850 |          |          |
|           |            | rs9462208  | Imputed   | 0.133    |                 |                 | 36735868 |          |          |
|           |            | rs9462209  | Genotyped | 0.141    |                 |                 | 36736020 |          |          |
|           |            |            |           |          | HEY1            | ENCODE ChIP-seq | 42       | 36736470 | 36736845 |
|           |            |            |           |          | STAT2           | ENCODE ChIP-seq | 161      | 36736508 | 36736967 |
|           |            |            |           |          | c-Myc           | ENCODE ChIP-seq | 354      | 36736511 | 36737120 |
|           |            |            |           |          | c-Jun           | ENCODE ChIP-seq | 107      | 36736539 | 36736941 |
|           |            |            |           |          | STAT1           | ENCODE ChIP-seq | 274      | 36736555 | 36737050 |
|           | DNaseI HS  |            |           |          |                 | ENCODE          | 387      | 36736600 | 36736890 |
|           |            |            |           |          | Max             | ENCODE ChIP-seq | 498      | 36736621 | 36737110 |
|           |            |            |           | BCL3     | ENCODE ChIP-seq | 1000            | 36736634 | 36737717 |          |
|           |            |            |           | PAX5-N19 | ENCODE ChIP-seq | 113             | 36736644 | 36737199 |          |
|           |            | rs6926780  | Imputed   | 0.386    |                 |                 | 36736650 |          |          |
|           |            |            |           |          | PAX5-C20        | ENCODE ChIP-seq | 164      | 36736721 | 36737085 |
|           |            |            |           |          | NFKB            | ENCODE ChIP-seq | 91       | 36736774 | 36737107 |
|           |            |            |           |          | Pbx3            | ENCODE ChIP-seq | 200      | 36736781 | 36737169 |
|           |            | rs9462210  | Imputed   | 7.82E-03 |                 |                 | 36736931 |          |          |
|           |            |            |           |          | Pbx3            | ENCODE ChIP-seq | 98       | 36737344 | 36737584 |
|           |            | rs10807170 | Genotyped | 0.031    |                 |                 | 36737422 |          |          |
|           | rs4713996  | Imputed    | 0.067     |          |                 | 36737692        |          |          |          |
|           | rs4713997  | Imputed    | 0.384     |          |                 | 36737811        |          |          |          |
| DNaseI HS |            |            |           |          | ENCODE          | 340             | 36738100 | 36738530 |          |
|           |            |            |           | NFKB     | ENCODE ChIP-seq | 119             | 36738149 | 36739005 |          |
|           |            |            |           | BATF     | ENCODE ChIP-seq | 63              | 36738245 | 36738550 |          |
|           |            |            |           | BAF155   | ENCODE ChIP-seq | 427             | 36738311 | 36739440 |          |

|           |            |           |       |          |                 |      |          |          |
|-----------|------------|-----------|-------|----------|-----------------|------|----------|----------|
|           |            |           |       | Ini1     | ENCODE ChIP-seq | 227  | 36738314 | 36739025 |
|           | rs9380585  | Imputed   | 0.383 |          |                 |      | 36738346 |          |
|           | rs9394368  | Imputed   | 0.066 |          |                 |      | 36738503 |          |
|           |            |           |       | BAF170   | ENCODE ChIP-seq | 868  | 36738562 | 36738905 |
| DNaseI HS |            |           |       |          | ENCODE          | 244  | 36738580 | 36738750 |
|           |            |           |       | CEBPB    | ENCODE ChIP-seq | 899  | 36738716 | 36739111 |
|           |            |           |       | HNF4A    | ENCODE ChIP-seq | 211  | 36738720 | 36739058 |
| DNaseI HS |            |           |       |          | ENCODE          | 336  | 36738780 | 36739050 |
|           |            |           |       | AREB6    | TransFac        | 2.6  | 36738824 | 36738837 |
|           |            |           |       | TBP      | TransFac        | 1.65 | 36739239 | 36739254 |
|           |            |           |       | NF-E2    | ENCODE ChIP-seq | 281  | 36739301 | 36739527 |
| DNaseI HS |            |           |       |          | ENCODE          | 83   | 36739320 | 36739510 |
|           | rs9380586  | Imputed   | 0.372 |          |                 |      | 36739542 |          |
|           |            |           |       | PU.1     | ENCODE ChIP-seq | 126  | 36739583 | 36739874 |
|           |            |           |       | IRF4     | ENCODE ChIP-seq | 134  | 36739602 | 36740231 |
| DNaseI HS |            |           |       |          | ENCODE          | 233  | 36739640 | 36739930 |
|           |            |           |       | NFKB     | ENCODE ChIP-seq | 110  | 36739800 | 36740167 |
|           |            |           |       | JunD     | ENCODE ChIP-seq | 352  | 36739844 | 36740258 |
|           |            |           |       | BATF     | ENCODE ChIP-seq | 325  | 36739856 | 36740214 |
| DNaseI HS |            |           |       |          | ENCODE          | 138  | 36739940 | 36740110 |
|           |            |           |       | POU2F2   | ENCODE ChIP-seq | 796  | 36740309 | 36740962 |
|           |            |           |       | HEY1     | ENCODE ChIP-seq | 160  | 36740336 | 36740644 |
|           |            |           |       | EBF      | ENCODE ChIP-seq | 285  | 36740385 | 36740865 |
| DNaseI HS | rs4711456  | Imputed   | 0.080 |          |                 |      | 36740460 |          |
|           |            |           |       |          | ENCODE          | 297  | 36740460 | 36740830 |
|           |            |           |       | NFKB     | ENCODE ChIP-seq | 164  | 36740478 | 36740942 |
|           |            |           |       | TCF12    | ENCODE ChIP-seq | 113  | 36740535 | 36740811 |
|           | rs13196885 | Imputed   | 0.365 |          |                 |      | 36740666 |          |
|           | rs4713999  | Genotyped | 0.072 |          |                 |      | 36741047 |          |
|           | rs4711457  | Imputed   | 0.065 |          |                 |      | 36741138 |          |
| DNaseI HS |            |           |       |          | ENCODE          | 132  | 36741240 | 36741390 |
|           | rs1969701  | Imputed   | 0.379 |          |                 |      | 36741676 |          |
| DNaseI HS |            |           |       |          | ENCODE          | 99   | 36742080 | 36742270 |
|           | rs6930083  | Imputed   | 0.063 |          |                 |      | 36742134 |          |
|           | rs6931097  | Imputed   | 0.166 |          |                 |      | 36742437 |          |
|           |            |           |       | NFKB     | ENCODE ChIP-seq | 193  | 36742534 | 36743469 |
|           |            |           |       | HNF4A    | ENCODE ChIP-seq | 172  | 36742679 | 36743175 |
|           |            |           |       | BAF155   | ENCODE ChIP-seq | 666  | 36742715 | 36743228 |
|           |            |           |       | FOSL2    | ENCODE ChIP-seq | 125  | 36742720 | 36743263 |
|           |            |           |       | TAF1     | ENCODE ChIP-seq | 447  | 36742738 | 36743359 |
|           |            |           |       | POU2F2   | ENCODE ChIP-seq | 728  | 36742754 | 36743292 |
|           |            |           |       | SP1      | ENCODE ChIP-seq | 682  | 36742760 | 36743311 |
|           |            |           |       | BCL11A   | ENCODE ChIP-seq | 699  | 36742763 | 36743257 |
|           |            |           |       | EBF      | ENCODE ChIP-seq | 1000 | 36742768 | 36743364 |
|           |            |           |       | PAX5-N19 | ENCODE ChIP-seq | 1000 | 36742780 | 36743370 |
|           |            |           |       | HSF1     | ENCODE ChIP-seq | 241  | 36742785 | 36743114 |
|           |            |           |       | CEBPB    | ENCODE ChIP-seq | 1000 | 36742789 | 36743242 |
|           |            |           |       | p300     | ENCODE ChIP-seq | 704  | 36742808 | 36743203 |
|           |            |           |       | IRF4     | ENCODE ChIP-seq | 555  | 36742808 | 36743390 |
|           |            |           |       | JunD     | ENCODE ChIP-seq | 492  | 36742809 | 36743180 |
|           |            |           |       | BATF     | ENCODE ChIP-seq | 732  | 36742809 | 36743196 |
|           |            |           |       | TCF12    | ENCODE ChIP-seq | 509  | 36742813 | 36743187 |
|           |            |           |       | c-Myc    | ENCODE ChIP-seq | 351  | 36742816 | 36743217 |
|           |            |           |       | PAX5-C20 | ENCODE ChIP-seq | 1000 | 36742816 | 36743384 |

|           |            |           |          |        |                 |      |          |          |
|-----------|------------|-----------|----------|--------|-----------------|------|----------|----------|
|           |            |           |          | c-Jun  | ENCODE ChIP-seq | 173  | 36742841 | 36743140 |
|           |            |           |          | Ini1   | ENCODE ChIP-seq | 232  | 36742843 | 36743217 |
|           |            |           |          | HEY1   | ENCODE ChIP-seq | 161  | 36742853 | 36743331 |
|           |            |           |          | BAF170 | ENCODE ChIP-seq | 647  | 36742862 | 36743206 |
|           |            |           |          | BCL3   | ENCODE ChIP-seq | 119  | 36742877 | 36743190 |
| DNaseI HS |            |           |          |        | ENCODE          | 1000 | 36742880 | 36743150 |
|           |            |           |          | PU.1   | ENCODE ChIP-seq | 822  | 36742890 | 36743338 |
|           |            |           |          | NRSF   | ENCODE ChIP-seq | 168  | 36742933 | 36743246 |
|           |            |           |          | Egr-2  | TransFac        | 2.35 | 36742947 | 36742959 |
|           |            |           |          | p53    | TransFac        | 4.55 | 36743012 | 36743032 |
|           |            |           |          | POU3F2 | TransFac        | 2.06 | 36743479 | 36743489 |
|           |            |           |          | Rad21  | ENCODE ChIP-seq | 139  | 36743573 | 36743630 |
|           | rs66761782 | Imputed   | 6.94E-03 |        |                 |      | 36744058 |          |
|           |            |           |          | AP-1   | TransFac        | 2.21 | 36744277 | 36744290 |
|           |            |           |          | Max    | ENCODE ChIP-seq | 639  | 36744338 | 36744565 |
|           |            |           |          | EBF    | ENCODE ChIP-seq | 53   | 36744369 | 36744656 |
| DNaseI HS |            |           |          |        | ENCODE          | 112  | 36744380 | 36744570 |
|           | rs6457934  | Imputed   | 0.178    |        |                 |      | 36745303 |          |
| DNaseI HS |            |           |          |        | ENCODE          | 140  | 36745480 | 36745650 |
|           | rs4714000  | Genotyped | 0.636    |        |                 |      | 36745575 |          |
| DNaseI HS |            |           |          |        | ENCODE          | 384  | 36746040 | 36746310 |
|           | rs4714001  | Imputed   | 0.035    |        |                 |      | 36746153 |          |
|           | rs1321309  | Imputed   | 0.209    |        |                 |      | 36746614 |          |
|           | rs1321308  | Imputed   | 0.222    |        |                 |      | 36746669 |          |
|           |            |           |          | HNF4A  | ENCODE ChIP-seq | 209  | 36747420 | 36747819 |
|           |            |           |          | USF-1  | ENCODE ChIP-seq | 55   | 36747495 | 36747701 |
|           | rs58714705 | Imputed   | 0.213    |        |                 |      | 36747591 |          |
| DNaseI HS |            |           |          |        | ENCODE          | 148  | 36747820 | 36748070 |
|           | rs11758192 | Imputed   | 0.213    |        |                 |      | 36747946 |          |
|           |            |           |          | STAT1  | ENCODE ChIP-seq | 50   | 36748055 | 36748275 |
|           | rs10947618 | Imputed   | 0.213    |        |                 |      | 36748110 |          |
|           | rs10947619 | Imputed   | 0.210    |        |                 |      | 36748135 |          |
|           | rs10947620 | Imputed   | 0.213    |        |                 |      | 36748166 |          |
|           | rs7452626  | Imputed   | 0.602    |        |                 |      | 36748219 |          |
|           | rs10947621 | Imputed   | 0.209    |        |                 |      | 36748250 |          |
|           | rs9349005  | Imputed   | 0.226    |        |                 |      | 36748346 |          |
|           | rs9380587  | Imputed   | 0.623    |        |                 |      | 36748461 |          |
|           | rs12174270 | Imputed   | 0.213    |        |                 |      | 36748647 |          |
|           | rs7747146  | Imputed   | 0.181    |        |                 |      | 36748968 |          |
|           | rs7764973  | Imputed   | 0.213    |        |                 |      | 36749014 |          |
|           | rs6457935  | Imputed   | 0.213    |        |                 |      | 36749116 |          |
|           | rs12199346 | Imputed   | 6.11E-03 |        |                 |      | 36749524 |          |
|           |            |           |          | LCR-F1 | TransFac        | 1.71 | 36749563 | 36749576 |
|           |            |           |          | XBP-1  | TransFac        | 2.35 | 36749614 | 36749631 |
| DNaseI HS |            |           |          |        | ENCODE          | 69   | 36749680 | 36749910 |
|           |            |           |          | SRF    | TransFac        | 1.99 | 36749730 | 36749745 |
|           | rs1977172  | Imputed   | 0.622    |        |                 |      | 36749732 |          |
|           | rs1010424  | Imputed   | 0.213    |        |                 |      | 36749763 |          |
|           | rs1010423  | Imputed   | 0.213    |        |                 |      | 36749769 |          |
|           |            |           |          | CUTL1  | TransFac        | 1.94 | 36749900 | 36749910 |
|           | rs4711458  | Imputed   | 0.213    |        |                 |      | 36749919 |          |
|           | rs4711459  | Imputed   | 0.213    |        |                 |      | 36750002 |          |
|           | rs4711460  | Imputed   | 0.213    |        |                 |      | 36750106 |          |
|           | rs4711461  | Imputed   | 0.230    |        |                 |      | 36750146 |          |

|                 |             |           |       |        |                 |      |          |          |          |
|-----------------|-------------|-----------|-------|--------|-----------------|------|----------|----------|----------|
|                 | rs4714002   | Imputed   | 0.213 |        |                 |      |          | 36750164 |          |
|                 | rs4711462   | Imputed   | 0.213 |        |                 |      |          | 36750168 |          |
|                 | rs4714003   | Imputed   | 0.213 |        |                 |      |          | 36750238 |          |
|                 |             |           |       | POU2F1 | TransFac        | 2.41 | 36750348 |          | 36750367 |
|                 |             |           |       | SRF    | TransFac        | 1.88 | 36750354 |          | 36750368 |
|                 | rs10947622  | Imputed   | 0.213 |        |                 |      | 36750380 |          |          |
|                 |             |           |       | p53    | TransFac        | 2.34 | 36750494 |          | 36750514 |
|                 | rs12214686  | Imputed   | 0.206 |        |                 |      | 36750529 |          |          |
|                 |             |           |       | Arnt   | TransFac        | 1.96 | 36750663 |          | 36750683 |
|                 |             |           |       | c-Myc  | TransFac        | 1.77 | 36750663 |          | 36750683 |
| DNaseI HS       |             |           |       |        | ENCODE          | 588  | 36750800 |          | 36750990 |
|                 |             |           |       | c-Jun  | ENCODE ChIP-seq | 428  | 36750808 |          | 36750975 |
|                 | rs10947623  | Imputed   | 0.206 |        |                 |      | 36750814 |          |          |
|                 |             |           |       | POU3F2 | TransFac        | 1.7  | 36750883 |          | 36750899 |
|                 |             |           |       | Lmo2   | TransFac        | 2.23 | 36750942 |          | 36750951 |
|                 | rs12192827  | Imputed   | 0.213 |        |                 |      | 36750949 |          |          |
|                 | rs12192877  | Imputed   | 0.213 |        |                 |      | 36750977 |          |          |
|                 | rs72853370  | Imputed   | 0.213 |        |                 |      | 36751200 |          |          |
|                 | rs11962192  | Imputed   | 0.213 |        |                 |      | 36751215 |          |          |
|                 | rs113041051 | Imputed   | 0.213 |        |                 |      | 36751224 |          |          |
|                 | rs9357222   | Imputed   | 0.213 |        |                 |      | 36751338 |          |          |
|                 | rs9368953   | Imputed   | 0.213 |        |                 |      | 36751444 |          |          |
|                 |             |           |       | STAT1  | ENCODE ChIP-seq | 155  | 36751645 |          | 36752569 |
|                 |             |           |       | NFKB   | ENCODE ChIP-seq | 148  | 36751723 |          | 36752524 |
|                 | rs9394371   | Imputed   | 0.213 |        |                 |      | 36751733 |          |          |
|                 |             |           |       | PU.1   | ENCODE ChIP-seq | 823  | 36751757 |          | 36752165 |
|                 |             |           |       | HEY1   | ENCODE ChIP-seq | 616  | 36751779 |          | 36752604 |
|                 |             |           |       | TAF1   | ENCODE ChIP-seq | 304  | 36751788 |          | 36752505 |
|                 |             |           |       | POU2F2 | ENCODE ChIP-seq | 181  | 36751815 |          | 36752543 |
| DNaseI HS       |             |           |       |        | ENCODE          | 308  | 36751840 |          | 36751990 |
|                 |             |           |       | SRF    | TransFac        | 2.46 | 36751897 |          | 36751911 |
|                 |             |           |       | p300   | ENCODE ChIP-seq | 106  | 36751941 |          | 36752370 |
|                 |             |           |       | CEBPB  | ENCODE ChIP-seq | 83   | 36752026 |          | 36752491 |
| DNaseI HS       |             |           |       |        | ENCODE          | 537  | 36752060 |          | 36752510 |
|                 |             |           |       | p53    | TransFac        | 3.3  | 36752203 |          | 36752223 |
| CDKN1A Exon 1   |             |           |       |        |                 |      | 36752245 |          | 36752279 |
| CDKN1A Intron 1 |             |           |       |        |                 |      | 36752280 |          | 36753656 |
|                 | rs3829963   | Genotyped | 0.252 |        |                 |      | 36752364 |          |          |
| DNaseI HS       |             |           |       |        | ENCODE          | 110  | 36752520 |          | 36752790 |
|                 |             |           |       | FOXC1  | TransFac        | 2.98 | 36752741 |          | 36752757 |
|                 |             |           |       | FOXD1  | TransFac        | 3.72 | 36752741 |          | 36752757 |
|                 |             |           |       | FOXF2  | TransFac        | 2.77 | 36752741 |          | 36752757 |
|                 |             |           |       | FOXI1  | TransFac        | 1.73 | 36752743 |          | 36752756 |
| DNaseI HS       |             |           |       |        | ENCODE          | 432  | 36752860 |          | 36753210 |
|                 |             |           |       | NFKB   | ENCODE ChIP-seq | 233  | 36752897 |          | 36755402 |
|                 |             |           |       | p53    | TransFac        | 3.36 | 36753085 |          | 36753105 |
|                 |             |           |       | SRF    | TransFac        | 1.73 | 36753093 |          | 36753111 |
|                 |             |           |       | p53    | TransFac        | 2.58 | 36753095 |          | 36753105 |
|                 |             |           |       | STAT3  | TransFac        | 1.85 | 36753095 |          | 36753116 |
|                 |             |           |       | HEY1   | ENCODE ChIP-seq | 1000 | 36753132 |          | 36755082 |
|                 |             |           |       | SRF    | ENCODE ChIP-seq | 1000 | 36753401 |          | 36753987 |
|                 |             |           |       | p300   | ENCODE ChIP-seq | 247  | 36753453 |          | 36753966 |
| DNaseI HS       |             |           |       |        | ENCODE          | 376  | 36753460 |          | 36753830 |
|                 |             |           |       | CEBPB  | ENCODE ChIP-seq | 86   | 36753482 |          | 36753903 |

