

Supplementary Table S2. Regions with loss of heterozygosity (LOH)

Supplementary Table S2. Regions with loss of heterozygosity (LOH) > 3 Mb in the patient

Type	Chromosome	Cytoband Start	Cytoband End	CytoRegions	Size (kb)	Marker Count	Gene Count	OMIM® Genes Count
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LOH

16

q22.1

q22.1

q22.1

3502,323

468

100

66

LOH	16	p11.2	p11.1	p11.2, p11.1	3590,042	236	18	1
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Supplementary Table S2. Regions with loss of heterozygosity (LOH) < 3 Mb in the patient

Type	Chromosome	Cytoband Start	Cytoband End	CytoRegions	Size (kb)	Marker Count	Gene Count	OMIM® Genes Count
LOH	1	p36.13	p36.13	p36.13	1439,436	221	21	12

LOH

3

q22.2

q22.3

q22.3, q22.2 1404,567

229

7

6

LOH

3

q29

q29

q29

1102,963

177

12

8

LOH	4	p15.1	p15.1	p15.1	2121,445	508	0	0
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LOH	4	q13.3	q13.3	q13.3	1262,461	276	5	5
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LOH	5	q12.1	q12.1	q12.1	1014,239	235	5	3
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LOH	5	q14.3	q14.3	q14.3	1199,182	298	3	1
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LOH	5	q33.3	q33.3	q33.3	1257,535	296	17	11
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LOH

7

q11.23

q11.23

q11.23

1323,292

49

23

8

LOH

7

q31.1

q31.1

q31.1

1162,935

292

3

2

LOH	8	q24.21	q24.21	q24.21	1100,735	295	1	0
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LOH	9	q22.32	q22.33	q22.33, q22.32	1097,566	200	18	4
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LOH

9

q34.11

q34.11

q34.11

1082,798

123

43

28

LOH	10	q11.22	q11.22	q11.22	1333,722	167	17	8
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LOH

10

q26.13

q26.13

q26.13

1017,369

287

19

10

LOH	11	p14.3	p14.2	p14.2, p14.3	1165,542	521	1	1
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LOH

12

q14.2

q14.3

q14.3, q14.2 1715,137

395

14

9

LOH	12	q24.31	q24.31	q24.31	1298,504	167	31	21
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LOH	13	q21.31	q21.32	q21.32, q21.31	1211,013	300	0	0
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LOH