|                 |           |                 |      |          |          |
|-----------------|-----------|-----------------|------|----------|----------|
|                 | JunD      | ENCODE ChIP-seq | 113  | 36753483 | 36753923 |
|                 | HNF4A     | ENCODE ChIP-seq | 195  | 36753496 | 36754726 |
|                 | Sin3Ak-20 | ENCODE ChIP-seq | 302  | 36753532 | 36753841 |
|                 | SREBP1    | ENCODE ChIP-seq | 12   | 36753532 | 36754042 |
|                 | USF-1     | ENCODE ChIP-seq | 164  | 36753628 | 36753872 |
| CDKN1A Exon 2   |           |                 |      | 36753657 | 36753688 |
| CDKN1A Intron 2 |           |                 |      | 36753689 | 36759851 |
|                 | BAF170    | ENCODE ChIP-seq | 310  | 36753819 | 36755474 |
|                 | STAT1     | ENCODE ChIP-seq | 414  | 36753877 | 36755452 |
|                 | PAX5-N19  | ENCODE ChIP-seq | 216  | 36754015 | 36755157 |
|                 | EBF       | ENCODE ChIP-seq | 453  | 36754026 | 36754594 |
|                 | Ini1      | ENCODE ChIP-seq | 282  | 36754031 | 36756991 |
|                 | GR        | ENCODE ChIP-seq | 1000 | 36754051 | 36756479 |
|                 | Nrf1      | ENCODE ChIP-seq | 237  | 36754070 | 36754496 |
|                 | Max       | ENCODE ChIP-seq | 415  | 36754074 | 36756527 |
|                 | BAF155    | ENCODE ChIP-seq | 414  | 36754074 | 36756900 |
|                 | SREBP1    | ENCODE ChIP-seq | 331  | 36754076 | 36755259 |
|                 | FOSL2     | ENCODE ChIP-seq | 300  | 36754077 | 36754697 |
|                 | JunD      | ENCODE ChIP-seq | 713  | 36754088 | 36754658 |
|                 | SP1       | ENCODE ChIP-seq | 162  | 36754089 | 36754806 |
|                 | c-Myc     | ENCODE ChIP-seq | 434  | 36754093 | 36755452 |
|                 | SREBP2    | ENCODE ChIP-seq | 45   | 36754109 | 36754121 |
|                 | PU.1      | ENCODE ChIP-seq | 303  | 36754112 | 36754350 |
|                 | GABP      | ENCODE ChIP-seq | 45   | 36754114 | 36754341 |
|                 | p300      | ENCODE ChIP-seq | 372  | 36754115 | 36754767 |
|                 | PAX5-C20  | ENCODE ChIP-seq | 553  | 36754116 | 36754746 |
|                 | TAF1      | ENCODE ChIP-seq | 864  | 36754125 | 36755421 |
|                 | TCF12     | ENCODE ChIP-seq | 1000 | 36754131 | 36754984 |
|                 | POU2F2    | ENCODE ChIP-seq | 1000 | 36754138 | 36756424 |
|                 | Sin3Ak-20 | ENCODE ChIP-seq | 1000 | 36754146 | 36754621 |
|                 | NRSF      | ENCODE ChIP-seq | 183  | 36754146 | 36754743 |
| DNaseI HS       |           | ENCODE          | 1000 | 36754160 | 36754590 |
|                 | CEBPB     | ENCODE ChIP-seq | 88   | 36754168 | 36754884 |
|                 | ERRA      | ENCODE ChIP-seq | 372  | 36754185 | 36754832 |
|                 | FOXP2     | ENCODE ChIP-seq | 380  | 36754241 | 36754341 |
|                 | Pbx3      | ENCODE ChIP-seq | 487  | 36754282 | 36754684 |
|                 | c-Jun     | ENCODE ChIP-seq | 185  | 36754286 | 36755391 |
|                 | BCL3      | ENCODE ChIP-seq | 124  | 36754308 | 36754654 |
|                 | SREBP2    | ENCODE ChIP-seq | 38   | 36754348 | 36754534 |
|                 | BCL11A    | ENCODE ChIP-seq | 101  | 36754368 | 36754774 |
|                 | PU.1      | ENCODE ChIP-seq | 97   | 36754369 | 36754576 |
|                 | E47       | TransFac        | 1.93 | 36754671 | 36754687 |
|                 | AREB6     | TransFac        | 1.88 | 36754673 | 36754685 |
|                 | MyoD      | TransFac        | 1.72 | 36754689 | 36754699 |
|                 | Sin3Ak-20 | ENCODE ChIP-seq | 134  | 36754751 | 36755010 |
| DNaseI HS       |           | ENCODE          | 430  | 36754780 | 36755610 |
|                 | FOSL2     | ENCODE ChIP-seq | 335  | 36754819 | 36755646 |
|                 | Input     | ENCODE ChIP-seq | 362  | 36754858 | 36755348 |
|                 | BCL11A    | ENCODE ChIP-seq | 210  | 36754992 | 36755313 |
|                 | JunD      | ENCODE ChIP-seq | 249  | 36755001 | 36755385 |
|                 | Sin3Ak-20 | ENCODE ChIP-seq | 237  | 36755016 | 36755312 |
|                 | EBF       | ENCODE ChIP-seq | 135  | 36755035 | 36755321 |
|                 | ATF       | TransFac        | 2.01 | 36755114 | 36755128 |
|                 | Bach1     | TransFac        | 2.57 | 36755186 | 36755201 |

|           |          |                 |      |          |          |
|-----------|----------|-----------------|------|----------|----------|
|           | AP-1     | TransFac        | 2.47 | 36755187 | 36755200 |
|           | E2F      | TransFac        | 2.41 | 36755330 | 36755342 |
|           | HEY1     | ENCODE ChIP-seq | 326  | 36755648 | 36756385 |
|           | EBF      | ENCODE ChIP-seq | 215  | 36755651 | 36756499 |
|           | TAF1     | ENCODE ChIP-seq | 325  | 36755663 | 36756186 |
| DNaseI HS |          | ENCODE          | 260  | 36755680 | 36755870 |
|           | STAT1    | ENCODE ChIP-seq | 162  | 36755702 | 36756840 |
|           | NFKB     | ENCODE ChIP-seq | 175  | 36755709 | 36757018 |
|           | FOSL2    | ENCODE ChIP-seq | 602  | 36755738 | 36756475 |
|           | BAF170   | ENCODE ChIP-seq | 411  | 36755741 | 36755958 |
|           | c-Myc    | ENCODE ChIP-seq | 641  | 36755769 | 36756511 |
|           | STAT2    | ENCODE ChIP-seq | 96   | 36755798 | 36756529 |
|           | c-Jun    | ENCODE ChIP-seq | 334  | 36755806 | 36756545 |
|           | c-Fos    | ENCODE ChIP-seq | 424  | 36755821 | 36756491 |
|           | JunD     | ENCODE ChIP-seq | 269  | 36755826 | 36756530 |
|           | PAX5-C20 | ENCODE ChIP-seq | 926  | 36755836 | 36756546 |
|           | Brg1     | ENCODE ChIP-seq | 555  | 36755838 | 36756729 |
|           | GABP     | ENCODE ChIP-seq | 39   | 36755840 | 36756203 |
| DNaseI HS |          | ENCODE          | 1000 | 36755880 | 36756370 |
|           | IRF4     | ENCODE ChIP-seq | 611  | 36755912 | 36756604 |
|           | RSRFC4   | TransFac        | 1.96 | 36755955 | 36755971 |
|           | CEBPB    | ENCODE ChIP-seq | 353  | 36755963 | 36756421 |
|           | Bach1    | TransFac        | 2.6  | 36755976 | 36755991 |
|           | BCL3     | ENCODE ChIP-seq | 501  | 36755976 | 36756551 |
|           | AP-1     | TransFac        | 2.54 | 36755977 | 36755990 |
|           | Arnt     | TransFac        | 1.87 | 36755981 | 36756001 |
|           | BATF     | ENCODE ChIP-seq | 636  | 36755984 | 36756497 |
|           | NRSF     | ENCODE ChIP-seq | 45   | 36755999 | 36756484 |
|           | PAX5-N19 | ENCODE ChIP-seq | 456  | 36756018 | 36756516 |
|           | TCF12    | ENCODE ChIP-seq | 661  | 36756022 | 36756393 |
|           | SP1      | ENCODE ChIP-seq | 165  | 36756065 | 36756506 |
|           | BCL11A   | ENCODE ChIP-seq | 442  | 36756086 | 36756505 |
|           | p300     | ENCODE ChIP-seq | 168  | 36756094 | 36756573 |
|           | LUN-1    | TransFac        | 2.34 | 36756132 | 36756149 |
|           | Rad21    | ENCODE ChIP-seq | 200  | 36756184 | 36756781 |
|           | CTCF     | ENCODE ChIP-seq | 680  | 36756191 | 36756542 |
|           | CREB     | TransFac        | 2.19 | 36756229 | 36756244 |
|           | HNF4A    | ENCODE ChIP-seq | 406  | 36756233 | 36756422 |
|           | E2F      | TransFac        | 2.21 | 36756256 | 36756264 |
|           | FOXP2    | ENCODE ChIP-seq | 543  | 36756271 | 36756371 |
|           | Bach1    | TransFac        | 2.14 | 36756283 | 36756298 |
|           | AP-1     | TransFac        | 1.86 | 36756284 | 36756297 |
|           | BAF170   | ENCODE ChIP-seq | 774  | 36756285 | 36756485 |
|           | Egr-1    | ENCODE ChIP-seq | 638  | 36756290 | 36756390 |
|           | Egr-2    | TransFac        | 1.95 | 36756322 | 36756334 |
|           | Egr-3    | TransFac        | 2.01 | 36756322 | 36756334 |
| DNaseI HS |          | ENCODE          | 251  | 36756560 | 36757010 |
|           | EBF      | ENCODE ChIP-seq | 173  | 36756693 | 36757001 |
|           | POU2F2   | ENCODE ChIP-seq | 99   | 36756756 | 36756932 |
| DNaseI HS |          | ENCODE          | 139  | 36757180 | 36757330 |
|           | PAX5-C20 | ENCODE ChIP-seq | 114  | 36757186 | 36757378 |
|           | NFKB     | ENCODE ChIP-seq | 102  | 36757305 | 36757343 |
|           | NFKB     | ENCODE ChIP-seq | 287  | 36757720 | 36759504 |
| DNaseI HS |          | ENCODE          | 97   | 36757780 | 36757930 |

|                 |          |                 |      |          |          |
|-----------------|----------|-----------------|------|----------|----------|
|                 | FOSL2    | ENCODE ChIP-seq | 877  | 36757904 | 36758525 |
|                 | RXRA     | ENCODE ChIP-seq | 761  | 36757938 | 36758419 |
|                 | JunD     | ENCODE ChIP-seq | 396  | 36757957 | 36759002 |
|                 | c-Jun    | ENCODE ChIP-seq | 495  | 36757984 | 36758961 |
|                 | BAF155   | ENCODE ChIP-seq | 789  | 36757991 | 36758995 |
|                 | c-Fos    | ENCODE ChIP-seq | 277  | 36757997 | 36759031 |
|                 | Max      | ENCODE ChIP-seq | 363  | 36758006 | 36758905 |
|                 | STAT1    | ENCODE ChIP-seq | 36   | 36758019 | 36758873 |
|                 | p300     | ENCODE ChIP-seq | 171  | 36758048 | 36758305 |
|                 | EBF      | ENCODE ChIP-seq | 1000 | 36758052 | 36758953 |
|                 | HNF4A    | ENCODE ChIP-seq | 274  | 36758055 | 36758427 |
|                 | BAF170   | ENCODE ChIP-seq | 593  | 36758074 | 36758806 |
|                 | c-Myc    | ENCODE ChIP-seq | 437  | 36758080 | 36758940 |
|                 | Ini1     | ENCODE ChIP-seq | 234  | 36758116 | 36758944 |
| DNaseI HS       |          | ENCODE          | 1000 | 36758140 | 36758490 |
|                 | POU2F2   | ENCODE ChIP-seq | 959  | 36758141 | 36759262 |
|                 | AP-4     | TransFac        | 2.12 | 36758267 | 36758277 |
|                 | TCF12    | ENCODE ChIP-seq | 1000 | 36758339 | 36758987 |
|                 | PAX5-N19 | ENCODE ChIP-seq | 100  | 36758431 | 36759049 |
|                 | BCL11A   | ENCODE ChIP-seq | 224  | 36758514 | 36759042 |
| DNaseI HS       |          | ENCODE          | 828  | 36758560 | 36758890 |
|                 | PU.1     | ENCODE ChIP-seq | 396  | 36758595 | 36759031 |
|                 | MIF-1    | TransFac        | 2    | 36758600 | 36758618 |
|                 | BATF     | ENCODE ChIP-seq | 146  | 36758649 | 36759000 |
|                 | SREBP-1a | TransFac        | 1.94 | 36758655 | 36758666 |
|                 | Arnt     | TransFac        | 1.74 | 36758780 | 36758800 |
|                 | E47      | TransFac        | 1.79 | 36758782 | 36758798 |
|                 | MyoD     | TransFac        | 2.14 | 36758785 | 36758795 |
|                 | Meis-1   | TransFac        | 2.24 | 36758787 | 36758799 |
|                 | TGIF     | TransFac        | 2.12 | 36758788 | 36758799 |
| DNaseI HS       |          | ENCODE          | 215  | 36759000 | 36759270 |
|                 | HNF4A    | ENCODE ChIP-seq | 659  | 36759144 | 36759793 |
|                 | CEBPB    | ENCODE ChIP-seq | 468  | 36759299 | 36759882 |
|                 | RXRA     | ENCODE ChIP-seq | 1000 | 36759311 | 36759804 |
|                 | JunD     | ENCODE ChIP-seq | 169  | 36759355 | 36759751 |
|                 | ERRA     | ENCODE ChIP-seq | 493  | 36759356 | 36760283 |
|                 | POU2F2   | ENCODE ChIP-seq | 535  | 36759357 | 36760199 |
|                 | p300     | ENCODE ChIP-seq | 348  | 36759367 | 36759716 |
|                 | ZBTB33   | ENCODE ChIP-seq | 400  | 36759378 | 36759715 |
| DNaseI HS       |          | ENCODE          | 237  | 36759540 | 36760190 |
|                 | NFKB     | ENCODE ChIP-seq | 42   | 36759589 | 36759715 |
|                 | Ini1     | ENCODE ChIP-seq | 181  | 36759686 | 36760016 |
| CDKN1A Exon 3   |          |                 |      | 36759852 | 36760301 |
|                 | c-Myc    | TransFac        | 2.58 | 36760176 | 36760196 |
|                 | c-Myc    | TransFac        | 3.17 | 36760179 | 36760193 |
|                 | POU2F2   | ENCODE ChIP-seq | 225  | 36760249 | 36760695 |
|                 | E2F      | TransFac        | 2.37 | 36760267 | 36760282 |
| CDKN1A Intron 3 |          |                 |      | 36760302 | 36761505 |
| DNaseI HS       |          | ENCODE          | 201  | 36760440 | 36760590 |
| DNaseI HS       |          | ENCODE          | 143  | 36761140 | 36761330 |
| DNaseI HS       |          | ENCODE          | 291  | 36761440 | 36761610 |
|                 | POU2F2   | ENCODE ChIP-seq | 1000 | 36761442 | 36763056 |
| CDKN1A Exon 4   |          |                 |      | 36761506 | 36763095 |
|                 | Cart-1   | TransFac        | 1.98 | 36761641 | 36761659 |



|           |            |           |       |            |                 |      |          |          |
|-----------|------------|-----------|-------|------------|-----------------|------|----------|----------|
|           |            |           |       | Cdc5       | TransFac        | 1.99 | 36761658 | 36761670 |
| DNaseI HS |            |           |       |            | ENCODE          | 390  | 36761820 | 36762090 |
|           |            |           |       | LCR-F1     | TransFac        | 2.17 | 36762093 | 36762106 |
| DNaseI HS |            |           |       |            | ENCODE          | 143  | 36762180 | 36762590 |
|           |            |           |       | NF-kappaB1 | TransFac        | 1.66 | 36762253 | 36762263 |
| DNaseI HS |            |           |       |            | ENCODE          | 125  | 36762680 | 36762830 |
|           |            |           |       | NFKB       | ENCODE ChIP-seq | 32   | 36762791 | 36763034 |
| DNaseI HS |            |           |       |            | ENCODE          | 226  | 36762840 | 36763070 |
|           |            |           |       | FOXF2      | TransFac        | 2.02 | 36762918 | 36762934 |
|           |            |           |       | POU2F2     | ENCODE ChIP-seq | 603  | 36763148 | 36763609 |
|           |            |           |       | Input      | ENCODE ChIP-seq | 172  | 36763218 | 36763437 |
| DNaseI HS |            |           |       |            | ENCODE          | 187  | 36763300 | 36763570 |
|           | rs876581   | Genotyped | 0.594 |            |                 |      | 36763423 |          |
|           |            |           |       | POU2F2     | ENCODE ChIP-seq | 704  | 36763633 | 36764117 |
|           |            |           |       | PAX5-N19   | ENCODE ChIP-seq | 94   | 36763718 | 36764126 |
| DNaseI HS |            |           |       |            | ENCODE          | 369  | 36763740 | 36764730 |
|           |            |           |       | POU2F2     | ENCODE ChIP-seq | 525  | 36764132 | 36764788 |
|           |            |           |       | NFKB       | ENCODE ChIP-seq | 101  | 36764148 | 36764798 |
|           | rs12207548 | Genotyped | 0.740 |            |                 |      | 36764234 |          |
| DNaseI HS |            |           |       |            | ENCODE          | 51   | 36765000 | 36765150 |
|           |            |           |       | NFKB       | ENCODE ChIP-seq | 92   | 36765558 | 36765674 |
| DNaseI HS |            |           |       |            | ENCODE          | 115  | 36766880 | 36767090 |
|           |            |           |       | NFKB       | ENCODE ChIP-seq | 87   | 36767820 | 36768456 |
|           |            |           |       | PAX5-N19   | ENCODE ChIP-seq | 139  | 36767846 | 36768332 |
|           |            |           |       | PU.1       | ENCODE ChIP-seq | 1000 | 36767880 | 36768436 |
|           |            |           |       | PAX5-C20   | ENCODE ChIP-seq | 92   | 36768009 | 36768254 |
| DNaseI HS |            |           |       |            | ENCODE          | 1000 | 36768020 | 36768270 |
|           |            |           |       | Hlf        | TransFac        | 2.4  | 36768262 | 36768272 |
|           |            |           |       | ATF-2      | TransFac        | 2.5  | 36768263 | 36768271 |
| DNaseI HS |            |           |       |            | ENCODE          | 112  | 36768660 | 36768810 |
| DNaseI HS |            |           |       |            | ENCODE          | 128  | 36768960 | 36769170 |
|           | rs2395656  | Genotyped | 0.656 |            |                 |      | 36770149 |          |

### 11q13.4 (rs3824999)

| Gene region | rsID       | SNP type  | P-value  | TF type | Source          | Score | Chrom start | Chrom end |
|-------------|------------|-----------|----------|---------|-----------------|-------|-------------|-----------|
|             | rs11236148 | Genotyped | 9.17E-03 |         |                 |       | 73941983    |           |
| DNaseI HS   |            |           |          |         | ENCODE          | 91    | 73942260    | 73942510  |
|             | rs1655483  | Genotyped | 0.034    |         |                 |       | 73942425    |           |
|             | rs1783200  | Imputed   | 2.81E-03 |         |                 |       | 73942542    |           |
|             | rs7930294  | Imputed   | 0.745    |         |                 |       | 73942862    |           |
|             | rs1791495  | Imputed   | 6.62E-05 |         |                 |       | 73943628    |           |
|             | rs2887046  | Genotyped | 0.379    |         |                 |       | 73943674    |           |
|             | rs4044287  | Imputed   | 0.133    |         |                 |       | 73944835    |           |
|             |            |           |          | PU.1    | ENCODE ChIP-seq | 355   | 73946169    | 73946357  |
|             | rs12795302 | Imputed   | 1.50E-03 |         |                 |       | 73946320    |           |
|             | rs12801229 | Imputed   | 9.35E-04 |         |                 |       | 73946819    |           |
|             | rs1292504  | Genotyped | 1.56E-06 |         |                 |       | 73947278    |           |
|             | rs3902018  | Genotyped | 1.52E-03 |         |                 |       | 73947465    |           |
|             | rs10793091 | Imputed   | 0.195    |         |                 |       | 73947740    |           |
| DNaseI HS   |            |           |          |         | ENCODE          | 98    | 73947940    | 73948230  |
|             | rs10501416 | Imputed   | 1.40E-03 |         |                 |       | 73947977    |           |

|           |            |           |          |       |                 |      |          |          |
|-----------|------------|-----------|----------|-------|-----------------|------|----------|----------|
|           |            |           |          | p300  | ENCODE ChIP-seq | 730  | 73948212 | 73948661 |
|           |            |           |          | RXRA  | ENCODE ChIP-seq | 218  | 73948221 | 73948637 |
|           |            |           |          | FOSL2 | ENCODE ChIP-seq | 157  | 73948226 | 73948906 |
| DNaseI HS |            |           |          |       | ENCODE          | 385  | 73948300 | 73948670 |
|           |            |           |          | c-Jun | ENCODE ChIP-seq | 200  | 73948379 | 73948973 |
|           |            |           |          | Elk-1 | TransFac        | 2.38 | 73948476 | 73948492 |
|           |            |           |          | CREB  | TransFac        | 2.2  | 73948667 | 73948679 |
|           |            |           |          | ATF-2 | TransFac        | 2    | 73948669 | 73948677 |
| DNaseI HS |            |           |          |       | ENCODE          | 356  | 73948700 | 73948930 |
|           |            |           |          | JunD  | ENCODE ChIP-seq | 647  | 73948853 | 73948869 |
|           | rs4315111  | Imputed   | 0.173    |       |                 |      | 73949244 |          |
|           | rs7947451  | Imputed   | 0.172    |       |                 |      | 73949417 |          |
|           | rs11236152 | Imputed   | 0.171    |       |                 |      | 73949906 |          |
|           | rs7130461  | Imputed   | 0.170    |       |                 |      | 73950487 |          |
|           | rs7941941  | Genotyped | 0.180    |       |                 |      | 73951606 |          |
|           | rs7929392  | Imputed   | 0.168    |       |                 |      | 73951655 |          |
|           | rs4439539  | Imputed   | 0.174    |       |                 |      | 73951677 |          |
|           | rs10899007 | Imputed   | 0.134    |       |                 |      | 73952433 |          |
| DNaseI HS |            |           |          |       | ENCODE          | 182  | 73953120 | 73953410 |
|           |            |           |          | FAC1  | TransFac        | 2.72 | 73953403 | 73953417 |
|           |            |           |          | FOXL1 | TransFac        | 2.39 | 73953420 | 73953436 |
|           |            |           |          | NFKB  | ENCODE ChIP-seq | 332  | 73953472 | 73953835 |
|           |            |           |          | USF-1 | ENCODE ChIP-seq | 119  | 73953534 | 73953681 |
|           | rs6592568  | Imputed   | 0.026    |       |                 |      | 73953542 |          |
| DNaseI HS |            |           |          |       | ENCODE          | 150  | 73953760 | 73953930 |
|           | rs10899009 | Imputed   | 3.50E-07 |       |                 |      | 73953815 |          |
|           | rs11604752 | Imputed   | 3.48E-07 |       |                 |      | 73954792 |          |
| DNaseI HS |            |           |          |       | ENCODE          | 280  | 73955180 | 73955510 |
|           |            |           |          | NFKB  | ENCODE ChIP-seq | 107  | 73955296 | 73955593 |
| DNaseI HS |            |           |          |       | ENCODE          | 208  | 73955660 | 73955810 |
|           | rs4944047  | Imputed   | 0.011    |       |                 |      | 73955738 |          |
|           | rs1433969  | Imputed   | 0.029    |       |                 |      | 73956081 |          |
| DNaseI HS |            |           |          |       | ENCODE          | 173  | 73956160 | 73956310 |
|           |            |           |          | MRF-2 | TransFac        | 1.97 | 73956182 | 73956196 |
|           |            |           |          | CUTL1 | TransFac        | 1.8  | 73956343 | 73956358 |
|           |            |           |          | Cdc5  | TransFac        | 1.65 | 73956353 | 73956365 |
|           | rs10793092 | Imputed   | 4.91E-07 |       |                 |      | 73956355 |          |
|           |            |           |          | Pbx1a | TransFac        | 1.71 | 73956355 | 73956370 |
|           | rs11826567 | Imputed   | 8.74E-03 |       |                 |      | 73956941 |          |
|           | rs7943504  | Imputed   | 8.72E-03 |       |                 |      | 73957654 |          |
|           | rs7121958  | Imputed   | 3.42E-07 |       |                 |      | 73957660 |          |
|           | rs4944903  | Genotyped | 0.203    |       |                 |      | 73957984 |          |
|           | rs6592570  | Imputed   | 1.13E-03 |       |                 |      | 73958558 |          |
|           | rs1971114  | Imputed   | 0.026    |       |                 |      | 73959054 |          |
|           |            |           |          | Rad21 | ENCODE ChIP-seq | 734  | 73959143 | 73959606 |
|           |            |           |          | CTCF  | ENCODE ChIP-seq | 1000 | 73959184 | 73959578 |
|           |            |           |          | NFKB  | ENCODE ChIP-seq | 51   | 73959261 | 73959526 |
| DNaseI HS |            |           |          |       | ENCODE          | 352  | 73959280 | 73959570 |
|           | rs7952661  | Imputed   | 0.026    |       |                 |      | 73959425 |          |
|           | rs7937232  | Imputed   | 0.025    |       |                 |      | 73959558 |          |
|           | rs72991606 | Imputed   | 3.16E-05 |       |                 |      | 73959768 |          |
|           | rs1433972  | Imputed   | 1.59E-03 |       |                 |      | 73960574 |          |
|           | rs10751238 | Imputed   | 0.036    |       |                 |      | 73960645 |          |
|           | rs10751239 | Imputed   | 0.026    |       |                 |      | 73960646 |          |
|           | rs11236154 | Imputed   | 0.026    |       |                 |      | 73960818 |          |
|           | rs7113533  | Genotyped | 0.064    |       |                 |      | 73961919 |          |
|           | rs4944904  | Imputed   | 0.026    |       |                 |      | 73963540 |          |
|           | rs10899010 | Imputed   | 0.026    |       |                 |      | 73963794 |          |

|           |                   |                |                 |          |                 |      |                 |          |
|-----------|-------------------|----------------|-----------------|----------|-----------------|------|-----------------|----------|
|           |                   |                |                 | MyoD     | TransFac        | 1.93 | 73965508        | 73965518 |
|           | rs4944048         | Imputed        | 0.027           |          |                 |      | 73965974        |          |
|           |                   |                |                 | Sox9     | TransFac        | 3.43 | 73966031        | 73966045 |
|           | rs12290016        | Imputed        | 1.26E-03        |          |                 |      | 73966297        |          |
|           |                   |                |                 | BCL3     | ENCODE ChIP-seq | 412  | 73966348        | 73966661 |
|           | rs36005373        | Imputed        | 7.91E-03        |          |                 |      | 73966365        |          |
|           |                   |                |                 | Pbx3     | ENCODE ChIP-seq | 151  | 73966379        | 73966589 |
|           | rs9665746         | Imputed        | 2.47E-03        |          |                 |      | 73966534        |          |
|           | rs9667597         | Imputed        | 2.47E-03        |          |                 |      | 73966555        |          |
|           | rs4944906         | Imputed        | 2.47E-03        |          |                 |      | 73966570        |          |
| DNaseI HS |                   |                |                 |          | ENCODE          | 497  | 73966780        | 73966990 |
|           | rs35165052        | Imputed        | 0.083           |          |                 |      | 73967273        |          |
|           | <b>rs11236158</b> | <b>Imputed</b> | <b>3.44E-07</b> |          |                 |      | <b>73967376</b> |          |
|           |                   |                |                 | CUTL1    | TransFac        | 2.63 | 73968423        | 73968438 |
|           | rs9919665         | Imputed        | 1.32E-03        |          |                 |      | 73968469        |          |
|           |                   |                |                 | FOXD1    | TransFac        | 1.76 | 73968479        | 73968495 |
|           |                   |                |                 | FOXO4    | TransFac        | 2.34 | 73968481        | 73968495 |
|           | rs9919666         | Genotyped      | 8.52E-03        |          |                 |      | 73968529        |          |
|           | rs10793093        | Genotyped      | 2.10E-04        |          |                 |      | 73968600        |          |
|           | rs4944907         | Imputed        | 2.85E-03        |          |                 |      | 73969005        |          |
|           | <b>rs11236161</b> | <b>Imputed</b> | <b>2.41E-07</b> |          |                 |      | <b>73969361</b> |          |
|           |                   |                |                 | PU.1     | ENCODE ChIP-seq | 502  | 73969746        | 73970154 |
|           |                   |                |                 | IRF4     | ENCODE ChIP-seq | 674  | 73969799        | 73970284 |
|           |                   |                |                 | SP1      | ENCODE ChIP-seq | 189  | 73969821        | 73970217 |
| DNaseI HS |                   |                |                 |          | ENCODE          | 468  | 73969840        | 73970190 |
|           |                   |                |                 | PAX5-N19 | ENCODE ChIP-seq | 221  | 73969851        | 73970326 |
|           |                   |                |                 | BATF     | ENCODE ChIP-seq | 1000 | 73969858        | 73970236 |
|           |                   |                |                 | BCL11A   | ENCODE ChIP-seq | 380  | 73969874        | 73970202 |
|           |                   |                |                 | NFKB     | ENCODE ChIP-seq | 85   | 73969904        | 73970215 |
|           |                   |                |                 | PAX5-C20 | ENCODE ChIP-seq | 98   | 73969975        | 73970268 |
|           | <b>rs60085010</b> | <b>Imputed</b> | <b>2.39E-07</b> |          |                 |      | <b>73970261</b> |          |
|           | rs9919663         | Imputed        | 7.40E-03        |          |                 |      | 73970373        |          |
|           | rs7935260         | Imputed        | 0.024           |          |                 |      | 73971187        |          |
|           | rs7938931         | Imputed        | 0.024           |          |                 |      | 73971868        |          |
|           | rs7939930         | Genotyped      | 8.57E-03        |          |                 |      | 73972014        |          |
|           | rs12292725        | Imputed        | 4.82E-03        |          |                 |      | 73972273        |          |
|           | rs7929876         | Imputed        | 0.024           |          |                 |      | 73972419        |          |
|           | rs11236164        | Genotyped      | 1.13E-04        |          |                 |      | 73972614        |          |
|           |                   |                |                 | Rad21    | ENCODE ChIP-seq | 228  | 73972786        | 73972969 |
|           |                   |                |                 | AP-2rep  | TransFac        | 2.23 | 73972888        | 73972895 |
|           | rs12364155        | Imputed        | 5.60E-03        |          |                 |      | 73973383        |          |
|           | rs7934776         | Imputed        | 4.91E-03        |          |                 |      | 73974498        |          |
|           |                   |                |                 | FOXC1    | TransFac        | 2.12 | 73975129        | 73975145 |
|           | rs34222102        | Imputed        | 3.16E-03        |          |                 |      | 73975606        |          |
|           | rs4417306         | Imputed        | 4.98E-03        |          |                 |      | 73975658        |          |
|           | rs4573702         | Imputed        | 4.99E-03        |          |                 |      | 73977147        |          |
|           | rs4627114         | Imputed        | 0.024           |          |                 |      | 73977466        |          |
|           | rs4335575         | Imputed        | 5.01E-03        |          |                 |      | 73977488        |          |
|           | rs34835838        | Imputed        | 0.046           |          |                 |      | 73977658        |          |
|           | rs10899012        | Imputed        | 3.11E-03        |          |                 |      | 73977882        |          |
|           | <b>rs72977282</b> | <b>Imputed</b> | <b>1.34E-07</b> |          |                 |      | <b>73978089</b> |          |
|           | rs7944514         | Imputed        | 3.46E-03        |          |                 |      | 73978840        |          |
| DNaseI HS |                   |                |                 |          | ENCODE          | 199  | 73979320        | 73979490 |
|           | rs11236165        | Imputed        | 5.09E-03        |          |                 |      | 73979918        |          |
|           | rs12362729        | Imputed        | 0.053           |          |                 |      | 73979965        |          |
| DNaseI HS |                   |                |                 |          | ENCODE          | 120  | 73980260        | 73980410 |
|           |                   |                |                 | STAT1    | ENCODE ChIP-seq | 921  | 73980589        | 73980597 |
|           |                   |                |                 | BAF170   | ENCODE ChIP-seq | 441  | 73980630        | 73980941 |

|                |            |           |          |           |                 |          |          |          |
|----------------|------------|-----------|----------|-----------|-----------------|----------|----------|----------|
| DNaseI HS      |            |           |          | ENCODE    | 121             | 73980800 | 73981050 |          |
|                |            |           |          | NFKB      | ENCODE ChIP-seq | 185      | 73981001 | 73981966 |
|                |            |           |          | STAT1     | ENCODE ChIP-seq | 60       | 73981011 | 73982132 |
|                |            |           |          | c-Myc     | ENCODE ChIP-seq | 365      | 73981018 | 73981981 |
|                |            |           |          | FOSL2     | ENCODE ChIP-seq | 126      | 73981022 | 73981396 |
|                |            |           |          | TAF1      | ENCODE ChIP-seq | 829      | 73981049 | 73981862 |
|                |            |           |          | HEY1      | ENCODE ChIP-seq | 233      | 73981052 | 73981611 |
|                |            |           |          | JunD      | ENCODE ChIP-seq | 97       | 73981059 | 73981355 |
|                |            |           |          | Sin3Ak-20 | ENCODE ChIP-seq | 1000     | 73981062 | 73981399 |
|                |            |           |          | SP1       | ENCODE ChIP-seq | 240      | 73981065 | 73981433 |
|                |            |           |          | HNF4A     | ENCODE ChIP-seq | 388      | 73981065 | 73981400 |
|                |            |           |          | TCF12     | ENCODE ChIP-seq | 177      | 73981072 | 73981400 |
|                |            |           |          | CEBPB     | ENCODE ChIP-seq | 153      | 73981079 | 73981941 |
|                |            |           |          | USF-1     | ENCODE ChIP-seq | 66       | 73981080 | 73981338 |
|                |            |           |          | Max       | ENCODE ChIP-seq | 528      | 73981082 | 73982034 |
|                |            |           |          | IRF4      | ENCODE ChIP-seq | 301      | 73981084 | 73981496 |
|                |            |           |          | Ini1      | ENCODE ChIP-seq | 207      | 73981085 | 73982092 |
|                |            |           |          | POU2F2    | ENCODE ChIP-seq | 378      | 73981086 | 73981577 |
|                |            |           |          | NRSF      | ENCODE ChIP-seq | 684      | 73981099 | 73981423 |
|                |            |           |          | BAF155    | ENCODE ChIP-seq | 622      | 73981104 | 73982086 |
|                |            |           |          | BATF      | ENCODE ChIP-seq | 73       | 73981111 | 73981345 |
| DNaseI HS      |            |           |          | ENCODE    | 1000            | 73981120 | 73981350 |          |
|                |            |           |          | HSF1      | ENCODE ChIP-seq | 274      | 73981126 | 73981546 |
|                |            |           |          | p300      | ENCODE ChIP-seq | 183      | 73981127 | 73981365 |
| POLD3 Exon 1   |            |           |          |           |                 | 73981277 | 73981411 |          |
|                |            |           |          | E2F       | TransFac        | 2.57     | 73981281 | 73981289 |
|                |            |           |          | BAF170    | ENCODE ChIP-seq | 572      | 73981296 | 73982112 |
| POLD3 Intron 1 |            |           |          |           |                 | 73981412 | 73982753 |          |
|                | rs2298572  | Imputed   | 0.025    |           |                 | 73981489 |          |          |
| DNaseI HS      |            |           |          | ENCODE    | 489             | 73981520 | 73981950 |          |
| DNaseI HS      |            |           |          | ENCODE    | 106             | 73981958 | 73982150 |          |
|                | rs7939226  | Genotyped | 0.082    |           |                 | 73982157 |          |          |
|                |            |           |          | p300      | ENCODE ChIP-seq | 105      | 73982168 | 73982433 |
| DNaseI HS      |            |           |          | ENCODE    | 107             | 73982260 | 73982450 |          |
|                |            |           |          | BAF170    | ENCODE ChIP-seq | 643      | 73982322 | 73982497 |
|                |            |           |          | NFKB      | ENCODE ChIP-seq | 164      | 73982329 | 73982375 |
| POLD3 Exon 2   |            |           |          |           |                 | 73982754 | 73982809 |          |
| POLD3 Intron 2 |            |           |          |           |                 | 73982810 | 73993384 |          |
|                |            |           |          | PAX5-C20  | ENCODE ChIP-seq | 83       | 73983298 | 73983567 |
|                | rs4944049  | Imputed   | 0.024    |           |                 | 73984058 |          |          |
|                | rs4944050  | Imputed   | 0.024    |           |                 | 73984142 |          |          |
|                | rs7125480  | Imputed   | 5.47E-03 |           |                 | 73984436 |          |          |
|                | rs4944911  | Imputed   | 0.024    |           |                 | 73985706 |          |          |
| DNaseI HS      |            |           |          | ENCODE    | 82              | 73986160 | 73986330 |          |
|                | rs61901537 | Imputed   | 3.05E-03 |           |                 | 73986667 |          |          |
|                | rs10899013 | Genotyped | 6.19E-03 |           |                 | 73987190 |          |          |
|                | rs61901538 | Imputed   | 5.86E-03 |           |                 | 73988107 |          |          |
|                | rs7934321  | Imputed   | 0.024    |           |                 | 73988327 |          |          |
|                | rs7934742  | Imputed   | 0.023    |           |                 | 73988699 |          |          |
|                | rs12801024 | Imputed   | 6.10E-03 |           |                 | 73989133 |          |          |
|                | rs34702292 | Imputed   | 3.35E-03 |           |                 | 73989272 |          |          |
|                | rs34060848 | Imputed   | 0.091    |           |                 | 73989315 |          |          |
|                | rs7932731  | Imputed   | 6.17E-03 |           |                 | 73989436 |          |          |
|                |            |           |          | BAF170    | ENCODE ChIP-seq | 1000     | 73989805 | 73989986 |
|                |            |           |          | POU3F2    | TransFac        | 2.6      | 73989932 | 73989942 |
|                | rs6592573  | Imputed   | 6.49E-03 |           |                 | 73990610 |          |          |
|                | rs7925556  | Imputed   | 0.025    |           |                 | 73990935 |          |          |
|                |            |           |          | POU2F1    | TransFac        | 1.82     | 73991924 | 73991937 |

|                |            |           |          |         |                 |          |          |          |
|----------------|------------|-----------|----------|---------|-----------------|----------|----------|----------|
| DNaseI HS      |            |           |          | ENCODE  | 68              | 73991960 | 73992210 |          |
|                | rs61901540 | Imputed   | 0.051    |         |                 | 73992513 |          |          |
|                |            |           |          | RSRFC4  | TransFac        | 1.77     | 73993128 | 73993144 |
| POLD3 Exon 3   |            |           |          |         |                 | 73993385 | 73993487 |          |
|                |            |           |          | HSF1    | TransFac        | 1.79     | 73993417 | 73993427 |
|                |            |           |          | E47     | TransFac        | 3.26     | 73993450 | 73993466 |
| POLD3 Intron 3 |            |           |          |         |                 | 73993488 | 74000191 |          |
|                |            |           |          | PU.1    | ENCODE ChIP-seq | 470      | 73994088 | 73994445 |
|                |            |           |          | EBF     | ENCODE ChIP-seq | 266      | 73994134 | 73994456 |
| DNaseI HS      |            |           |          | ENCODE  | 63              | 73994700 | 73994850 |          |
|                | rs7940880  | Imputed   | 1.54E-06 |         |                 | 73995062 |          |          |
|                | rs11236171 | Imputed   | 0.051    |         |                 | 73995166 |          |          |
|                | rs61901541 | Imputed   | 0.080    |         |                 | 73995633 |          |          |
|                | rs7124188  | Imputed   | 1.28E-06 |         |                 | 73995994 |          |          |
|                |            |           |          | CEBPB   | ENCODE ChIP-seq | 890      | 73998625 | 73998873 |
|                |            |           |          | RXRA    | ENCODE ChIP-seq | 225      | 73998635 | 73998801 |
|                | rs4944912  | Imputed   | 1.36E-03 |         |                 | 73998929 |          |          |
|                | rs4944913  | Imputed   | 6.69E-03 |         |                 | 73998997 |          |          |
|                | rs4944914  | Imputed   | 1.37E-03 |         |                 | 73999053 |          |          |
|                | rs12296094 | Imputed   | 0.027    |         |                 | 73999348 |          |          |
|                | rs4944915  | Imputed   | 0.018    |         |                 | 73999612 |          |          |
|                | rs4944916  | Imputed   | 0.018    |         |                 | 73999688 |          |          |
|                |            |           |          | CTCF    | ENCODE ChIP-seq | 155      | 73999999 | 74000392 |
| DNaseI HS      |            |           |          | ENCODE  | 866             | 74000080 | 74000590 |          |
|                |            |           |          | Rad21   | ENCODE ChIP-seq | 267      | 74000105 | 74000338 |
|                |            |           |          | Ini1    | ENCODE ChIP-seq | 786      | 74000156 | 74000372 |
| POLD3 Exon 4   |            |           |          |         |                 | 74000192 | 74000231 |          |
| POLD3 Intron 4 |            |           |          |         |                 | 74000232 | 74001570 |          |
|                | rs6592574  | Genotyped | 5.43E-04 |         |                 | 74000610 |          |          |
|                | rs6592575  | Imputed   | 0.019    |         |                 | 74000654 |          |          |
|                | rs7122710  | Imputed   | 0.052    |         |                 | 74000823 |          |          |
|                | rs4944917  | Imputed   | 0.049    |         |                 | 74000929 |          |          |
|                |            |           |          | CREB    | TransFac        | 1.67     | 74001330 | 74001345 |
|                |            |           |          | E47     | TransFac        | 1.93     | 74001341 | 74001357 |
| POLD3 Exon 5   |            |           |          |         |                 | 74001571 | 74001703 |          |
|                |            |           |          | LUN-1   | TransFac        | 1.92     | 74001577 | 74001594 |
| DNaseI HS      |            |           |          | ENCODE  | 259             | 74001640 | 74001850 |          |
|                |            |           |          | AP-2rep | TransFac        | 2.23     | 74001640 | 74001647 |
|                |            |           |          | MRF-2   | TransFac        | 2.29     | 74001650 | 74001664 |
|                |            |           |          | TBP     | TransFac        | 1.92     | 74001672 | 74001682 |
| POLD3 Intron 5 |            |           |          |         |                 | 74001704 | 74007229 |          |
|                |            |           |          | Nkx3-1  | TransFac        | 1.74     | 74001839 | 74001851 |
|                | rs10219203 | Genotyped | 3.66E-04 |         |                 | 74002571 |          |          |
|                | rs10899014 | Imputed   | 0.057    |         |                 | 74002757 |          |          |
|                | rs4944051  | Imputed   | 0.053    |         |                 | 74002983 |          |          |
|                | rs7120069  | Imputed   | 0.018    |         |                 | 74003036 |          |          |
|                | rs7130839  | Imputed   | 3.05E-03 |         |                 | 74003231 |          |          |
|                | rs7101472  | Imputed   | 3.06E-03 |         |                 | 74003265 |          |          |
|                | rs4584608  | Imputed   | 0.018    |         |                 | 74003296 |          |          |
|                | rs4996254  | Imputed   | 1.66E-03 |         |                 | 74003306 |          |          |
|                |            |           |          | Input   | ENCODE ChIP-seq | 252      | 74003652 | 74003836 |
|                |            |           |          | PU.1    | ENCODE ChIP-seq | 473      | 74004062 | 74004339 |
| DNaseI HS      |            |           |          | ENCODE  | 356             | 74004080 | 74004310 |          |
|                | rs6592576  | Imputed   | 0.018    |         |                 | 74004469 |          |          |
|                | rs7932194  | Imputed   | 0.027    |         |                 | 74005026 |          |          |
|                | rs6592577  | Genotyped | 0.021    |         |                 | 74005262 |          |          |
|                |            |           |          | AP-2rep | TransFac        | 2.23     | 74005292 | 74005299 |
|                |            |           |          | E47     | TransFac        | 1.81     | 74005614 | 74005630 |