13

q32.3

q32.3

q32.3

1326,902

364

13

6

LOH

19

p13.2

p13.2

p13.2

1030,625

219

31

11

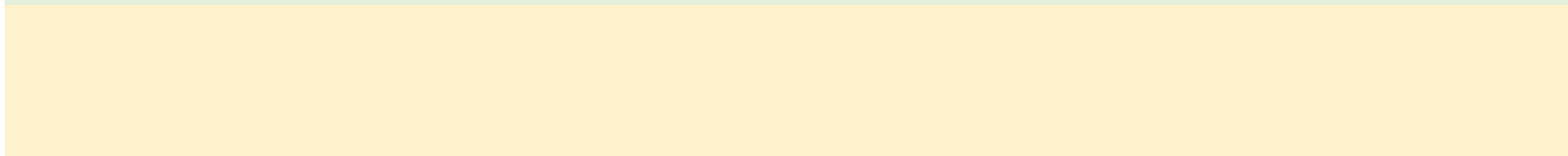
OMIM[®] Genes

Full Location

TRADD (603500), FBXL8 (609077), HSF4 (602438), NOL3 (605235), EXOC3L1 (614117), E2F4 (600659), ELMO3 (606422), MIR328 (613701), FHOD1 (606881), SLC9A5 (600477), PLEKHG4 (609526), TPPP3 (616957), HSD11B2 (614232), ATP6V0D1 (607028), AGRP (602311), CTCF (604167), CARMIL2 (610859), ACD (609377), PARD6A (607484), RANBP10 (614031), TSNAXIP1 (607720), CENPT (611510), THAP11 (609119), NUTF2 (605813), EDC4 (606030), PSKH1 (177015), CTRL (118888), PSMB10 (176847), LCAT (606967), SLC12A4 (604119), DPEP3 (609926), DPEP2 (609925), DDX28 (607618), DUS2 (609707), NFATC3 (602698), ESRP2 (612960), PLA2G15 (609362), SLC7A6 (605641), PRMT7 (610087), SMPD3 (605777), ZFP90 (609451), CDH3 (114021), CDH1 (192090), HAS3 (602428), CHTF8 (613202), UTP4 (607456), SNTB2 (600027), VPS4A (609982), COG8 (606979), TERF2 (602027), CYB5B (611964), NFAT5 (604708), NQO1 (125860), NOB1 (613586), WWP2 (602308), MIR140 (611894), CLEC18A (616571), CLEC18C (616573), EXOSC6 (606490), AARS (601065), DDX19B (605812), ST3GAL2 (607188), FUK (608675), COG4 (606976), SF3B3 (605592), IL34 (612081) chr16:67174211-70676534

ZNF267 (604752)

chr16:31630502-35220544



OMIM® Genes**Full Location**

NECAP2 (611624), NBPF1 (610501), MFAP2 (156790), ATP13A2 (610513), SDHB (185470), PADI2 (607935), PADI1 (607934), PADI3 (606755), PADI4 (605347), PADI6 (610363), RCC2 (609587), ARHGEF10L (612494)

chr1:16736737-18176173

PPP2R3A (604944), MSL2 (614802), PCCB (232050), STAG1 (604358), NCK1 (600508), IL20RB
(605621)

chr3:135285824-136690391

XXYLT1 (614552), ACAP2 (607766), PPP1R2 (601792), APOD (107740), MIR570 (614538), MUC20 (610360), MUC4 (158372), TNK2 (606994) chr3:194650488-195753451

chr4:32417743-34539188

MOB1B (609282), DCK (125450), SLC4A4 (603345), GC (139200), NPFFR2 (607449)

chr4:71709992-72972453

KIF2A (602591), DIMT1 (612499), IPO11 (610889)

chr5:61362329-62376568

COX7C (603774)

chr5:84996731-86195913

SGCD (601411), TIMD4 (610096), HAVCR1 (606518), HAVCR2 (606652), MED7 (605045), ITK (186973), CYFIP2 (606323), NIPAL4 (609383), ADAM19 (603640), SOX30 (606698), CLINT1 (607265) chr5:156038647-157296182

GTF2IRD1 (604318), GTF2I (601679), NCF1 (608512), GTF2IRD2 (608899), GTF2IRD2B (608900), chr7:73843868-75167160
TRIM73 (612549), TRIM74 (612550), HIP1 (601767)

IMMP2L (605977), DOCK4 (607679)

chr7:110303526-111466461

chr8:129124948-130225683

SLC35D2 (609182), ZNF367 (610160), CDC14B (603505), CTSV (603308)

chr9:99066432-100163998

ENG (131195), AK1 (103000), ST6GALNAC6 (610135), ST6GALNAC4 (606378), PIP5KL1 (612865), chr9:130607711-131690509
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COQ4 (612898), SLC27A4 (604194), URM1 (612693), ODF2 (602015), GLE1 (603371), SPTAN1
(182810), WDR34 (613363), SET (600960), PKN3 (610714), ENDOG (600440), CCBL1 (600547),
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RBP3 (180290), GDF2 (605120), GDF10 (601361), PTPN20B (610631), PTPN20A (610630), FRMPD2 (613323), MAPK8 (601158), ARHGAP22 (610585) chr10:48365294-49699016

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ANO3 (610110)

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chr13:65352957-66563970

GPR18 (602042), GPR183 (605741), TM9SF2 (604678), CLYBL (609686), ZIC2 (603073), PCCA
(232000)

chr13:99774561-101101463

PRKCSH (177060), ELAVL3 (603458), ZNF653 (611371), ECSIT (608388), CNN1 (600806), ACP5 (171640), ZNF627 (612248), ZNF69 (194543), ZNF20 (194557), ZNF136 (604078), ZNF44 (194542) chr19:11509008-12539633