|                |            |           |          |         |                 |      |          |          |
|----------------|------------|-----------|----------|---------|-----------------|------|----------|----------|
|                | rs10899015 | Imputed   | 1.71E-03 |         |                 |      | 74006732 |          |
| POLD3 Exon 6   |            |           |          |         |                 |      | 74007230 | 74007497 |
|                | rs4944052  | Imputed   | 1.67E-03 |         |                 |      | 74007263 |          |
|                |            |           |          | CUTL1   | TransFac        | 2.36 | 74007355 | 74007370 |
|                |            |           |          | E47     | TransFac        | 1.78 | 74007367 | 74007382 |
| POLD3 Intron 6 |            |           |          |         |                 |      | 74007498 | 74008709 |
|                | rs7943085  | Imputed   | 1.59E-03 |         |                 |      | 74007856 |          |
|                | rs7944208  | Imputed   | 1.62E-03 |         |                 |      | 74008042 |          |
|                | rs11236172 | Imputed   | 0.050    |         |                 |      | 74008536 |          |
|                | rs10501417 | Imputed   | 1.53E-03 |         |                 |      | 74008628 |          |
| DNaseI HS      |            |           |          |         | ENCODE          | 70   | 74008680 | 74008850 |
| POLD3 Exon 7   |            |           |          |         |                 |      | 74008710 | 74008782 |
| POLD3 Intron 7 |            |           |          |         |                 |      | 74008783 | 74014100 |
| DNaseI HS      |            |           |          |         | ENCODE          | 211  | 74009340 | 74009530 |
|                | rs4145953  | Genotyped | 0.042    |         |                 |      | 74009527 |          |
|                | rs10899016 | Imputed   | 0.018    |         |                 |      | 74009849 |          |
|                | rs11236173 | Imputed   | 1.38E-03 |         |                 |      | 74009910 |          |
|                | rs12288519 | Imputed   | 1.45E-03 |         |                 |      | 74010227 |          |
|                | rs12288525 | Imputed   | 0.026    |         |                 |      | 74010237 |          |
|                | rs12287097 | Imputed   | 0.050    |         |                 |      | 74010312 |          |
|                | rs12280434 | Imputed   | 0.026    |         |                 |      | 74010475 |          |
|                | rs59768581 | Imputed   | 1.55E-06 |         |                 |      | 74010784 |          |
|                | rs10899017 | Imputed   | 0.050    |         |                 |      | 74010945 |          |
|                | rs12807398 | Imputed   | 0.046    |         |                 |      | 74011387 |          |
|                | rs11236174 | Imputed   | 0.050    |         |                 |      | 74011849 |          |
|                | rs11236175 | Imputed   | 1.30E-03 |         |                 |      | 74011992 |          |
|                | rs11236176 | Imputed   | 1.30E-03 |         |                 |      | 74011996 |          |
|                | rs10899018 | Imputed   | 1.28E-03 |         |                 |      | 74012303 |          |
|                | rs10899019 | Imputed   | 0.026    |         |                 |      | 74012810 |          |
|                |            |           |          | RSRFC4  | TransFac        | 1.92 | 74013247 | 74013263 |
|                | rs10899020 | Imputed   | 1.14E-06 |         |                 |      | 74013417 |          |
|                | rs10793094 | Imputed   | 1.14E-06 |         |                 |      | 74013473 |          |
| POLD3 Exon 8   |            |           |          |         |                 |      | 74014101 | 74014266 |
| POLD3 Intron 8 |            |           |          |         |                 |      | 74014267 | 74017909 |
|                |            |           |          | Input   | ENCODE ChIP-seq | 184  | 74014337 | 74014513 |
|                |            |           |          | FOXO4   | TransFac        | 2.11 | 74014658 | 74014672 |
|                |            |           |          | Lmo2    | TransFac        | 2.18 | 74014830 | 74014839 |
|                |            |           |          | AP-2rep | TransFac        | 2.23 | 74014929 | 74014936 |
|                | rs2117222  | Imputed   | 9.76E-07 |         |                 |      | 74015333 |          |
|                | rs10899021 | Imputed   | 0.056    |         |                 |      | 74015702 |          |
|                | rs57796856 | Imputed   | 9.24E-07 |         |                 |      | 74016003 |          |
|                |            |           |          | BATF    | ENCODE ChIP-seq | 80   | 74016179 | 74016359 |
|                | rs59830047 | Imputed   | 9.09E-07 |         |                 |      | 74016183 |          |
|                |            |           |          | NFKB    | ENCODE ChIP-seq | 152  | 74016209 | 74016400 |
|                | rs11236177 | Imputed   | 0.027    |         |                 |      | 74016715 |          |
|                | rs1433970  | Imputed   | 0.035    |         |                 |      | 74016841 |          |
|                | rs10899022 | Imputed   | 0.027    |         |                 |      | 74017034 |          |
|                | rs2155935  | Imputed   | 8.04E-07 |         |                 |      | 74017225 |          |
|                | rs2298792  | Imputed   | 7.40E-07 |         |                 |      | 74017844 |          |
|                |            |           |          | Nkx3-1  | TransFac        | 2.13 | 74017852 | 74017864 |
| POLD3 Exon 9   |            |           |          |         |                 |      | 74017910 | 74018016 |
|                |            |           |          | IRF-7A  | TransFac        | 2.56 | 74017943 | 74017961 |
| POLD3 Intron 9 |            |           |          |         |                 |      | 74018017 | 74023275 |
|                |            |           |          | PU.1    | ENCODE ChIP-seq | 164  | 74018799 | 74019095 |
|                | rs11236178 | Imputed   | 7.22E-07 |         |                 |      | 74018984 |          |
|                | rs4944053  | Imputed   | 8.30E-04 |         |                 |      | 74019358 |          |
|                |            |           |          | NFKB    | ENCODE ChIP-seq | 281  | 74020374 | 74020707 |
|                |            |           |          | BATF    | ENCODE ChIP-seq | 135  | 74020409 | 74020651 |

|                 |                   |                  |                 |        |                 |          |                 |
|-----------------|-------------------|------------------|-----------------|--------|-----------------|----------|-----------------|
| DNaseI HS       |                   |                  |                 | ENCODE | 877             | 74020440 | 74020630        |
|                 | rs11236180        | Genotyped        | 0.329           |        |                 | 74020576 |                 |
|                 |                   |                  |                 | E47    | TransFac        | 2.02     | 74020704        |
|                 |                   |                  |                 | STAT3  | TransFac        | 2.09     | 74020898        |
| DNaseI HS       |                   |                  |                 | ENCODE | 95              | 74021640 | 74021790        |
|                 |                   |                  |                 | IRF-7A | TransFac        | 2.23     | 74022227        |
|                 |                   |                  |                 | POU2F1 | TransFac        | 1.74     | 74022231        |
| DNaseI HS       |                   |                  |                 | ENCODE | 83              | 74022240 | 74022430        |
|                 |                   |                  |                 | AREB6  | TransFac        | 2.31     | 74022258        |
|                 |                   |                  |                 | AML1a  | TransFac        | 1.68     | 74022307        |
|                 |                   |                  |                 | Lmo2   | TransFac        | 2.05     | 74022507        |
|                 |                   |                  |                 | JunD   | ENCODE ChIP-seq | 173      | 74023143        |
|                 |                   |                  |                 | c-Jun  | ENCODE ChIP-seq | 176      | 74023188        |
|                 | <b>rs3824999</b>  | <b>Genotyped</b> | <b>1.49E-06</b> |        |                 |          | <b>74023198</b> |
| DNaseI HS       |                   |                  |                 | ENCODE | 1000            | 74023200 | 74023570        |
|                 |                   |                  |                 | ISGF-3 | TransFac        | 2.26     | 74023258        |
| POLD3 Exon 10   |                   |                  |                 |        |                 | 74023276 | 74023388        |
|                 |                   |                  |                 | CTCF   | ENCODE ChIP-seq | 564      | 74023287        |
|                 |                   |                  |                 | Meis-1 | TransFac        | 1.97     | 74023377        |
|                 |                   |                  |                 | Rad21  | ENCODE ChIP-seq | 133      | 74023382        |
| POLD3 Intron 10 |                   |                  |                 |        |                 | 74023389 | 74024889        |
|                 |                   |                  |                 | STAT1  | ENCODE ChIP-seq | 130      | 74023407        |
|                 | rs11236181        | Genotyped        | 0.072           |        |                 | 74023471 |                 |
|                 |                   |                  |                 | AREB6  | TransFac        | 1.9      | 74023555        |
|                 | <b>rs11236182</b> | <b>Imputed</b>   | <b>5.40E-07</b> |        |                 |          | <b>74024170</b> |
| DNaseI HS       |                   |                  |                 | ENCODE | 144             | 74024180 | 74024370        |
|                 | rs10899023        | Imputed          | 1.42E-03        |        |                 | 74024340 |                 |
|                 | rs3741127         | Imputed          | 0.173           |        |                 | 74024581 |                 |
| POLD3 Exon 11   |                   |                  |                 |        |                 | 74024890 | 74024968        |
| POLD3 Intron 11 |                   |                  |                 |        |                 | 74024969 | 74029256        |
|                 | rs4944920         | Imputed          | 0.011           |        |                 | 74025082 |                 |
|                 |                   |                  |                 | c-Jun  | ENCODE ChIP-seq | 1000     | 74025218        |
|                 |                   |                  |                 | JunD   | ENCODE ChIP-seq | 569      | 74025265        |
|                 |                   |                  |                 | c-Fos  | ENCODE ChIP-seq | 386      | 74025373        |
|                 |                   |                  |                 | Max    | ENCODE ChIP-seq | 578      | 74025392        |
|                 |                   |                  |                 | BAF170 | ENCODE ChIP-seq | 464      | 74025399        |
| DNaseI HS       |                   |                  |                 | ENCODE | 1000            | 74025420 | 74025690        |
|                 |                   |                  |                 | BAF155 | ENCODE ChIP-seq | 405      | 74025425        |
|                 |                   |                  |                 | c-Myc  | ENCODE ChIP-seq | 179      | 74025452        |
|                 | rs7112384         | Imputed          | 0.068           |        |                 | 74025853 |                 |
|                 | rs7112664         | Imputed          | 0.068           |        |                 | 74026074 |                 |
|                 | rs7936385         | Imputed          | 1.59E-03        |        |                 | 74027598 |                 |
| DNaseI HS       |                   |                  |                 | ENCODE | 237             | 74027680 | 74027870        |
|                 |                   |                  |                 | Input  | ENCODE ChIP-seq | 293      | 74027778        |
|                 |                   |                  |                 | CUTL1  | TransFac        | 2.32     | 74028066        |
|                 |                   |                  |                 | Oct-B1 | TransFac        | 2.32     | 74028147        |
| DNaseI HS       |                   |                  |                 | ENCODE | 101             | 74028240 | 74028570        |
|                 |                   |                  |                 | c-Jun  | ENCODE ChIP-seq | 217      | 74028384        |
|                 | rs7925747         | Genotyped        | 0.012           |        |                 | 74028431 |                 |
|                 | rs7925341         | Imputed          | 9.97E-03        |        |                 | 74028736 |                 |
|                 | <b>rs12282266</b> | <b>Imputed</b>   | <b>8.52E-07</b> |        |                 |          | <b>74028972</b> |
|                 |                   |                  |                 | AP-1   | TransFac        | 1.95     | 74029254        |
| POLD3 Exon 12   |                   |                  |                 |        |                 | 74029257 | 74031413        |
|                 |                   |                  |                 | CUTL1  | TransFac        | 2.4      | 74029428        |
|                 |                   |                  |                 | ZIC2   | TransFac        | 2.18     | 74029487        |
|                 |                   |                  |                 | Pax-2  | TransFac        | 1.72     | 74029489        |
|                 |                   |                  |                 | c-Jun  | ENCODE ChIP-seq | 480      | 74029621        |
|                 | rs1051058         | Imputed          | 0.038           |        |                 | 74029849 |                 |

|           |            |           |          |        |                 |          |          |          |
|-----------|------------|-----------|----------|--------|-----------------|----------|----------|----------|
| DNaseI HS |            |           |          | ENCODE | 712             | 74030040 | 74030330 |          |
|           |            |           |          | Brg1   | ENCODE ChIP-seq | 1000     | 74030082 | 74030465 |
|           |            |           |          | c-Jun  | ENCODE ChIP-seq | 250      | 74030124 | 74030426 |
| DNaseI HS |            |           |          | ENCODE | 533             | 74030700 | 74030930 |          |
|           |            |           |          | CREB   | TransFac        | 2.28     | 74030919 | 74030934 |
|           |            |           |          | Bach1  | TransFac        | 2.36     | 74030922 | 74030937 |
|           | rs10857    | Imputed   | 9.91E-03 |        |                 |          | 74031019 |          |
|           | rs1051076  | Genotyped | 0.435    |        |                 |          | 74031247 |          |
| DNaseI HS |            |           |          | ENCODE | 175             | 74031380 | 74031590 |          |
|           |            |           |          | c-Jun  | ENCODE ChIP-seq | 329      | 74031685 | 74032027 |
| DNaseI HS |            |           |          | ENCODE | 62              | 74031760 | 74031937 |          |
|           | rs7947345  | Imputed   | 9.80E-03 |        |                 |          | 74031875 |          |
|           | rs59958321 | Imputed   | 0.052    |        |                 |          | 74032262 |          |
|           | rs7935057  | Imputed   | 9.52E-03 |        |                 |          | 74032431 |          |
|           |            |           |          | SRF    | TransFac        | 2.84     | 74032881 | 74032896 |
|           |            |           |          | CUTL1  | TransFac        | 2.36     | 74032935 | 74032947 |
|           |            |           |          | FOXC1  | TransFac        | 1.85     | 74033117 | 74033133 |
|           |            |           |          | p300   | TransFac        | 1.66     | 74033199 | 74033213 |
|           |            |           |          | LCR-F1 | TransFac        | 2.43     | 74033208 | 74033221 |
|           |            |           |          | ZIC2   | TransFac        | 1.85     | 74033270 | 74033279 |
|           |            |           |          | ZIC2   | TransFac        | 2.33     | 74033284 | 74033293 |
|           |            |           |          | E47    | TransFac        | 1.89     | 74033440 | 74033456 |
|           | rs7123887  | Imputed   | 0.033    |        |                 |          | 74033737 |          |
|           | rs4944921  | Imputed   | 9.30E-03 |        |                 |          | 74034319 |          |
|           | rs4944922  | Imputed   | 0.033    |        |                 |          | 74034353 |          |
|           | rs4944923  | Imputed   | 0.033    |        |                 |          | 74034638 |          |
|           | rs12293317 | Imputed   | 0.105    |        |                 |          | 74034815 |          |
| DNaseI HS |            |           |          | ENCODE | 168             | 74034980 | 74035130 |          |
|           | rs10793095 | Imputed   | 0.010    |        |                 |          | 74035012 |          |
|           | rs1893362  | Imputed   | 9.21E-03 |        |                 |          | 74035085 |          |
|           | rs11236183 | Genotyped | 0.414    |        |                 |          | 74035494 |          |
| DNaseI HS |            |           |          | ENCODE | 75              | 74035740 | 74035910 |          |
|           | rs7102084  | Imputed   | 0.108    |        |                 |          | 74035780 |          |
|           | rs4944054  | Imputed   | 9.11E-03 |        |                 |          | 74036143 |          |
| DNaseI HS |            |           |          | ENCODE | 264             | 74036200 | 74036410 |          |
|           | rs10899024 | Imputed   | 6.81E-05 |        |                 |          | 74036330 |          |
|           | rs7130195  | Imputed   | 0.034    |        |                 |          | 74036574 |          |
|           | rs61900635 | Imputed   | 0.111    |        |                 |          | 74036658 |          |
|           | rs7106219  | Imputed   | 0.034    |        |                 |          | 74036714 |          |
|           | rs4944924  | Imputed   | 9.05E-03 |        |                 |          | 74036946 |          |
|           |            |           |          | PU.1   | ENCODE ChIP-seq | 95       | 74037088 | 74037275 |
|           | rs4944925  | Genotyped | 0.052    |        |                 |          | 74037177 |          |
|           | rs61900636 | Imputed   | 0.113    |        |                 |          | 74037239 |          |
|           | rs4944926  | Imputed   | 8.71E-03 |        |                 |          | 74037464 |          |
|           | rs7932922  | Genotyped | 3.64E-03 |        |                 |          | 74037678 |          |
| DNaseI HS |            |           |          | ENCODE | 374             | 74037720 | 74037890 |          |
|           |            |           |          | CREB   | TransFac        | 2.32     | 74037876 | 74037891 |
|           | rs7110993  | Imputed   | 8.68E-03 |        |                 |          | 74037961 |          |
|           |            |           |          | Nrf1   | ENCODE ChIP-seq | 635      | 74038002 | 74038122 |
|           | rs7122810  | Imputed   | 8.88E-03 |        |                 |          | 74038283 |          |
|           |            |           |          | FOXL1  | TransFac        | 1.78     | 74039225 | 74039241 |
| DNaseI HS |            |           |          | ENCODE | 1000            | 74039260 | 74039470 |          |
|           | rs1944933  | Imputed   | 1.19E-06 |        |                 |          | 74039262 |          |
|           |            |           |          | ITF-2  | TransFac        | 1.77     | 74039308 | 74039324 |
|           |            |           |          | AML1a  | TransFac        | 1.68     | 74039352 | 74039358 |
|           |            |           |          | POU2F1 | TransFac        | 1.69     | 74039353 | 74039368 |
|           |            |           |          | ARP-1  | TransFac        | 1.7      | 74039464 | 74039480 |
|           |            |           |          | p53    | TransFac        | 2        | 74039518 | 74039528 |



|           |            |           |          |        |                 |      |          |          |
|-----------|------------|-----------|----------|--------|-----------------|------|----------|----------|
|           |            |           |          | AP-4   | TransFac        | 1.81 | 74039605 | 74039615 |
|           |            |           |          | FOX11  | TransFac        | 1.97 | 74039634 | 74039647 |
|           |            |           |          | FOXD1  | TransFac        | 3.29 | 74039637 | 74039653 |
|           |            |           |          | FOXC1  | TransFac        | 4.02 | 74039637 | 74039653 |
|           |            |           |          | FOXF2  | TransFac        | 2.77 | 74039637 | 74039653 |
|           |            |           |          | FOXO4  | TransFac        | 2.77 | 74039637 | 74039651 |
|           |            |           |          | NRSF   | ENCODE ChIP-seq | 93   | 74039732 | 74039965 |
|           | rs11236185 | Imputed   | 1.01E-06 |        |                 |      | 74040179 |          |
|           | rs11236186 | Imputed   | 0.116    |        |                 |      | 74040513 |          |
|           | rs4145954  | Imputed   | 6.58E-05 |        |                 |      | 74040814 |          |
|           | rs7934301  | Imputed   | 0.033    |        |                 |      | 74041304 |          |
|           |            |           |          | IRF-7A | TransFac        | 2.19 | 74041350 | 74041368 |
|           | rs7924706  | Imputed   | 6.53E-05 |        |                 |      | 74041492 |          |
|           | rs7935426  | Imputed   | 0.116    |        |                 |      | 74041538 |          |
|           |            |           |          | NRSF   | TransFac        | 2.25 | 74041665 | 74041686 |
|           | rs7924701  | Imputed   | 8.65E-03 |        |                 |      | 74041686 |          |
|           |            |           |          | POU2F1 | TransFac        | 2.41 | 74041782 | 74041797 |
|           |            |           |          | FOXO4  | TransFac        | 1.85 | 74041803 | 74041817 |
|           | rs7937753  | Imputed   | 1.08E-04 |        |                 |      | 74041841 |          |
|           |            |           |          | CUTL1  | TransFac        | 2.07 | 74041920 | 74041935 |
|           | rs6592578  | Imputed   | 0.033    |        |                 |      | 74042175 |          |
|           | rs11236187 | Imputed   | 1.09E-06 |        |                 |      | 74042214 |          |
|           | rs11236188 | Imputed   | 1.09E-06 |        |                 |      | 74042378 |          |
|           | rs11236189 | Imputed   | 1.15E-06 |        |                 |      | 74042418 |          |
|           | rs55719957 | Imputed   | 0.097    |        |                 |      | 74042693 |          |
|           | rs12421418 | Imputed   | 8.54E-03 |        |                 |      | 74042787 |          |
|           | rs12421429 | Imputed   | 1.14E-06 |        |                 |      | 74042833 |          |
|           | rs9651760  | Imputed   | 8.50E-03 |        |                 |      | 74043104 |          |
|           | rs9651761  | Imputed   | 8.48E-03 |        |                 |      | 74043242 |          |
|           | rs9651762  | Imputed   | 8.41E-03 |        |                 |      | 74043298 |          |
|           | rs9651763  | Imputed   | 8.40E-03 |        |                 |      | 74043387 |          |
| DNaseI HS |            |           |          | ENCODE |                 | 152  | 74043780 | 74043970 |
|           | rs6592579  | Genotyped | 8.13E-03 |        |                 |      | 74043979 |          |
|           | rs6592580  | Imputed   | 8.35E-03 |        |                 |      | 74044106 |          |
|           | rs6592581  | Imputed   | 8.33E-03 |        |                 |      | 74044162 |          |
|           | rs11236190 | Imputed   | 0.145    |        |                 |      | 74044403 |          |
|           | rs6592582  | Imputed   | 9.92E-03 |        |                 |      | 74044512 |          |
|           | rs7933770  | Imputed   | 9.94E-03 |        |                 |      | 74044557 |          |
|           | rs10899025 | Imputed   | 0.134    |        |                 |      | 74044688 |          |
|           | rs7118314  | Imputed   | 0.010    |        |                 |      | 74044932 |          |
|           | rs7128944  | Imputed   | 0.010    |        |                 |      | 74045032 |          |
|           | rs7128944  | Imputed   | 6.55E-05 |        |                 |      | 74045033 |          |
|           | rs58659624 | Imputed   | 1.53E-06 |        |                 |      | 74045135 |          |
|           | rs4944928  | Imputed   | 0.010    |        |                 |      | 74045235 |          |
|           | rs12271394 | Imputed   | 0.010    |        |                 |      | 74045291 |          |
|           | rs4944055  | Imputed   | 0.011    |        |                 |      | 74045814 |          |
|           | rs4944929  | Imputed   | 0.011    |        |                 |      | 74045853 |          |
|           | rs11236193 | Imputed   | 0.140    |        |                 |      | 74045955 |          |
|           | rs4944056  | Imputed   | 0.011    |        |                 |      | 74046068 |          |
|           | rs4944930  | Imputed   | 0.011    |        |                 |      | 74046089 |          |
|           | rs61621556 | Imputed   | 0.028    |        |                 |      | 74046227 |          |
|           | rs7103197  | Imputed   | 6.70E-05 |        |                 |      | 74046364 |          |
|           | rs7126573  | Imputed   | 0.011    |        |                 |      | 74046743 |          |
|           | rs10793098 | Imputed   | 6.79E-05 |        |                 |      | 74046995 |          |
|           | rs7106984  | Imputed   | 0.011    |        |                 |      | 74047007 |          |
|           | rs7126957  | Imputed   | 0.011    |        |                 |      | 74047023 |          |
|           | rs7127628  | Imputed   | 0.011    |        |                 |      | 74047532 |          |
|           | rs7127630  | Imputed   | 0.011    |        |                 |      | 74047537 |          |

|           |            |           |          |        |                 |      |  |          |          |
|-----------|------------|-----------|----------|--------|-----------------|------|--|----------|----------|
|           | rs7127668  | Imputed   | 0.011    |        |                 |      |  | 74047800 |          |
|           | rs7111251  | Imputed   | 0.012    |        |                 |      |  | 74047839 |          |
|           | rs7111273  | Imputed   | 0.012    |        |                 |      |  | 74047879 |          |
|           | rs7131096  | Imputed   | 0.012    |        |                 |      |  | 74048004 |          |
|           | rs4514450  | Imputed   | 0.012    |        |                 |      |  | 74048604 |          |
|           | rs4644667  | Imputed   | 7.01E-05 |        |                 |      |  | 74048721 |          |
|           | rs4944931  | Imputed   | 0.012    |        |                 |      |  | 74049014 |          |
|           | rs11236198 | Imputed   | 0.201    |        |                 |      |  | 74049848 |          |
| DNaseI HS |            |           |          |        | ENCODE          | 168  |  | 74049940 | 74050110 |
|           | rs11236199 | Imputed   | 2.29E-06 |        |                 |      |  | 74050133 |          |
|           |            |           |          | USF-1  | ENCODE ChIP-seq | 223  |  | 74051271 | 74051480 |
|           |            |           |          | HNF4A  | ENCODE ChIP-seq | 813  |  | 74051446 | 74051817 |
|           |            |           |          | p300   | ENCODE ChIP-seq | 120  |  | 74051524 | 74051741 |
| DNaseI HS |            |           |          |        | ENCODE          | 68   |  | 74051540 | 74051730 |
|           |            |           |          | ERRA   | ENCODE ChIP-seq | 783  |  | 74051545 | 74051816 |
|           | rs11236200 | Imputed   | 2.60E-06 |        |                 |      |  | 74051588 |          |
|           | rs10219168 | Imputed   | 0.122    |        |                 |      |  | 74052101 |          |
|           | rs4420291  | Imputed   | 2.83E-06 |        |                 |      |  | 74052598 |          |
|           | rs2186955  | Imputed   | 0.110    |        |                 |      |  | 74053123 |          |
|           | rs11236201 | Imputed   | 0.177    |        |                 |      |  | 74053281 |          |
|           | rs7948555  | Imputed   | 0.023    |        |                 |      |  | 74053444 |          |
|           | rs11236202 | Imputed   | 0.183    |        |                 |      |  | 74054340 |          |
| DNaseI HS |            |           |          |        | ENCODE          | 154  |  | 74054880 | 74055030 |
|           | rs10899029 | Imputed   | 3.59E-06 |        |                 |      |  | 74055159 |          |
|           | rs10793101 | Imputed   | 2.45E-05 |        |                 |      |  | 74055394 |          |
|           | rs11236203 | Genotyped | 1.04E-03 |        |                 |      |  | 74055648 |          |
|           | rs4944057  | Imputed   | 0.011    |        |                 |      |  | 74056151 |          |
|           | rs11825804 | Imputed   | 3.88E-06 |        |                 |      |  | 74056519 |          |
|           | rs7109940  | Imputed   | 3.89E-06 |        |                 |      |  | 74056548 |          |
|           | rs4944933  | Imputed   | 0.094    |        |                 |      |  | 74056594 |          |
|           |            |           |          | PU.1   | ENCODE ChIP-seq | 78   |  | 74056773 | 74056895 |
|           | rs1944935  | Imputed   | 0.016    |        |                 |      |  | 74056814 |          |
|           |            |           |          | E47    | TransFac        | 1.78 |  | 74058089 | 74058105 |
|           |            |           |          | Lmo2   | TransFac        | 1.65 |  | 74058091 | 74058103 |
|           |            |           |          | AREB6  | TransFac        | 2.1  |  | 74058091 | 74058103 |
|           |            |           |          | MyoD   | TransFac        | 1.7  |  | 74058092 | 74058102 |
|           |            |           |          | c-Myb  | TransFac        | 3.21 |  | 74058145 | 74058155 |
|           |            |           |          | SRF    | TransFac        | 1.75 |  | 74058350 | 74058365 |
|           | rs6592590  | Genotyped | 3.86E-05 |        |                 |      |  | 74058677 |          |
|           | rs7129492  | Imputed   | 3.65E-05 |        |                 |      |  | 74058829 |          |
|           |            |           |          | BAF155 | ENCODE ChIP-seq | 632  |  | 74060407 | 74060857 |
|           |            |           |          | Ini1   | ENCODE ChIP-seq | 458  |  | 74060450 | 74060526 |
| DNaseI HS |            |           |          |        | ENCODE          | 645  |  | 74060500 | 74060770 |
|           | rs7933422  | Imputed   | 7.65E-03 |        |                 |      |  | 74060954 |          |
| DNaseI HS |            |           |          |        | ENCODE          | 80   |  | 74061120 | 74061330 |
|           |            |           |          | BAF155 | ENCODE ChIP-seq | 752  |  | 74061293 | 74062255 |
|           |            |           |          | c-Myc  | ENCODE ChIP-seq | 255  |  | 74061351 | 74062396 |
|           |            |           |          | Ini1   | ENCODE ChIP-seq | 191  |  | 74061390 | 74062244 |
|           |            |           |          | BAF170 | ENCODE ChIP-seq | 623  |  | 74061390 | 74062314 |
|           |            |           |          | USF-1  | ENCODE ChIP-seq | 84   |  | 74061437 | 74062359 |
|           |            |           |          | GR     | ENCODE ChIP-seq | 916  |  | 74061480 | 74062381 |
|           |            |           |          | JunD   | ENCODE ChIP-seq | 206  |  | 74061504 | 74062392 |
|           |            |           |          | c-Jun  | ENCODE ChIP-seq | 161  |  | 74061556 | 74062388 |
|           |            |           |          | NRSF   | ENCODE ChIP-seq | 127  |  | 74061572 | 74061938 |
|           |            |           |          | TAF1   | ENCODE ChIP-seq | 64   |  | 74061591 | 74061997 |
|           | rs11236207 | Imputed   | 7.47E-03 |        |                 |      |  | 74061645 |          |
| DNaseI HS |            |           |          |        | ENCODE          | 1000 |  | 74061680 | 74061950 |
|           |            |           |          | AP-4   | TransFac        | 1.87 |  | 74061714 | 74061724 |

|           |            |           |          |        |                 |      |          |          |
|-----------|------------|-----------|----------|--------|-----------------|------|----------|----------|
| DNaseI HS |            |           |          |        | ENCODE          | 1000 | 74061960 | 74062330 |
|           |            |           |          | Brg1   | ENCODE ChIP-seq | 623  | 74062123 | 74062456 |
| DNaseI HS |            |           |          |        | ENCODE          | 581  | 74062340 | 74062610 |
|           | rs10793102 | Imputed   | 0.206    |        |                 |      | 74062485 |          |
| DNaseI HS |            |           |          |        | ENCODE          | 317  | 74062660 | 74062910 |
|           |            |           |          | BAF155 | ENCODE ChIP-seq | 688  | 74062695 | 74062888 |
|           | rs11822234 | Imputed   | 2.04E-05 |        |                 |      | 74062794 |          |
|           | rs11602237 | Imputed   | 1.95E-05 |        |                 |      | 74063339 |          |
|           |            |           |          | PU.1   | ENCODE ChIP-seq | 154  | 74063350 | 74063581 |
|           | rs7104259  | Genotyped | 0.015    |        |                 |      | 74064089 |          |
|           | rs7104675  | Imputed   | 9.61E-03 |        |                 |      | 74064380 |          |
|           | rs7104802  | Genotyped | 3.66E-03 |        |                 |      | 74064448 |          |
| DNaseI HS |            |           |          |        | ENCODE          | 471  | 74064580 | 74064730 |
|           | rs7127935  | Imputed   | 6.92E-03 |        |                 |      | 74064655 |          |
| DNaseI HS |            |           |          |        | ENCODE          | 387  | 74065840 | 74066170 |
|           |            |           |          | ARP-1  | TransFac        | 1.97 | 74066489 | 74066505 |
| DNaseI HS |            |           |          |        | ENCODE          | 95   | 74067040 | 74067190 |
|           |            |           |          | Ini1   | ENCODE ChIP-seq | 244  | 74067226 | 74067389 |
|           | rs7940591  | Imputed   | 5.78E-03 |        |                 |      | 74067530 |          |
|           |            |           |          | c-Jun  | ENCODE ChIP-seq | 264  | 74067531 | 74067852 |
| DNaseI HS |            |           |          |        | ENCODE          | 267  | 74067620 | 74067850 |
|           | rs11236208 | Imputed   | 0.020    |        |                 |      | 74067969 |          |
| DNaseI HS |            |           |          |        | ENCODE          | 175  | 74068840 | 74069050 |
|           |            |           |          | Ini1   | ENCODE ChIP-seq | 203  | 74068891 | 74069051 |
|           |            |           |          | Arnt   | TransFac        | 1.85 | 74069611 | 74069631 |
| DNaseI HS |            |           |          |        | ENCODE          | 53   | 74069880 | 74070030 |
|           |            |           |          | p53    | TransFac        | 1.67 | 74070525 | 74070535 |
| DNaseI HS |            |           |          |        | ENCODE          | 127  | 74070560 | 74070710 |
|           |            |           |          | c-Myc  | TransFac        | 1.86 | 74070594 | 74070614 |
|           |            |           |          | RSRFC4 | TransFac        | 3.51 | 74070621 | 74070637 |
|           |            |           |          | POU3F2 | TransFac        | 2.06 | 74070652 | 74070662 |
| DNaseI HS |            |           |          |        | ENCODE          | 95   | 74071080 | 74071330 |
|           | rs12282262 | Genotyped | 0.013    |        |                 |      | 74071586 |          |
|           | rs10793104 | Genotyped | 0.530    |        |                 |      | 74071663 |          |

## Xp22.2 (rs5934683)

| Gene region   | rsID      | SNP type  | P-value  | TF type | Source          | Score | Chrom start | Chrom end |
|---------------|-----------|-----------|----------|---------|-----------------|-------|-------------|-----------|
|               | rs2521579 | Genotyped | 0.991    |         |                 |       | 9493090     |           |
|               |           |           |          | FAC1    | TransFac        | 1.83  | 9493996     | 9494010   |
| TBL1X Exon 18 |           |           |          | HSF1    | TransFac        | 2.36  | 9494556     | 9494566   |
|               |           |           |          | E4BP4   | TransFac        | 2.29  | 9494941     | 9494953   |
|               |           |           |          | ATF-2   | TransFac        | 2.31  | 9494943     | 9494951   |
|               |           |           |          | Nkx3-1  | TransFac        | 2.03  | 9494989     | 9495001   |
|               |           |           |          | Ini1    | ENCODE ChIP-seq | 274   | 9495217     | 9495357   |
|               |           |           |          | POU2F1  | TransFac        | 1.89  | 9496114     | 9496126   |
|               |           |           |          | ZID     | TransFac        | 3.28  | 9497097     | 9497110   |
|               |           |           |          | FAC1    | TransFac        | 2.21  | 9497431     | 9497445   |
|               | rs5979160 | Imputed   | 0.012    |         |                 |       | 9499933     |           |
|               |           |           |          | FOSL2   | ENCODE ChIP-seq | 211   | 9500429     | 9501193   |
|               | rs734253  | Genotyped | 3.83E-03 |         |                 |       | 9501246     |           |
| DNaseI HS     |           |           |          |         | ENCODE          | 307   | 9502576     | 9502806   |

|                 |           |           |       |          |                 |      |  |         |         |
|-----------------|-----------|-----------|-------|----------|-----------------|------|--|---------|---------|
|                 | rs1476510 | Genotyped | 0.010 |          |                 |      |  | 9503079 |         |
| GPR143 Exon 9   |           |           |       |          |                 |      |  | 9503190 | 9503616 |
|                 | rs3044    | Genotyped | 0.013 |          |                 |      |  | 9503388 |         |
| GPR143 Intron 8 |           |           |       |          |                 |      |  | 9503617 | 9517260 |
|                 |           |           |       | FOSL2    | ENCODE ChIP-seq | 170  |  | 9503876 | 9504268 |
|                 | rs6640489 | Imputed   | 0.013 |          |                 |      |  | 9504607 |         |
|                 |           |           |       | FOSL2    | ENCODE ChIP-seq | 920  |  | 9504930 | 9506097 |
|                 |           |           |       | PU.1     | ENCODE ChIP-seq | 1000 |  | 9505984 | 9506393 |
|                 | rs5978337 | Imputed   | 0.013 |          |                 |      |  | 9506038 |         |
|                 | rs5979162 | Imputed   | 0.013 |          |                 |      |  | 9506243 |         |
|                 |           |           |       | USF-1    | ENCODE ChIP-seq | 236  |  | 9507026 | 9507200 |
| DNaseI HS       |           |           |       |          | ENCODE          | 112  |  | 9507096 | 9507306 |
| DNaseI HS       |           |           |       |          | ENCODE          | 152  |  | 9507336 | 9507586 |
| DNaseI HS       |           |           |       |          | ENCODE          | 206  |  | 9507816 | 9507966 |
| DNaseI HS       |           |           |       |          | ENCODE          | 125  |  | 9508116 | 9508306 |
|                 |           |           |       | EBF      | ENCODE ChIP-seq | 54   |  | 9508520 | 9508867 |
|                 |           |           |       | PAX5-N19 | ENCODE ChIP-seq | 106  |  | 9508543 | 9508819 |
|                 |           |           |       | BCL11A   | ENCODE ChIP-seq | 181  |  | 9508550 | 9508870 |
|                 |           |           |       | BATF     | ENCODE ChIP-seq | 425  |  | 9508591 | 9508878 |
|                 |           |           |       | NFKB     | ENCODE ChIP-seq | 339  |  | 9508610 | 9508782 |
| DNaseI HS       |           |           |       |          | ENCODE          | 546  |  | 9508616 | 9508850 |
|                 | rs6530311 | Genotyped | 0.015 |          |                 |      |  | 9509233 |         |
|                 |           |           |       | POU2F1   | TransFac        | 3.29 |  | 9514099 | 9514111 |
|                 |           |           |       | Oct-B1   | TransFac        | 2.44 |  | 9514100 | 9514113 |
| DNaseI HS       |           |           |       |          | ENCODE          | 137  |  | 9515216 | 9515426 |
| DNaseI HS       |           |           |       |          | ENCODE          | 106  |  | 9515956 | 9516186 |
|                 | rs6654731 | Genotyped | 0.717 |          |                 |      |  | 9516373 |         |
|                 |           |           |       | PU.1     | ENCODE ChIP-seq | 1000 |  | 9516943 | 9517222 |
|                 |           |           |       | Pbx3     | ENCODE ChIP-seq | 491  |  | 9516959 | 9517187 |
|                 |           |           |       | BCL11A   | ENCODE ChIP-seq | 338  |  | 9516972 | 9517208 |
| GPR143 Exon 8   |           |           |       |          |                 |      |  | 9517261 | 9517495 |
|                 |           |           |       | AP-2rep  | TransFac        | 2.23 |  | 9517392 | 9517399 |
|                 |           |           |       | SRF      | TransFac        | 3.07 |  | 9517444 | 9517459 |
| GPR143 Intron 7 |           |           |       |          |                 |      |  | 9517496 | 9519113 |
| GPR143 Exon 7   |           |           |       |          |                 |      |  | 9519114 | 9519231 |
|                 |           |           |       | c-Myc    | TransFac        | 2.29 |  | 9519119 | 9519133 |
|                 |           |           |       | AML1a    | TransFac        | 1.68 |  | 9519125 | 9519131 |
|                 |           |           |       | CUTL1    | TransFac        | 1.92 |  | 9519206 | 9519221 |
| GPR143 Intron 6 |           |           |       |          |                 |      |  | 9519232 | 9521340 |
| GPR143 Exon 6   |           |           |       |          |                 |      |  | 9521341 | 9521449 |
| GPR143 Intron 5 |           |           |       |          |                 |      |  | 9521450 | 9523819 |
|                 |           |           |       | Cart-1   | TransFac        | 2.06 |  | 9523181 | 9523199 |
|                 |           |           |       | c-Jun    | ENCODE ChIP-seq | 444  |  | 9523207 | 9523574 |
|                 |           |           |       | PU.1     | ENCODE ChIP-seq | 219  |  | 9523238 | 9523542 |
| DNaseI HS       |           |           |       |          | ENCODE          | 649  |  | 9523296 | 9523546 |
|                 |           |           |       | GABP     | ENCODE ChIP-seq | 45   |  | 9523301 | 9523445 |
| DNaseI HS       |           |           |       |          | ENCODE          | 179  |  | 9523596 | 9523746 |
|                 |           |           |       | GR       | ENCODE ChIP-seq | 48   |  | 9523613 | 9523926 |
|                 |           |           |       | c-Jun    | ENCODE ChIP-seq | 328  |  | 9523819 | 9524657 |
| GPR143 Exon 5   |           |           |       |          |                 |      |  | 9523820 | 9523929 |
|                 |           |           |       | CREB     | TransFac        | 2.13 |  | 9523886 | 9523898 |
|                 |           |           |       | GATA-2   | TransFac        | 1.79 |  | 9523898 | 9523908 |
|                 |           |           |       | XBP-1    | TransFac        | 1.95 |  | 9523899 | 9523916 |
|                 |           |           |       | PU.1     | ENCODE ChIP-seq | 315  |  | 9523912 | 9524106 |