DGV

Segmental Duplications

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DGV**Segmental Duplications**

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Genes

Microarray Nomenclature

C16orf70, B3GNT9, TRADD, FBXL8, HSF4, NOL3, KIAA0895L, EXOC3L1, E2F4, ELMO3, MIR328, LRRC29, TMEM208, FHOD1, SLC9A5, PLEKHG4, KCTD19, LRRC36, TPPP3, ZDHHC1, HSD11B2, ATP6V0D1, AGRP, LOC100505942, FAM65A, CTCF, CARMIL2, ACD, PARD6A, ENKD1, C16orf86, GFOD2, RANBP10, TSNAXIP1, CENPT, THAP11, NUTF2, EDC4, NRN1L, PSKH1, CTRL, PSMB10, LCAT, SLC12A4, DPEP3, DPEP2, LOC100131303, DDX28, DUS2, NFATC3, ESRP2, MIR6773, PLA2G15, SLC7A6, SLC7A6OS, PRMT7, SMPD3, ZFP90, CDH3, CDH1, MIR7641-2, TANGO6, HAS3, CHTF8, UTP4, SNTB2, VPS4A, PDF, COG8, NIP7, TMED6, TERF2, CYB5B, MIR1538, NFAT5, NQO1, NOB1, WWP2, MIR140, CLEC18A, PDXDC2P, MIR1972-2, MIR1972-1, PDPR, LOC400541, CLEC18C, LOC105371328, SMG1P7, EXOSC6, AARS, DDX19B, LOC100506083, DDX19A, ST3GAL2, FUK, COG4, SF3B3, SNORD111B, SNORD111, IL34

arr[hg19] 16q22.1(67,174,211-70,676,534) hmz

CLUHP3, ZNF720, ZNF267, LOC102723753, HERC2P4, TP53TG3D, LOC390705, arr[hg19] 16p11.2p11.1(31,630,502-35,220,544) hmz
TP53TG3, TP53TG3B, TP53TG3C, SLC6A10P, ENPP7P13, LINC00273, UBE2MP1,
LINC01566, FRG2DP, TP53TG3HP, FLJ26245

Genes

SPATA21, NECAP2, CROCCP3, MIR3675, NBPF1, CROCCP2, MST1P2, ESPNP, MST1L, CROCC, MFAP2, ATP13A2, SDHB, PADI2, PADI1, PADI3, PADI4, PADI6, RCC2, ARHGEF10L, ACTL8

Microarray Nomenclature

arr[hg19] 1p36.13(16,736,737-18,176,173) hmz

PPP2R3A, MSL2, PCCB, STAG1, SLC35G2, NCK1, IL20RB

arr[hg19] 3q22.2q22.3(135,285,824-136,690,391) hmz

XXYLT1, XXYLT1-AS2, ACAP2, MIR5692C1, PPP1R2, APOD, SDHAP2, MIR570, arr[hg19] 3q29(194,650,488-195,753,451) hmz
MUC20, MUC4, TNK2, SDHAP1

arr[hg19] 4p15.1(32,417,743-34,539,188) hmz

MOB1B, DCK, SLC4A4, GC, NPFFR2

arr[hg19] 4q13.3(71,709,992-72,972,453) hmz

KIF2A, DIMT1, IPO11, LRRC70, IPO11-LRRC70

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NBPF22P, COX7C, MIR3607

arr[hg19] 5q14.3(84,996,731-86,195,913) hmz

SGCD, PPP1R2P3, TIMD4, HAVCR1, HAVCR2, MED7, FAM71B, ITK, CYFIP2,
FNDC9, NIPAL4, ADAM19, SOX30, C5orf52, THG1L, LSM11, CLINT1

arr[hg19] 5q33.3(156,038,647-157,296,182) hmz

GTF2IRD1, GTF2I, NCF1, GTF2IRD2, STAG3L2, PMS2P5, GATSL1, WBSR16,
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STAG3L1, TRIM73, TRIM74, NSUN5P1, POM121C, SPDYE5, PMS2P3, HIP1