|                 |                  |                  |                 |         |                 |         |                |         |
|-----------------|------------------|------------------|-----------------|---------|-----------------|---------|----------------|---------|
| GPR143 Intron 4 |                  |                  |                 | 9523930 | 9526349         |         |                |         |
| DNaseI HS       |                  |                  |                 | ENCODE  | 576             | 9523936 | 9524286        |         |
|                 |                  |                  |                 | TBP     | TransFac        | 2.45    | 9524181        | 9524191 |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 255     | 9524356        | 9524626 |
| GPR143 Exon 4   |                  |                  |                 | 9526350 | 9526442         |         |                |         |
| GPR143 Intron 3 |                  |                  |                 | 9526443 | 9537107         |         |                |         |
| DNaseI HS       |                  |                  |                 | ENCODE  | 131             | 9527040 | 9527310        |         |
|                 |                  |                  |                 | PU.1    | ENCODE ChIP-seq | 103     | 9527835        | 9528075 |
|                 | rs6640499        | Genotyped        | 0.588           |         |                 |         | 9530781        |         |
|                 | rs6638961        | Imputed          | 0.603           |         |                 |         | 9531752        |         |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 78      | 9532016        | 9532206 |
|                 |                  |                  |                 | EBF     | ENCODE ChIP-seq | 132     | 9533232        | 9533473 |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 149     | 9533260        | 9533446 |
|                 |                  |                  |                 | NFKB    | ENCODE ChIP-seq | 152     | 9533290        | 9533506 |
|                 | rs11796366       | Genotyped        | 0.012           |         |                 |         | 9533879        |         |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 664     | 9535216        | 9535426 |
| GPR143 Exon 3   |                  |                  |                 | 9537108 | 9537202         |         |                |         |
|                 |                  |                  |                 | USF-1   | ENCODE ChIP-seq | 78      | 9537125        | 9537510 |
|                 |                  |                  |                 | RP58    | TransFac        | 2.05    | 9537183        | 9537195 |
| GPR143 Intron 2 |                  |                  |                 | 9537203 | 9538492         |         |                |         |
|                 | rs2732872        | Genotyped        | 0.511           |         |                 |         | 9538257        |         |
| GPR143 Exon 2   |                  |                  |                 | 9538493 | 9538602         |         |                |         |
| GPR143 Intron 1 |                  |                  |                 | 9538603 | 9543343         |         |                |         |
|                 |                  |                  |                 | EBF     | ENCODE ChIP-seq | 148     | 9542734        | 9542962 |
|                 |                  |                  |                 | TAF1    | ENCODE ChIP-seq | 490     | 9542793        | 9543804 |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 207     | 9542836        | 9543326 |
| GPR143 Exon 1   |                  |                  |                 | 9543344 | 9543653         |         |                |         |
|                 |                  |                  |                 | AP-4    | TransFac        | 2.19    | 9543531        | 9543541 |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 367     | 9543636        | 9543846 |
|                 |                  |                  |                 | Arnt    | TransFac        | 1.71    | 9543643        | 9543663 |
|                 |                  |                  |                 | c-Myc   | TransFac        | 2.09    | 9543762        | 9543776 |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 286     | 9545180        | 9545486 |
|                 |                  |                  |                 | HNF4A   | ENCODE ChIP-seq | 578     | 9545222        | 9545451 |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 56      | 9546096        | 9546270 |
|                 |                  |                  |                 | EBF     | ENCODE ChIP-seq | 79      | 9546112        | 9546331 |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 550     | 9548156        | 9548506 |
|                 |                  |                  |                 | c-Jun   | ENCODE ChIP-seq | 374     | 9548204        | 9548563 |
|                 | rs2521651        | Imputed          | 0.141           |         |                 |         | 9550636        |         |
|                 | rs2521655        | Genotyped        | 0.143           |         |                 |         | 9551779        |         |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 740     | 9553736        | 9554070 |
|                 |                  |                  |                 | CEBPB   | ENCODE ChIP-seq | 463     | 9558164        | 9558367 |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 61      | 9559596        | 9559826 |
|                 | rs2521661        | Genotyped        | 0.499           |         |                 |         | 9559988        |         |
|                 | <b>rs5934683</b> | <b>Genotyped</b> | <b>5.11E-04</b> |         |                 |         | <b>9561210</b> |         |
|                 |                  |                  |                 | c-Fos   | ENCODE ChIP-seq | 398     | 9562194        | 9562419 |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 72      | 9563096        | 9563530 |
|                 |                  |                  |                 | AREB6   | TransFac        | 1.82    | 9563681        | 9563693 |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 95      | 9563736        | 9563946 |
|                 |                  |                  |                 | HEY1    | ENCODE ChIP-seq | 209     | 9563946        | 9564280 |
| DNaseI HS       |                  |                  |                 |         | ENCODE          | 170     | 9563960        | 9564466 |
| SHROOM2 Exon 1  |                  |                  |                 | 9564232 | 9564486         |         |                |         |
|                 |                  |                  |                 | Egr-1   | TransFac        | 2.03    | 9564282        | 9564294 |
|                 |                  |                  |                 | Egr-2   | TransFac        | 2.17    | 9564282        | 9564294 |
|                 |                  |                  |                 | Egr-3   | TransFac        | 2.2     | 9564282        | 9564294 |

|                     |           |           |          |        |                 |      |         |         |
|---------------------|-----------|-----------|----------|--------|-----------------|------|---------|---------|
|                     |           |           |          | Pax-5  | TransFac        | 2.29 | 9564415 | 9564443 |
| SHROOM2 Intron<br>1 |           |           |          |        |                 |      | 9564487 | 9651427 |
| DNaseI HS           |           |           |          |        | ENCODE          | 95   | 9564600 | 9564846 |
|                     |           |           |          | AP-1   | TransFac        | 1.78 | 9564607 | 9564618 |
|                     |           |           |          | Arnt   | TransFac        | 1.68 | 9564607 | 9564627 |
|                     |           |           |          | MZF-1  | TransFac        | 2.98 | 9564710 | 9564723 |
|                     |           |           |          | AREB6  | TransFac        | 1.82 | 9564785 | 9564797 |
|                     |           |           |          | Lmo2   | TransFac        | 1.7  | 9564785 | 9564797 |
| DNaseI HS           |           |           |          |        | ENCODE          | 136  | 9564936 | 9565306 |
|                     | rs2521662 | Imputed   | 0.347    |        |                 |      | 9566359 |         |
|                     |           |           |          | p300   | ENCODE ChIP-seq | 123  | 9566723 | 9566997 |
|                     |           |           |          | CEBPB  | ENCODE ChIP-seq | 413  | 9566740 | 9567025 |
| DNaseI HS           |           |           |          |        | ENCODE          | 718  | 9566776 | 9566990 |
|                     |           |           |          | POU2F1 | TransFac        | 1.65 | 9566881 | 9566904 |
|                     |           |           |          | FOXC1  | TransFac        | 3.27 | 9566883 | 9566899 |
|                     | rs757018  | Genotyped | 0.355    |        |                 |      | 9567034 |         |
| DNaseI HS           |           |           |          |        | ENCODE          | 1000 | 9567416 | 9567586 |
|                     |           |           |          | AP-4   | TransFac        | 1.78 | 9567467 | 9567477 |
|                     |           |           |          | c-Myb  | TransFac        | 3.21 | 9568555 | 9568565 |
|                     | rs2040621 | Imputed   | 0.345    |        |                 |      | 9568681 |         |
| DNaseI HS           |           |           |          |        | ENCODE          | 1000 | 9568716 | 9568946 |
|                     |           |           |          | POU2F1 | TransFac        | 2.33 | 9569008 | 9569021 |
|                     |           |           |          | Oct-B1 | TransFac        | 2.53 | 9569150 | 9569163 |
| DNaseI HS           |           |           |          |        | ENCODE          | 240  | 9569316 | 9569486 |
|                     |           |           |          | AP-4   | TransFac        | 1.76 | 9573639 | 9573649 |
|                     |           |           |          | Nkx3-1 | TransFac        | 2.34 | 9575362 | 9575374 |
| DNaseI HS           |           |           |          |        | ENCODE          | 171  | 9575856 | 9576006 |
|                     |           |           |          | CEBPB  | ENCODE ChIP-seq | 496  | 9575891 | 9576124 |
|                     | rs6530318 | Genotyped | 0.020    |        |                 |      | 9575936 |         |
|                     |           |           |          | CUTL1  | TransFac        | 1.64 | 9576134 | 9576149 |
| DNaseI HS           |           |           |          |        | ENCODE          | 107  | 9576756 | 9576906 |
|                     |           |           |          | c-Myb  | TransFac        | 1.72 | 9577823 | 9577833 |
|                     |           |           |          | NRSF   | ENCODE ChIP-seq | 29   | 9579754 | 9579922 |
|                     | rs7059068 | Genotyped | 9.92E-03 |        |                 |      | 9581936 |         |
| DNaseI HS           |           |           |          |        | ENCODE          | 83   | 9582076 | 9582246 |
|                     |           |           |          | Pax-2  | TransFac        | 1.7  | 9583461 | 9583480 |
| DNaseI HS           |           |           |          |        | ENCODE          | 298  | 9583796 | 9584026 |
| DNaseI HS           |           |           |          |        | ENCODE          | 112  | 9584796 | 9584946 |
|                     |           |           |          | PU.1   | ENCODE ChIP-seq | 236  | 9585578 | 9585791 |
| DNaseI HS           |           |           |          |        | ENCODE          | 333  | 9585736 | 9585906 |
| DNaseI HS           |           |           |          |        | ENCODE          | 264  | 9587756 | 9587946 |
| DNaseI HS           |           |           |          |        | ENCODE          | 142  | 9588056 | 9588286 |
| DNaseI HS           |           |           |          |        | ENCODE          | 301  | 9594216 | 9594446 |
|                     |           |           |          | AREB6  | TransFac        | 1.64 | 9595165 | 9595174 |
|                     |           |           |          | Hlf    | TransFac        | 3.1  | 9595642 | 9595652 |
|                     |           |           |          | RP58   | TransFac        | 1.74 | 9595754 | 9595766 |
|                     |           |           |          | FOXL1  | TransFac        | 1.96 | 9595791 | 9595807 |
|                     |           |           |          | RSRFC4 | TransFac        | 2.35 | 9595791 | 9595807 |
|                     |           |           |          | TBP    | TransFac        | 1.97 | 9595795 | 9595803 |
|                     |           |           |          | ISGF-3 | TransFac        | 1.72 | 9596058 | 9596073 |
|                     |           |           |          | HSF1   | TransFac        | 1.93 | 9596134 | 9596144 |
|                     |           |           |          | CUTL1  | TransFac        | 2.38 | 9596180 | 9596195 |
|                     |           |           |          | AP-4   | TransFac        | 1.76 | 9596235 | 9596245 |
|                     |           |           |          | Pbx1a  | TransFac        | 2.37 | 9596519 | 9596534 |

|           |           |           |       |          |                 |      |         |         |
|-----------|-----------|-----------|-------|----------|-----------------|------|---------|---------|
|           |           |           |       | CUTL1    | TransFac        | 1.64 | 9596520 | 9596535 |
|           |           |           |       | Elk-1    | TransFac        | 1.66 | 9596564 | 9596580 |
|           |           |           |       | POU2F1   | TransFac        | 1.91 | 9596618 | 9596641 |
|           |           |           |       | ARP-1    | TransFac        | 2.05 | 9596654 | 9596670 |
|           |           |           |       | Pax-2    | TransFac        | 1.72 | 9596881 | 9596900 |
|           |           |           |       | p53      | TransFac        | 1.95 | 9596888 | 9596898 |
|           |           |           |       | AREB6    | TransFac        | 2.2  | 9596959 | 9596968 |
|           |           |           |       | CUTL1    | TransFac        | 1.7  | 9597043 | 9597053 |
|           |           |           |       | CREB     | TransFac        | 2.42 | 9597223 | 9597238 |
|           |           |           |       | FOXL1    | TransFac        | 2.19 | 9597328 | 9597344 |
|           |           |           |       | ATF-2    | TransFac        | 1.76 | 9597329 | 9597337 |
|           |           |           |       | FOXO4    | TransFac        | 2.13 | 9597330 | 9597344 |
|           |           |           |       | MRF-2    | TransFac        | 1.93 | 9597417 | 9597431 |
| DNaseI HS |           |           |       |          | ENCODE          | 118  | 9599036 | 9599186 |
| DNaseI HS |           |           |       |          | ENCODE          | 532  | 9602336 | 9602610 |
|           |           |           |       | MRF-2    | TransFac        | 2.56 | 9602458 | 9602472 |
| DNaseI HS |           |           |       |          | ENCODE          | 171  | 9602916 | 9603186 |
|           |           |           |       | FOXO4    | TransFac        | 1.8  | 9603308 | 9603322 |
| DNaseI HS |           |           |       |          | ENCODE          | 371  | 9603336 | 9603546 |
|           |           |           |       | RelA     | TransFac        | 1.81 | 9603407 | 9603417 |
|           |           |           |       | HNF4A    | ENCODE ChIP-seq | 233  | 9603448 | 9604073 |
|           |           |           |       | p300     | ENCODE ChIP-seq | 114  | 9603882 | 9604083 |
|           |           |           |       | Ini1     | ENCODE ChIP-seq | 230  | 9606971 | 9607151 |
|           |           |           |       | TCF12    | ENCODE ChIP-seq | 122  | 9607009 | 9607324 |
| DNaseI HS |           |           |       |          | ENCODE          | 177  | 9607120 | 9607270 |
|           |           |           |       | IRF4     | ENCODE ChIP-seq | 940  | 9607193 | 9607690 |
|           |           |           |       | NFKB     | ENCODE ChIP-seq | 172  | 9607253 | 9607719 |
|           |           |           |       | BCL11A   | ENCODE ChIP-seq | 1000 | 9607272 | 9607706 |
|           |           |           |       | p300     | ENCODE ChIP-seq | 234  | 9607275 | 9607687 |
|           |           |           |       | PAX5-N19 | ENCODE ChIP-seq | 603  | 9607291 | 9607678 |
|           |           |           |       | JunD     | ENCODE ChIP-seq | 672  | 9607308 | 9607728 |
|           |           |           |       | BATF     | ENCODE ChIP-seq | 1000 | 9607310 | 9607704 |
|           |           |           |       | EBF      | ENCODE ChIP-seq | 70   | 9607320 | 9607599 |
|           |           |           |       | TAF1     | ENCODE ChIP-seq | 90   | 9607336 | 9607669 |
|           |           |           |       | TCF12    | ENCODE ChIP-seq | 778  | 9607336 | 9607682 |
|           |           |           |       | c-Jun    | ENCODE ChIP-seq | 357  | 9607338 | 9607742 |
| DNaseI HS |           |           |       |          | ENCODE          | 1000 | 9607351 | 9607626 |
|           |           |           |       | PAX5-C20 | ENCODE ChIP-seq | 324  | 9607381 | 9607661 |
|           |           |           |       | BAF155   | ENCODE ChIP-seq | 1000 | 9607385 | 9607622 |
|           |           |           |       | POU2F2   | ENCODE ChIP-seq | 235  | 9607387 | 9607640 |
|           | rs5934697 | Imputed   | 0.427 |          |                 |      | 9608931 |         |
|           |           |           |       | Max      | ENCODE ChIP-seq | 1000 | 9610157 | 9610486 |
| DNaseI HS |           |           |       |          | ENCODE          | 41   | 9610316 | 9610426 |
| DNaseI HS |           |           |       |          | ENCODE          | 982  | 9610460 | 9610906 |
|           | rs5933761 | Imputed   | 0.456 |          |                 |      | 9613230 |         |
|           | rs2405940 | Genotyped | 0.391 |          |                 |      | 9613280 |         |
|           |           |           |       | p53      | TransFac        | 1.79 | 9613419 | 9613439 |
|           | rs1859013 | Imputed   | 0.463 |          |                 |      | 9613481 |         |
|           | rs6530329 | Imputed   | 0.491 |          |                 |      | 9614078 |         |
|           | rs6638967 | Imputed   | 0.627 |          |                 |      | 9615667 |         |
| DNaseI HS |           |           |       |          | ENCODE          | 331  | 9616136 | 9616306 |
|           | rs4830660 | Imputed   | 0.646 |          |                 |      | 9617325 |         |
|           | rs4830661 | Genotyped | 0.285 |          |                 |      | 9617429 |         |
|           | rs6530331 | Imputed   | 0.788 |          |                 |      | 9617838 |         |

|           |            |           |       |        |                 |      |         |         |
|-----------|------------|-----------|-------|--------|-----------------|------|---------|---------|
|           | rs5933762  | Genotyped | 0.668 |        |                 |      | 9618602 |         |
|           |            |           |       | FOXO1  | TransFac        | 1.87 | 9620267 | 9620283 |
|           |            |           |       | NFKB   | ENCODE ChIP-seq | 63   | 9620303 | 9620527 |
| DNaseI HS |            |           |       |        | ENCODE          | 562  | 9620336 | 9620686 |
|           | rs6530332  | Imputed   | 0.710 |        |                 |      | 9620377 |         |
|           |            |           |       | MZF-1  | TransFac        | 1.66 | 9620411 | 9620424 |
|           | rs960417   | Genotyped | 0.604 |        |                 |      | 9620831 |         |
| DNaseI HS |            |           |       |        | ENCODE          | 112  | 9622756 | 9622906 |
|           |            |           |       | HEY1   | ENCODE ChIP-seq | 47   | 9622907 | 9623250 |
| DNaseI HS |            |           |       |        | ENCODE          | 107  | 9622976 | 9623270 |
|           |            |           |       | CEBPB  | ENCODE ChIP-seq | 595  | 9623275 | 9623740 |
|           |            |           |       | p300   | ENCODE ChIP-seq | 221  | 9623298 | 9623670 |
| DNaseI HS |            |           |       |        | ENCODE          | 628  | 9623336 | 9623666 |
|           | rs2405942  | Genotyped | 0.825 |        |                 |      | 9623871 |         |
| DNaseI HS |            |           |       |        | ENCODE          | 264  | 9624416 | 9624706 |
|           | rs6530333  | Genotyped | 0.637 |        |                 |      | 9624668 |         |
|           |            |           |       | BAF155 | ENCODE ChIP-seq | 539  | 9624923 | 9625160 |
|           |            |           |       | c-Jun  | ENCODE ChIP-seq | 321  | 9624941 | 9625255 |
|           |            |           |       | CEBPB  | ENCODE ChIP-seq | 355  | 9624963 | 9625294 |
|           |            |           |       | FOSL2  | ENCODE ChIP-seq | 100  | 9624969 | 9625203 |
|           |            |           |       | JunD   | ENCODE ChIP-seq | 143  | 9624971 | 9625303 |
| DNaseI HS |            |           |       |        | ENCODE          | 1000 | 9624976 | 9625226 |
|           | rs5933763  | Imputed   | 0.580 |        |                 |      | 9625101 |         |
|           | rs5933764  | Imputed   | 0.565 |        |                 |      | 9625257 |         |
|           | rs5933765  | Genotyped | 0.538 |        |                 |      | 9625538 |         |
| DNaseI HS |            |           |       |        | ENCODE          | 141  | 9626956 | 9627166 |
|           | rs5934702  | Genotyped | 0.365 |        |                 |      | 9627249 |         |
|           | rs5934703  | Imputed   | 0.359 |        |                 |      | 9627315 |         |
|           | rs16985744 | Imputed   | 0.696 |        |                 |      | 9627419 |         |
|           | rs4830418  | Imputed   | 0.357 |        |                 |      | 9627483 |         |
|           | rs4830419  | Imputed   | 0.353 |        |                 |      | 9627715 |         |
| DNaseI HS |            |           |       |        | ENCODE          | 1000 | 9629736 | 9629966 |
|           | rs5934705  | Genotyped | 0.294 |        |                 |      | 9631135 |         |
| DNaseI HS |            |           |       |        | ENCODE          | 186  | 9631136 | 9631346 |
|           | rs12395804 | Genotyped | 0.634 |        |                 |      | 9632657 |         |
| DNaseI HS |            |           |       |        | ENCODE          | 212  | 9633356 | 9633726 |
| DNaseI HS |            |           |       |        | ENCODE          | 269  | 9633816 | 9634026 |
|           |            |           |       | Max    | ENCODE ChIP-seq | 713  | 9634448 | 9634827 |
| DNaseI HS |            |           |       |        | ENCODE          | 113  | 9634456 | 9634606 |
|           | rs7051365  | Genotyped | 0.096 |        |                 |      | 9634583 |         |
| DNaseI HS |            |           |       |        | ENCODE          | 44   | 9635456 | 9635606 |
| DNaseI HS |            |           |       |        | ENCODE          | 394  | 9635856 | 9636166 |
|           |            |           |       | GR     | ENCODE ChIP-seq | 181  | 9635934 | 9636308 |
|           |            |           |       | MZF-1  | TransFac        | 2.27 | 9635995 | 9636008 |
|           |            |           |       | c-Rel  | TransFac        | 1.82 | 9636084 | 9636094 |
|           |            |           |       | FOXO1  | TransFac        | 1.83 | 9636096 | 9636109 |
|           |            |           |       | ISGF-3 | TransFac        | 1.67 | 9636150 | 9636165 |
|           | rs7062290  | Genotyped | 0.094 |        |                 |      | 9637173 |         |
|           | rs5979192  | Imputed   | 0.107 |        |                 |      | 9637705 |         |
| DNaseI HS |            |           |       |        | ENCODE          | 334  | 9637716 | 9637906 |
|           |            |           |       | Bach1  | TransFac        | 2.5  | 9637766 | 9637781 |
| DNaseI HS |            |           |       |        | ENCODE          | 597  | 9637996 | 9638206 |
| DNaseI HS |            |           |       |        | ENCODE          | 165  | 9638676 | 9638946 |
|           |            |           |       | CTCF   | ENCODE ChIP-seq | 88   | 9640324 | 9640554 |