arr[hg19] 7q11.23(73,843,868-75,167,160) hmz

IMMP2L, LRRN3, DOCK4

arr[hg19] 7q31.1(110,303,526-111,466,461) hmz

MIR1208

arr[hg19] 8q24.21(129,124,948-130,225,683) hmz

SLC35D2, ZNF367, HABP4, CDC14B, AAED1, LOC441455, ZNF510, ZNF782,
LOC100132781, LOC441454, NUTM2G, HIATL2, CTSL2, LOC340508,
LOC100499484, LOC100499484-C9ORF174, CCDC180, LOC286359

arr[hg19] 9q22.32q22.33(99,066,432-100,163,998) hmz

ENG, AK1, MIR4672, ST6GALNAC6, ST6GALNAC4, PIP5KL1, DPM2, FAM102A, arr[hg19] 9q34.11(130,607,711-131,690,509) hmz
NAIF1, SLC25A25, LOC100289019, PTGES2, PTGES2-AS1, LCN2, C9orf16, CIZ1,
DNM1, MIR199B, MIR3154, GOLGA2, SWI5, TRUB2, COQ4, SLC27A4, URM1,
MIR219-2, MIR2964A, CERCAM, ODF2, GLE1, SPTAN1, WDR34, SET, PKN3,
ZDHHC12, LOC100506100, ZER1, TBC1D13, ENDOG, C9orf114, CCBL1, LRRC8A,
PHYHD1

ZNF488, RBP3, GDF2, GDF10, PTPN20A, PTPN20B, FRMPD2P1, BMS1P1,
BMS1P5, GLUD1P7, FAM25B, FAM25C, FAM25G, LOC399753, FRMPD2,
MAPK8, ARHGAP22

arr[hg19] 10q11.22(48,365,294-49,699,016) hmz

PLEKHA1, MIR3941, ARMS2, HTRA1, DMBT1, C10orf120, FLJ46361, CUZD1,
FAM24B-CUZD1, FAM24B, LOC399815, FAM24A, C10orf88, PSTK, IKZF5,
ACADSB, HMX3, HMX2, BUB3

arr[hg19] 10q26.13(124,165,614-125,182,983) hmz

ANO3

arr[hg19] 11p14.3p14.2(25,377,880-26,543,422) hmz

SRGAP1, C12orf66, C12orf56, XPOT, TBK1, RASSF3, MIR548Z, MIR548C, GNS, arr[hg19] 12q14.2q14.3(64,434,226-66,149,363) hmz
TBC1D30, FLJ41278, WIF1, LEMD3, MSRB3

HCAR3, HCAR1, DENR, CCDC62, HIP1R, VPS37B, ABCB9, OGFOD2, ARL6IP4, arr[hg19] 12q24.31(123,201,038-124,499,542) hmz
PITPNM2, MIR4304, LOC100507091, MPHOSPH9, C12orf65, CDK2AP1, SBNO1,
SETD8, RILPL2, SNRNP35, RILPL1, MIR3908, TMED2, DDX55, EIF2B1, GTF2H3,
TCTN2, ATP6VOA2, DNAH10, CCDC92, ZNF664, ZNF664-FAM101A

arr[hg19] 13q21.31q21.32(65,352,957-66,563,970) hmz

UBAC2-AS1, UBAC2, MIR548AN, GPR18, GPR183, FKSG29, MIR623, TM9SF2, arr[hg19] 13q32.3(99,774,561-101,101,463) hmz
CLYBL, MIR4306, ZIC5, ZIC2, PCCA

RGL3, CCDC151, PRKCSH, ELAVL3, ZNF653, ECSIT, CNN1, ELOF1, ACP5, ZNF627, arr[hg19] 19p13.2(11,509,008-12,539,633) hmz
ZNF833P, ZNF823, ZNF441, ZNF491, ZNF440, ZNF439, ZNF69, ZNF700, ZNF763,
ZNF433, ZNF878, ZNF844, ZNF788, ZNF20, ZNF625-ZNF20, ZNF625, ZNF136,
ZNF44, ZNF563, ZNF442, ZNF799