|                  |            |           |       |        |                 |      |         |         |
|------------------|------------|-----------|-------|--------|-----------------|------|---------|---------|
|                  |            |           |       | CEBPB  | ENCODE ChIP-seq | 948  | 9640446 | 9640612 |
|                  | rs1016625  | Genotyped | 0.047 |        |                 |      | 9640837 |         |
|                  | rs1016626  | Imputed   | 0.428 |        |                 |      | 9641162 |         |
|                  | rs1016627  | Genotyped | 0.078 |        |                 |      | 9641307 |         |
| DNaseI HS        |            |           |       |        | ENCODE          | 296  | 9641756 | 9642106 |
|                  |            |           |       | XBP-1  | TransFac        | 1.67 | 9643244 | 9643261 |
|                  |            |           |       | Ini1   | ENCODE ChIP-seq | 235  | 9643639 | 9643851 |
| DNaseI HS        |            |           |       |        | ENCODE          | 108  | 9643640 | 9643790 |
|                  |            |           |       | NFKB   | ENCODE ChIP-seq | 993  | 9643732 | 9644277 |
|                  |            |           |       | GR     | ENCODE ChIP-seq | 424  | 9643749 | 9644173 |
|                  |            |           |       | BAF155 | ENCODE ChIP-seq | 663  | 9643809 | 9644371 |
|                  |            |           |       | c-Jun  | ENCODE ChIP-seq | 354  | 9643858 | 9644367 |
|                  |            |           |       | JunD   | ENCODE ChIP-seq | 466  | 9643868 | 9644599 |
|                  |            |           |       | BAF170 | ENCODE ChIP-seq | 653  | 9643905 | 9644706 |
| DNaseI HS        |            |           |       |        | ENCODE          | 435  | 9643916 | 9644186 |
|                  |            |           |       | AP-1   | TransFac        | 1.72 | 9644019 | 9644028 |
|                  | rs5934708  | Genotyped | 0.115 |        |                 |      | 9644055 |         |
|                  |            |           |       | E47    | TransFac        | 1.76 | 9644156 | 9644172 |
|                  |            |           |       | AP-1   | TransFac        | 1.79 | 9644166 | 9644175 |
| DNaseI HS        |            |           |       |        | ENCODE          | 366  | 9644356 | 9644786 |
|                  |            |           |       | Ini1   | ENCODE ChIP-seq | 376  | 9644429 | 9644840 |
|                  |            |           |       | p300   | TransFac        | 1.82 | 9644532 | 9644546 |
|                  | rs12007032 | Imputed   | 0.353 |        |                 |      | 9644753 |         |
| DNaseI HS        |            |           |       |        | ENCODE          | 159  | 9644856 | 9645006 |
|                  | rs5979195  | Imputed   | 0.355 |        |                 |      | 9644975 |         |
|                  | rs5933773  | Imputed   | 0.399 |        |                 |      | 9645028 |         |
|                  | rs6640534  | Genotyped | 0.317 |        |                 |      | 9645430 |         |
| DNaseI HS        |            |           |       |        | ENCODE          | 471  | 9645556 | 9645886 |
|                  | rs4830665  | Imputed   | 0.195 |        |                 |      | 9645855 |         |
|                  | rs5934710  | Imputed   | 0.442 |        |                 |      | 9647321 |         |
|                  | rs723259   | Genotyped | 0.498 |        |                 |      | 9648934 |         |
| DNaseI HS        |            |           |       |        | ENCODE          | 107  | 9649136 | 9649306 |
|                  | rs723261   | Imputed   | 0.466 |        |                 |      | 9649446 |         |
|                  |            |           |       | NFKB   | ENCODE ChIP-seq | 101  | 9649584 | 9649750 |
|                  | rs5933775  | Genotyped | 0.459 |        |                 |      | 9649608 |         |
| DNaseI HS        |            |           |       |        | ENCODE          | 287  | 9649716 | 9649946 |
| DNaseI HS        |            |           |       |        | ENCODE          | 59   | 9650676 | 9650886 |
|                  | rs763375   | Imputed   | 0.484 |        |                 |      | 9651274 |         |
| SHROOM2 Exon 2   |            |           |       |        |                 |      | 9651428 | 9651579 |
|                  |            |           |       | CUTL1  | TransFac        | 1.92 | 9651487 | 9651497 |
| SHROOM2 Intron 2 |            |           |       |        |                 |      | 9651580 | 9668752 |
|                  |            |           |       | Rad21  | ENCODE ChIP-seq | 152  | 9651725 | 9652121 |
|                  |            |           |       | CTCF   | ENCODE ChIP-seq | 364  | 9651740 | 9652075 |
|                  |            |           |       | NFKB   | ENCODE ChIP-seq | 78   | 9651784 | 9652122 |
| DNaseI HS        |            |           |       |        | ENCODE          | 284  | 9651816 | 9652026 |
|                  | rs2239416  | Imputed   | 0.497 |        |                 |      | 9652249 |         |
| DNaseI HS        |            |           |       |        | ENCODE          | 516  | 9652336 | 9652586 |
| DNaseI HS        |            |           |       |        | ENCODE          | 150  | 9654096 | 9654266 |
| DNaseI HS        |            |           |       |        | ENCODE          | 333  | 9655976 | 9656206 |
|                  |            |           |       | Pbx1a  | TransFac        | 2.07 | 9656058 | 9656073 |
|                  |            |           |       | RP58   | TransFac        | 2.12 | 9656424 | 9656436 |
|                  | rs6640537  | Imputed   | 0.430 |        |                 |      | 9657453 |         |
|                  | rs17280844 | Genotyped | 0.407 |        |                 |      | 9657652 |         |
|                  |            |           |       | Elk-1  | TransFac        | 1.8  | 9658008 | 9658024 |

|                  |            |           |       |          |                 |      |         |         |
|------------------|------------|-----------|-------|----------|-----------------|------|---------|---------|
|                  | rs2051687  | Genotyped | 0.213 |          |                 |      | 9659037 |         |
|                  | rs2239427  | Imputed   | 0.472 |          |                 |      | 9659259 |         |
|                  |            |           |       | Max      | ENCODE ChIP-seq | 696  | 9660400 | 9661200 |
|                  |            |           |       | c-Jun    | ENCODE ChIP-seq | 1000 | 9660503 | 9660980 |
| DNaseI HS        |            |           |       |          | ENCODE          | 437  | 9660536 | 9660926 |
|                  | rs1858883  | Genotyped | 0.511 |          |                 |      | 9660624 |         |
|                  |            |           |       | TBP      | TransFac        | 1.92 | 9660651 | 9660661 |
| DNaseI HS        |            |           |       |          | ENCODE          | 195  | 9661896 | 9662126 |
|                  |            |           |       | IRF4     | ENCODE ChIP-seq | 496  | 9662910 | 9663280 |
|                  |            |           |       | CTCF     | ENCODE ChIP-seq | 882  | 9662948 | 9663277 |
|                  |            |           |       | Rad21    | ENCODE ChIP-seq | 249  | 9662959 | 9663389 |
|                  |            |           |       | NFKB     | ENCODE ChIP-seq | 194  | 9662995 | 9663300 |
| DNaseI HS        |            |           |       |          | ENCODE          | 514  | 9662996 | 9663226 |
|                  | rs13340753 | Genotyped | 0.505 |          |                 |      | 9663006 |         |
|                  |            |           |       | Ini1     | ENCODE ChIP-seq | 141  | 9663024 | 9663233 |
|                  | rs6640541  | Imputed   | 0.537 |          |                 |      | 9664230 |         |
|                  | rs12557532 | Genotyped | 0.819 |          |                 |      | 9664501 |         |
| DNaseI HS        |            |           |       |          | ENCODE          | 62   | 9664956 | 9665106 |
|                  | rs5979199  | Genotyped | 0.654 |          |                 |      | 9665537 |         |
|                  |            |           |       | NRSF     | ENCODE ChIP-seq | 832  | 9666720 | 9667086 |
| DNaseI HS        |            |           |       |          | ENCODE          | 310  | 9666896 | 9667086 |
|                  | rs6638974  | Imputed   | 0.575 |          |                 |      | 9666988 |         |
|                  | rs6640542  | Imputed   | 0.577 |          |                 |      | 9667110 |         |
| DNaseI HS        |            |           |       |          | ENCODE          | 101  | 9667416 | 9667606 |
| SHROOM2 Exon 3   |            |           |       |          |                 |      | 9668753 | 9668884 |
|                  |            |           |       | p300     | TransFac        | 2.6  | 9668773 | 9668787 |
| SHROOM2 Intron 3 | rs6640543  | Imputed   | 0.915 |          |                 |      | 9668834 |         |
|                  |            |           |       |          |                 |      | 9668885 | 9672133 |
|                  | rs6640544  | Genotyped | 0.929 |          |                 |      | 9668933 |         |
|                  | rs17327524 | Genotyped | 0.515 |          |                 |      | 9671845 |         |
|                  | rs17327531 | Imputed   | 0.593 |          |                 |      | 9671904 |         |
| DNaseI HS        |            |           |       |          | ENCODE          | 84   | 9671916 | 9672106 |
| SHROOM2 Exon 4   |            |           |       |          |                 |      | 9672134 | 9674474 |
|                  |            |           |       | NRSF     | TransFac        | 2.77 | 9672197 | 9672218 |
|                  |            |           |       | E47      | TransFac        | 2.17 | 9672273 | 9672289 |
|                  |            |           |       | c-Myc    | TransFac        | 1.94 | 9672274 | 9672288 |
|                  |            |           |       | ZID      | TransFac        | 1.65 | 9672313 | 9672326 |
|                  |            |           |       | AML1a    | TransFac        | 1.68 | 9672345 | 9672351 |
|                  |            |           |       | HSF1     | TransFac        | 1.87 | 9672381 | 9672391 |
|                  |            |           |       | SRF      | TransFac        | 1.76 | 9672568 | 9672582 |
|                  |            |           |       | Arnt     | TransFac        | 1.8  | 9673051 | 9673071 |
|                  |            |           |       | AREB6    | TransFac        | 2.65 | 9673055 | 9673068 |
|                  |            |           |       | SREBP-1a | TransFac        | 1.86 | 9673056 | 9673067 |
|                  |            |           |       | TBP      | TransFac        | 2.21 | 9673720 | 9673735 |
|                  |            |           |       | XBP-1    | TransFac        | 2.01 | 9673759 | 9673776 |
| SHROOM2 Intron 4 |            |           |       |          |                 |      | 9674475 | 9675965 |
| DNaseI HS        |            |           |       |          | ENCODE          | 1000 | 9675556 | 9675946 |
| SHROOM2 Exon 5   |            |           |       |          |                 |      | 9675966 | 9676066 |
| SHROOM2 Intron 5 |            |           |       |          |                 |      | 9676067 | 9709950 |
|                  | rs2073944  | Imputed   | 0.817 |          |                 |      | 9676156 |         |
| DNaseI HS        |            |           |       |          | ENCODE          | 97   | 9676476 | 9676686 |
|                  |            |           |       | Nkx6-1   | TransFac        | 2.03 | 9676544 | 9676557 |

|           |           |           |       |          |                 |      |         |         |
|-----------|-----------|-----------|-------|----------|-----------------|------|---------|---------|
|           |           |           |       | Cdc5     | TransFac        | 2.24 | 9676565 | 9676577 |
|           | rs2012821 | Genotyped | 0.360 |          |                 |      | 9676743 |         |
|           |           |           |       | IRF4     | ENCODE ChIP-seq | 763  | 9677186 | 9677753 |
|           |           |           |       | POU2F2   | ENCODE ChIP-seq | 180  | 9677274 | 9677751 |
|           |           |           |       | BCL11A   | ENCODE ChIP-seq | 902  | 9677277 | 9677666 |
|           |           |           |       | PAX5-N19 | ENCODE ChIP-seq | 130  | 9677301 | 9677708 |
|           |           |           |       | EBF      | ENCODE ChIP-seq | 158  | 9677333 | 9677881 |
|           |           |           |       | BATF     | ENCODE ChIP-seq | 309  | 9677355 | 9677620 |
|           |           |           |       | TCF12    | ENCODE ChIP-seq | 797  | 9677361 | 9677714 |
|           |           |           |       | FOXL1    | TransFac        | 2.31 | 9677369 | 9677385 |
| DNaseI HS |           |           |       |          | ENCODE          | 914  | 9677376 | 9677766 |
|           |           |           |       | E4BP4    | TransFac        | 2.03 | 9677437 | 9677449 |
|           |           |           |       | NFKB     | ENCODE ChIP-seq | 344  | 9677438 | 9677624 |
|           |           |           |       | c-Myc    | TransFac        | 2.28 | 9677563 | 9677583 |
|           |           |           |       | Arnt     | TransFac        | 2.62 | 9677563 | 9677583 |
|           |           |           |       | SREBP-1a | TransFac        | 2.19 | 9677567 | 9677578 |
|           |           |           |       | c-Myc    | TransFac        | 2.52 | 9677567 | 9677579 |
|           |           |           |       | FOXL1    | TransFac        | 1.67 | 9680386 | 9680402 |
| DNaseI HS |           |           |       |          | ENCODE          | 171  | 9680536 | 9680746 |
| DNaseI HS |           |           |       |          | ENCODE          | 94   | 9680996 | 9681246 |
|           | rs6640551 | Genotyped | 0.639 |          |                 |      | 9681893 |         |
|           | rs6640552 | Genotyped | 0.609 |          |                 |      | 9682157 |         |
| DNaseI HS |           |           |       |          | ENCODE          | 597  | 9683016 | 9683386 |
| DNaseI HS |           |           |       |          | ENCODE          | 612  | 9684776 | 9685086 |
|           |           |           |       | FOSL2    | ENCODE ChIP-seq | 117  | 9685347 | 9685592 |
|           |           |           |       | JunD     | ENCODE ChIP-seq | 244  | 9685350 | 9685626 |
|           |           |           |       | CEBPB    | ENCODE ChIP-seq | 694  | 9685351 | 9685631 |
|           |           |           |       | c-Jun    | ENCODE ChIP-seq | 271  | 9685355 | 9685660 |
| DNaseI HS |           |           |       |          | ENCODE          | 780  | 9685416 | 9685726 |
| DNaseI HS |           |           |       |          | ENCODE          | 813  | 9685756 | 9686066 |
| DNaseI HS |           |           |       |          | ENCODE          | 528  | 9689336 | 9689626 |
|           |           |           |       | c-Jun    | ENCODE ChIP-seq | 296  | 9689399 | 9689707 |
|           |           |           |       | POU3F2   | TransFac        | 2.82 | 9689532 | 9689548 |
| DNaseI HS |           |           |       |          | ENCODE          | 793  | 9690116 | 9690366 |
|           |           |           |       | RP58     | TransFac        | 2.25 | 9690235 | 9690247 |
|           |           |           |       | Sox9     | TransFac        | 1.97 | 9690561 | 9690575 |
|           |           |           |       | FOXF2    | TransFac        | 2.09 | 9690563 | 9690579 |
|           |           |           |       | FOXL1    | TransFac        | 1.68 | 9690563 | 9690579 |
|           |           |           |       | Oct-B1   | TransFac        | 2.77 | 9690596 | 9690609 |
|           |           |           |       | p53      | TransFac        | 1.82 | 9690906 | 9690916 |
| DNaseI HS |           |           |       |          | ENCODE          | 529  | 9690916 | 9691126 |
|           |           |           |       | FOXC1    | TransFac        | 1.89 | 9691403 | 9691419 |
|           |           |           |       | LCR-F1   | TransFac        | 1.74 | 9691405 | 9691427 |
|           |           |           |       | AP-1     | TransFac        | 2.34 | 9691411 | 9691424 |
|           |           |           |       | POU2F1   | TransFac        | 2.23 | 9692075 | 9692090 |
|           | rs6640557 | Genotyped | 0.826 |          |                 |      | 9692207 |         |
|           | rs2239419 | Genotyped | 0.700 |          |                 |      | 9694918 |         |
|           | rs2239420 | Imputed   | 0.677 |          |                 |      | 9695076 |         |
|           | rs2239421 | Genotyped | 0.345 |          |                 |      | 9695788 |         |
|           | rs2239422 | Genotyped | 0.398 |          |                 |      | 9695900 |         |
|           | rs2239423 | Imputed   | 0.352 |          |                 |      | 9695930 |         |
|           |           |           |       | BCL3     | ENCODE ChIP-seq | 173  | 9697480 | 9697840 |
| DNaseI HS |           |           |       |          | ENCODE          | 54   | 9697556 | 9697746 |
| DNaseI HS |           |           |       |          | ENCODE          | 165  | 9697776 | 9697926 |

|                  |           |           |       |          |                 |         |         |         |
|------------------|-----------|-----------|-------|----------|-----------------|---------|---------|---------|
| DNaseI HS        |           |           |       | ENCODE   | 189             | 9698856 | 9699026 |         |
|                  |           |           |       | CEBPB    | ENCODE ChIP-seq | 1000    | 9699190 | 9699552 |
|                  |           |           |       | HNF4A    | ENCODE ChIP-seq | 686     | 9699231 | 9699486 |
|                  | rs2283703 | Genotyped | 0.523 |          |                 |         | 9700159 |         |
| DNaseI HS        |           |           |       | ENCODE   | 86              | 9701216 | 9701426 |         |
|                  |           |           |       | AML1a    | TransFac        | 1.68    | 9701666 | 9701672 |
|                  |           |           |       | c-Myc    | TransFac        | 2.21    | 9701831 | 9701843 |
| DNaseI HS        |           |           |       | ENCODE   | 99              | 9703916 | 9704086 |         |
| DNaseI HS        |           |           |       | ENCODE   | 195             | 9705496 | 9705746 |         |
|                  |           |           |       | TAF1     | ENCODE ChIP-seq | 35      | 9705871 | 9706089 |
| DNaseI HS        |           |           |       | ENCODE   | 1000            | 9705996 | 9706226 |         |
|                  |           |           |       | Nkx6-1   | TransFac        | 2.08    | 9706894 | 9706907 |
|                  |           |           |       | FOXO4    | TransFac        | 1.87    | 9708946 | 9708957 |
| SHROOM2 Exon 6   |           |           |       |          |                 |         | 9709951 | 9710646 |
|                  |           |           |       | NRSF     | ENCODE ChIP-seq | 52      | 9710472 | 9710647 |
| SHROOM2 Intron 6 |           |           |       |          |                 |         | 9710647 | 9714909 |
| DNaseI HS        |           |           |       | ENCODE   | 171             | 9711196 | 9711346 |         |
|                  | rs2238886 | Genotyped | 0.989 |          |                 |         | 9712112 |         |
| DNaseI HS        |           |           |       | ENCODE   | 65              | 9713276 | 9713446 |         |
|                  |           |           |       | CUTL1    | TransFac        | 1.88    | 9714909 | 9714919 |
| SHROOM2 Exon 7   |           |           |       |          |                 |         | 9714910 | 9715461 |
|                  |           |           |       | MRF-2    | TransFac        | 2.09    | 9714925 | 9714939 |
|                  |           |           |       | FAC1     | TransFac        | 1.76    | 9714925 | 9714939 |
|                  |           |           |       | ZID      | TransFac        | 2.14    | 9714942 | 9714955 |
| SHROOM2 Intron 7 |           |           |       |          |                 |         | 9715462 | 9716970 |
| SHROOM2 Exon 8   |           |           |       |          |                 |         | 9716971 | 9717142 |
|                  |           |           |       | XBP-1    | TransFac        | 1.8     | 9717024 | 9717041 |
|                  |           |           |       | CREB     | TransFac        | 1.88    | 9717028 | 9717043 |
| SHROOM2 Intron 8 |           |           |       |          |                 |         | 9717143 | 9722416 |
|                  | rs4830676 | Genotyped | 0.933 |          |                 |         | 9717327 |         |
|                  | rs4830677 | Imputed   | 0.981 |          |                 |         | 9717408 |         |
|                  | rs3788949 | Genotyped | 0.901 |          |                 |         | 9718077 |         |
|                  |           |           |       | c-Jun    | ENCODE ChIP-seq | 287     | 9718122 | 9718532 |
|                  |           |           |       | Max      | ENCODE ChIP-seq | 617     | 9718167 | 9718668 |
|                  | rs3788950 | Genotyped | 0.782 |          |                 |         | 9718173 |         |
| DNaseI HS        |           |           |       | ENCODE   | 1000            | 9718196 | 9718466 |         |
|                  | rs3788951 | Imputed   | 0.620 |          |                 |         | 9718313 |         |
| DNaseI HS        |           |           |       | ENCODE   | 263             | 9718960 | 9719466 |         |
|                  |           |           |       | NRSF     | ENCODE ChIP-seq | 1000    | 9719025 | 9719285 |
|                  |           |           |       | POU2F2   | ENCODE ChIP-seq | 219     | 9719081 | 9719535 |
|                  |           |           |       | EBF      | ENCODE ChIP-seq | 124     | 9719213 | 9719527 |
|                  |           |           |       | NFKB     | ENCODE ChIP-seq | 136     | 9719215 | 9719495 |
|                  |           |           |       | BATF     | ENCODE ChIP-seq | 324     | 9719224 | 9719494 |
|                  |           |           |       | PAX5-C20 | ENCODE ChIP-seq | 1000    | 9719717 | 9720249 |
|                  |           |           |       | p300     | ENCODE ChIP-seq | 890     | 9719748 | 9720412 |
|                  |           |           |       | TCF12    | ENCODE ChIP-seq | 1000    | 9719769 | 9720334 |
|                  |           |           |       | NFKB     | ENCODE ChIP-seq | 373     | 9719775 | 9720393 |
|                  |           |           |       | IRF4     | ENCODE ChIP-seq | 985     | 9719783 | 9720382 |
|                  |           |           |       | PAX5-N19 | ENCODE ChIP-seq | 1000    | 9719786 | 9720294 |
|                  |           |           |       | EBF      | ENCODE ChIP-seq | 983     | 9719795 | 9720338 |
|                  |           |           |       | BCL11A   | ENCODE ChIP-seq | 1000    | 9719799 | 9720337 |
|                  |           |           |       | SP1      | ENCODE ChIP-seq | 450     | 9719821 | 9720356 |

|                  |           |           |       |          |                 |      |         |         |
|------------------|-----------|-----------|-------|----------|-----------------|------|---------|---------|
|                  |           |           |       | PU.1     | ENCODE ChIP-seq | 221  | 9719823 | 9720227 |
|                  |           |           |       | BCL3     | ENCODE ChIP-seq | 1000 | 9719828 | 9720318 |
|                  |           |           |       | POU2F2   | ENCODE ChIP-seq | 440  | 9719847 | 9720327 |
|                  |           |           |       | BATF     | ENCODE ChIP-seq | 1000 | 9719859 | 9720273 |
|                  |           |           |       | Max      | ENCODE ChIP-seq | 425  | 9719901 | 9720280 |
|                  | rs5934722 | Genotyped | 0.476 |          |                 |      | 9719918 |         |
| DNaseI HS        |           |           |       |          | ENCODE          | 724  | 9719920 | 9720166 |
|                  |           |           |       | Pbx3     | ENCODE ChIP-seq | 543  | 9719975 | 9720240 |
|                  |           |           |       | EBF      | ENCODE ChIP-seq | 364  | 9721122 | 9721539 |
|                  |           |           |       | IRF4     | ENCODE ChIP-seq | 299  | 9721138 | 9721546 |
|                  |           |           |       | POU2F2   | ENCODE ChIP-seq | 211  | 9721144 | 9721542 |
|                  |           |           |       | BCL11A   | ENCODE ChIP-seq | 517  | 9721173 | 9721533 |
|                  |           |           |       | PAX5-N19 | ENCODE ChIP-seq | 467  | 9721180 | 9721503 |
|                  |           |           |       | NFKB     | ENCODE ChIP-seq | 143  | 9721201 | 9721606 |
|                  |           |           |       | PAX5-C20 | ENCODE ChIP-seq | 523  | 9721212 | 9721497 |
|                  |           |           |       | TCF12    | ENCODE ChIP-seq | 701  | 9721240 | 9721552 |
|                  |           |           |       | BATF     | ENCODE ChIP-seq | 405  | 9721248 | 9721530 |
|                  |           |           |       | Pbx3     | ENCODE ChIP-seq | 112  | 9721288 | 9721445 |
| DNaseI HS        |           |           |       |          | ENCODE          | 329  | 9721296 | 9721570 |
|                  | rs4830679 | Genotyped | 0.721 |          |                 |      | 9722022 |         |
|                  | rs3815015 | Imputed   | 0.561 |          |                 |      | 9722222 |         |
|                  |           |           |       | POU2F1   | TransFac        | 1.81 | 9722274 | 9722293 |
|                  | rs3815016 | Genotyped | 0.626 |          |                 |      | 9722278 |         |
| SHROOM2 Exon 9   |           |           |       |          |                 |      | 9722417 | 9722689 |
|                  |           |           |       | E47      | TransFac        | 2.39 | 9722444 | 9722460 |
|                  |           |           |       | Lmo2     | TransFac        | 2.01 | 9722446 | 9722458 |
|                  |           |           |       | RP58     | TransFac        | 1.95 | 9722567 | 9722579 |
|                  |           |           |       | Pax-5    | TransFac        | 1.87 | 9722607 | 9722635 |
| SHROOM2 Intron 9 |           |           |       |          |                 |      | 9722690 | 9724446 |
|                  | rs2238887 | Imputed   | 0.847 |          |                 |      | 9723258 |         |
|                  | rs2238888 | Genotyped | 0.926 |          |                 |      | 9723489 |         |
|                  | rs2238889 | Imputed   | 0.878 |          |                 |      | 9724026 |         |
|                  | rs2073940 | Imputed   | 0.758 |          |                 |      | 9724075 |         |
|                  | rs2073941 | Genotyped | 0.715 |          |                 |      | 9724403 |         |
| SHROOM2 Exon 10  |           |           |       |          |                 |      | 9724447 | 9727217 |
|                  |           |           |       | Pax-5    | TransFac        | 1.71 | 9724604 | 9724632 |
|                  | rs2073942 | Imputed   | 0.937 |          |                 |      | 9724683 |         |
|                  |           |           |       | EBF      | ENCODE ChIP-seq | 52   | 9724919 | 9725203 |
|                  |           |           |       | Pbx3     | ENCODE ChIP-seq | 136  | 9724982 | 9725197 |
| DNaseI HS        |           |           |       |          | ENCODE          | 127  | 9725016 | 9725306 |
|                  |           |           |       | BCL11A   | ENCODE ChIP-seq | 447  | 9725157 | 9725532 |
|                  |           |           |       | NFKB     | ENCODE ChIP-seq | 47   | 9725161 | 9725519 |
|                  |           |           |       | LCR-F1   | TransFac        | 1.7  | 9725217 | 9725239 |
|                  |           |           |       | BATF     | ENCODE ChIP-seq | 118  | 9725221 | 9725509 |
|                  |           |           |       | TCF12    | ENCODE ChIP-seq | 288  | 9725245 | 9725470 |
|                  | rs1858884 | Genotyped | 0.553 |          |                 |      | 9725585 |         |
| DNaseI HS        |           |           |       |          | ENCODE          | 182  | 9726296 | 9726486 |
| DNaseI HS        |           |           |       |          | ENCODE          | 98   | 9727256 | 9727406 |
|                  |           |           |       | BCL3     | ENCODE ChIP-seq | 127  | 9727338 | 9727577 |
|                  | rs2188384 | Genotyped | 0.275 |          |                 |      | 9729314 |         |

**Supplementary Table 5: Relationship between SNP genotype and gene expression in colonic tissue, CRC, lymphoblastoid cell lines (LCL), fibroblasts, T-cells and adipocytes.** Box plots are shown only for selected associations.

| Variant                   | Gene/probe                     | Colonic epithelium <sup>y</sup><br>P-value            | CRC <sup>§</sup><br>P-value         | LCL<br>P-value<br>Twin1<br>Twin2<br>Geneva <sup>1,2</sup> | Fibroblast<br>P-value<br>Twin1<br>Twin2<br>Geneva <sup>1,2</sup> | Adipose<br>P-value<br>Twin1<br>Twin2 <sup>1,2</sup> | T-cell<br>P-value<br>Geneva <sup>1</sup> |
|---------------------------|--------------------------------|---|-------------------------------------|---|--|---|--|
| rs1321311<br>(6p21)       | <i>CDKN1A</i><br>ILMN_1784602  | 0.28  | -                                   | 0.91<br>0.98<br>0.15                                      | 0.97<br>0.95<br>0.42   | 0.53<br>0.38  | 0.0001                                   |
| rs1321311<br>(6p21)       | <i>CDKN1A</i><br>ILMN_1787212  | -   | -                                   | 4.0x10 <sup>-5</sup><br>0.008<br>0.002                    | 0.14<br>0.37<br>0.16   | 0.73<br>0.17  | 0.46                                     |
| rs1321310*<br>(6p21)      | <i>CDKN1A</i><br>A_24_P89457   | -   | 0.46                                | -<br>-<br>-   | -<br>-<br>-  | -<br>-<br>-   | -  |
| rs3824999<br>(11q13.4)    | <i>POLD3</i><br>ILMN_1751773   | 0.25  | -                                   | 0.73<br>0.79<br>0.18                                      | 0.37<br>0.72<br>0.04   | 0.24<br>0.64  | 0.17                                     |
| rs3824999<br>(11q13.4)    | <i>POLD3</i><br>ILMN_2233552   | -   | -                                   | 0.16<br>0.06<br>0.12                                      | 0.43<br>0.33<br>0.02   | 0.54<br>0.49  | 0.56                                     |
| rs11236187**<br>(11q13.4) | <i>POLD3</i><br>A_32_P182439   |   | 0.12                                | -<br>-<br>-   | -<br>-<br>-  | -<br>-<br>-   | -  |
| rs5934683<br>(Xp22.2)     | <i>SHROOM2</i><br>ILMN_1681777 | 1.28x10 <sup>-7</sup><br>Male-0.0003<br>Female-0.0004 |                                     | -<br>-<br>-   | -<br>-<br>-  | -<br>-<br>-   | -  |
| rs5934683<br>(Xp22.2)     | <i>SHROOM2</i><br>A_23_P217753 | -   | 0.015<br>Male-0.77<br>Female-0.0037 | -<br>-<br>-   | -<br>-<br>-  | -<br>-<br>-   | -  |
| rs5934683<br>(Xp22.2)     | <i>GPR143</i><br>ILMN_1756261  | 0.47<br>Male-0.89<br>Female-0.24                      |                                     | -<br>-<br>-   | -<br>-<br>-  | -<br>-<br>-   | -  |

¥Illumina analysis of 42 colon tissue samples

§Data on AgilentG4502A\_07\_3 custom gene expression platform derived from the analysis of 154 CRCs as part of the Cancer Genome Atlas project: <http://cancergenome.nih.gov>

Geneva: Fibroblast, LCL and T-cells derived from the umbilical cords of 75 Geneva GenCord individuals<sup>1</sup>

Twin1 and Twin 2: 166 adipose, 156 LCL and 160 skin samples derived from a subset of healthy female twins of the MuTHER resource<sup>2</sup>

\*Linkage disequilibrium between rs1321311 and rs1321310 –  $r^2=1.0$ ,  $D'=1.0$ .

\*\*Linkage disequilibrium between rs3824999 and rs11236187 –  $r^2=1.0$ ,  $D'=1.0$ .

## REFERENCES

1. Dimas, A.S. et al. Common regulatory variation impacts gene expression in a cell type-dependent manner. *Science* **325**, 1246-50 (2009).
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## SUPPLEMENTARY NOTE

### 1. Subjects and datasets

Scotland1 (COGS)<sup>8</sup> included 1,012 CRC cases (51% male; mean age at diagnosis 49.6 years, SD±6.1) and 1,012 cancer-free population controls (51% male; mean age 51.0 years; SD±5.9). Cases were for early age at onset (age ≤55 years). Known dominant polyposis syndromes, HNPCC/Lynch syndrome or bi-allelic *MYH* mutation carriers were excluded. Control subjects were sampled from the Scottish population NHS registers, matched by age (±5 years), gender and area of residence within Scotland.

Scotland2 (SOCCS)<sup>8</sup> comprised 2,057 CRC cases (61% male; mean age at diagnosis 65.8 years, SD±8.4) and 2,111 population controls (60% males; mean age 67.9 years, SD±9.0) ascertained in Scotland. Cases were taken from an independent, prospective, incident CRC case series and aged <80 years at diagnosis. Control subjects were population controls matched by age (±5 years), gender and area of residence within Scotland.

UK1 (CORGI)<sup>8</sup> comprised 940 cases with colorectal neoplasia (47% male) ascertained through the Colorectal Tumour Gene Identification (CoRGI) consortium. All had at least one first-degree relative affected by CRC and one or more of the following phenotypes: CRC at age 75 or less; any colorectal adenoma (CRAd) at age 45 or less; ≥3 colorectal adenomas at age 75 or less; or a large (>1 cm diameter) or aggressive (villous and/or severely dysplastic) adenoma at age 75 or less. The 965 controls (45% males) were spouses or partners unaffected by cancer and without a personal family history (to 2<sup>nd</sup> degree relative level) of colorectal neoplasia. Known dominant polyposis syndromes, HNPCC/Lynch syndrome or bi-allelic *MYH* mutation carriers were excluded. All cases and controls were of white UK ethnic origin.



UK2 (NSCCG)<sup>8</sup> consisted of 2,873 CRC cases (58% male, mean age at diagnosis 59.3 years; SD±8.7) ascertained through two ongoing initiatives at the Institute of Cancer Research/Royal Marsden Hospital NHS Trust (RMHNSHT) from 1999 onwards - The National Study of Colorectal Cancer Genetics (NSCCG)<sup>29</sup> and the Royal Marsden Hospital Trust/Institute of Cancer Research Family History and DNA Registry. The 2,871 controls (41% males; mean age 59.8 years; SD±10.8) were the spouses or unrelated friends of patients with malignancies. None had a personal history of malignancy at time of ascertainment. All cases and controls had self-reported European ancestry, and there were no obvious differences in the demography of cases and controls in terms of place of residence within the UK.

VQ58 comprised 1,800 CRC cases (1,099 males, mean age of diagnosis 62.5 years; SD±10.9) from the VICTOR<sup>30</sup> and QUASAR2 ([www.oct-oxford.org.uk/alltrials/trials/q2.html](http://www.oct-oxford.org.uk/alltrials/trials/q2.html)) trials. There were 2,690 population control genotypes (1,391 males,) from the Wellcome Trust Case-Control Consortium 2 (WTCCC2) 1958 birth cohort (also known as the National Child Development Study), which included all births in England, Wales and Scotland during a single week in 1958<sup>31</sup>.

The COIN samples were 2,183 cases derived from the COIN and COIN-B clinical trials of metastatic CRC<sup>32</sup>. Median age was 63 years. COIN cases were compared against genotypes from 2,501 population controls (1,237 males,) from the WTCCC2 National Blood Service (NBS) cohort (50% male; mean age at diagnosis 53.2 years, SD±15.4).

The Colon Cancer Family Registry (CCFR1) data set comprised 1,290 familial CRC cases and 1,055 controls Colon Cancer Family Registry (Colon-CFR) ([http://epi.grants.cancer.gov/CFR/about\\_colon.html](http://epi.grants.cancer.gov/CFR/about_colon.html))<sup>33</sup>. The cases were recently diagnosed CRC cases reported to population complete cancer registries in the USA (Puget Sound, Washington State) who were recruited by the Seattle Familial Colorectal Cancer Registry; in Canada (Ontario) who were recruited by the Ontario Familial Cancer Registry; and in Australia (Melbourne, Victoria) who were recruited by the Australasian

Colorectal Cancer Family Study. Controls were population-based and for this analysis were restricted to those without a family history of colorectal cancer.

UK3 (NSCCG)<sup>8</sup> comprised 7,862 CRC cases (65% male; mean age at diagnosis 59 years, SD±8.2) and 4,431 controls (40% male; mean age 62 years, SD±11.5) ascertained through NSCCG post-2005<sup>29</sup>.

UK4 (CORGI2BCD)<sup>8</sup> consisted of 589 CRC cases (46% male; mean age at diagnosis 58.3 years; SD±14.1) and 1,087 cancer-free population or spouse controls (45% male; mean age 45.1 years, SD±15.9).

Scotland3 (SOCCS)<sup>8</sup> comprised 736 CRC cases (50% male; mean age at diagnosis 53.2 years, SD±15.4) and 1,522 cancer-free population controls (47% male; mean age 51.8 years, SD±11.5). Controls comprised cancer-free participants in the Lothian Birth Cohort 1921 and Lothian Birth Cohort 1936.

Cambridge/SEARCH consisted of 2,248 CRC cases (56% male; mean age at diagnosis 59.2 years, SD±8.1) and 2,288 controls (42% males; mean age 57.6 years; SD±15.1). Samples were ascertained through the SEARCH (Studies of Epidemiology and Risk Factors in Cancer Heredity, <http://www.cancerhelp.org.uk/trials/a-study-looking-at-genetic-causes-of-cancer>) study based in Cambridge, UK. Recruitment started in 2000; initial patient contact was through the general practitioner. Control samples were collected post-2003. Eligible individuals were sex- and frequency-matched in five-year age bands to cases.

In Croatia, the cases (n=375) were recruited during preoperative work-up and individuals coming for a regular follow-up after the surgery for colorectal cancer, at the surgical unit of the University hospital "Sestre Milosrdnice" in Zagreb, Croatia, in the period between 2008-2010. Only individuals with adenocarcinoma were included in this study. Although all recruited cases come to Zagreb for treatment, a substantial proportion

were resident elsewhere in Croatia. Controls (N=1,051) comprised individuals with no malignant disease recruited from the surgical unit of the same hospital and healthy subjects from Split, Croatia. Both Zagreb and Split represent urban centres that attract immigration from other parts of Croatia. Hence both populations are mixed and comprise diverse Croatian subpopulations. Nevertheless, we acknowledge there may be population substructure within Croatian population.

The Helsinki (FCCPS) study (<http://research.med.helsinki.fi/gsb/aaltonen/>) comprised 1,013 cases from a population-based collection centred on south-eastern Finland and 827 population controls from the same collection.

The Swedish study comprised 3,217 CRC patients were recruited within a Swedish national study conducted by the Swedish Low-Risk Colorectal Cancer Study Group. Samples were obtained during 2004-2009 from 14 different surgical clinics in central Sweden. All CRC patients during the study period were eligible for recruitment and were invited to participate. Only those too ill or too frail to consent were excluded. Controls (n=2,895) comprised blood donors from Stockholm and Uppsala. Fully informed consent was obtained in accordance with the Swedish law concerning ethical approval of research on human subjects (refs:2002:489,2003:198,2010:1213-31/4).

The Japanese study comprised 1,583 colorectal cancer cases and 1,898 control subjects as described previously<sup>34</sup>. All cases and controls were obtained from Biobank Japan. These samples were genotyped using the Illumina Human610-QuadBeadChip in cases and the Illumina HumanHap550v3 BeadChip in controls. Exclusion criteria: Samples with a call rate of <0.98, SNP quality call rates <0.95, Hardy-Weinberg  $P < 1.0 \times 10^{-7}$  in controls.

In all cases CRC was defined according to the ninth revision of the International Classification of Diseases (ICD) by codes 153–154.

## 2. ADDITIONAL PCA BASED ANALYSIS

Although no substantial population structure was indicated by analysis of the QQ plots, the Eigenstrat software was utilised to check for the effect of any substructure on the results for the SNPs reported in this paper. Adjusting along the lowest ten principal components and recalculating  $p$ -values showed minimal changes in the association statistics (shown below). It should be noted that the eigenstrat software does not facilitate proper testing of the x-chromosome and the results reported here are based on males being treated as female homozygotes.

**Table of Eigenstrat adjusted  $p$ -values for implicated SNPs (10PCs)**

|             | Inflation ( $\lambda$ ) |      | rs1321311            |                      | rs3824999            |                      | rs5934683            |                      |
|-------------|-------------------------|------|----------------------|----------------------|----------------------|----------------------|----------------------|----------------------|
|             | PRE                     | POST | PRE                  | POST                 | PRE                  | POST                 | PRE                  | POST                 |
| <b>LP1</b>  | 1.02                    | 1.01 | $2.0 \times 10^{-2}$ | $1.8 \times 10^{-2}$ | $4.9 \times 10^{-2}$ | $7.3 \times 10^{-2}$ | $3.3 \times 10^{-4}$ | $4.9 \times 10^{-4}$ |
| <b>SP1</b>  | 1.01                    | 1.00 | $2.4 \times 10^{-2}$ | $1.6 \times 10^{-2}$ | $5.9 \times 10^{-2}$ | $4.4 \times 10^{-2}$ | $5.8 \times 10^{-1}$ | $5.8 \times 10^{-1}$ |
| <b>LP2</b>  | 1.04                    | 1.03 | $7.2 \times 10^{-2}$ | $6.6 \times 10^{-2}$ | $1.7 \times 10^{-2}$ | $1.4 \times 10^{-2}$ | $1.2 \times 10^{-1}$ | $1.2 \times 10^{-1}$ |
| <b>SP2</b>  | 1.03                    | 1.04 | $3.6 \times 10^{-1}$ | $3.4 \times 10^{-1}$ | $2.0 \times 10^{-1}$ | $2.2 \times 10^{-1}$ | $6.5 \times 10^{-2}$ | $8.3 \times 10^{-2}$ |
| <b>VQ</b>   | 1.01                    | 1.01 | $4.8 \times 10^{-1}$ | $5.2 \times 10^{-1}$ | $5.4 \times 10^{-5}$ | $9.2 \times 10^{-5}$ | $9.0 \times 10^{-2}$ | $1.1 \times 10^{-1}$ |
| <b>CFR1</b> | 1.02                    | 1.01 | $1.4 \times 10^{-1}$ | $9.7 \times 10^{-2}$ | $1.9 \times 10^{-2}$ | $2.4 \times 10^{-2}$ | $5.4 \times 10^{-2}$ | $4.6 \times 10^{-2}$ |