

Supplementary File 1

Mutation analysis of the following cells lines (used in our study) from COSMIC (Catalogue Of Somatic Mutations In Cancer <http://cancer.sanger.ac.uk/cosmic>)

Cell lines » Sample » Overview » NCI-H1650

View in GRCh37 Arc

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|---------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|---------------------------------|-------------|-------------------------------------|----------------------------------|-------------------|--------------|------------|------------------------------|--|
| Non-Coding mutation CNV & Expr Methylation | | | | | | | | | |
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| Gene | Transcript | Census Gene | AA Mutation | CDS Mutation | Somatic status | Zygoty | Validated | Type | |
| APC | ENST00000457016 | Yes | p.A1358T | c.4072G>A | Unknown | Heterozygous | Unverified | Substitution - Missense | |
| CAMTA1 | ENST00000303635 | Yes | p.S239L | c.716C>T | Unknown | Heterozygous | Unverified | Substitution - Missense | |
| DEK | ENST00000397239 | Yes | p.K221K | c.663A>G | Unknown | Homozygous | Unverified | Substitution - coding silent | |
| EGFR | ENST00000275493 | Yes | p.E746_A750delELREA | c.2235_2249del15 | Confirmed Somatic | Heterozygous | Verified | Deletion - In frame | |
| NF1 | ENST00000358273 | Yes | p.? | c.3114-7T>C | Unknown | Heterozygous | Unverified | Unknown | |
| SBDS | ENST00000246868 | Yes | p.I133V | c.397A>G | Unknown | Heterozygous | Unverified | Substitution - Missense | |
| SFPO | ENST00000357214 | Yes | p.A651A | c.1953A>T | Unknown | Heterozygous | Unverified | Substitution - coding silent | |
| TP53 | ENST00000269305 | Yes | p.? | c.673-2A>G | Confirmed Somatic | Homozygous | Verified | Unknown | |

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| Gene | Transcript | Census Gene | AA Mutation | CDS Mutation | Somatic status | Zygoty | Validated | Type |
|-------------------------|---------------------------------|-------------|----------------------------|----------------------------------|---------------------|--------------|------------|------------------------------|
| ATR | NM_001184 | Yes | p.?? | c.2634-1G>A | Unknown | Heterozygous | Unverified | Unknown |
| ATR | NM_001184 | Yes | p.Q1441Q | c.4323A>G | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| CARD11 | ENST00000396946 | Yes | p.M1124I | c.3372G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| CBL | ENST00000264033 | Yes | p.R585C | c.1753C>T | Unknown | Homozygous | Unverified | Substitution - Missense |
| CTNNA1 | ENST00000302763 | Yes | p.E865* | c.2593G>T | Unknown | Heterozygous | Unverified | Substitution - Nonsense |
| ERC1 | ENST00000397203 | Yes | p.T1032T | c.3096C>A | Unknown | Homozygous | Unverified | Substitution - coding silent |
| FH | ENST00000366560 | Yes | p.G464V | c.1391G>T | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| FLT3 | ENST00000241453 | Yes | p.N323K | c.969C>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| FUS | ENST00000254108 | Yes | p.R383H | c.1148G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| HIP1 | ENST00000336926 | Yes | p.S277N | c.830G>A | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| KRAS | ENST00000311936 | Yes | p.G12S | c.34G>A | Confirmed Somatic | Homozygous | Verified | Substitution - Missense |
| PCSK7 | ENST00000320934 | Yes | p.? | c.1156-9C>T | Unknown | Homozygous | Unverified | Unknown |
| POLE | ENST00000320574 | Yes | p.Q1475Q | c.4425G>A | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| SMARCA4 | ENST00000429416 | Yes | p.Q729fs*4 | c.2184_2206del23 | Confirmed Somatic | Heterozygous | Verified | Deletion - Frameshift |
| SMARCA4 | ENST00000429416 | Yes | p.Q729fs*4 | c.2184_2206del23 | Previously Reported | Homozygous | Verified | Deletion - Frameshift |
| STK11 | ENST00000326873 | Yes | p.Q37* | c.109C>T | Confirmed Somatic | Homozygous | Verified | Substitution - Nonsense |
| SUFU | ENST00000369902 | Yes | p.T411M | c.1232C>T | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| TBL1XR1 | ENST00000430069 | Yes | p.T290A | c.868A>G | Unknown | Homozygous | Unverified | Substitution - Missense |
| USP6 | NM_004505.1 | Yes | p.P31T | c.91C>A | Unknown | Heterozygous | Unverified | Substitution - Missense |

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| Gene | Transcript | Census Gene | AA Mutation | CDS Mutation | Somatic status | Zygoty | Validated | Type |
|-------------------------|---------------------------------|-------------|--------------------------------------|----------------------------------|---------------------|--------------|------------|------------------------------|
| ATP2B3 | ENST00000359149 | Yes | p.V882E | c.2645T>A | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| BCR | ENST00000305877 | Yes | p.I456T | c.1367T>C | Unknown | Heterozygous | Unverified | Substitution - Missense |
| CLIP1 | ENST00000358808 | Yes | p.N890S | c.2669A>G | Unknown | Heterozygous | Unverified | Substitution - Missense |
| DNM2 | ENST00000355667 | Yes | p.A800V | c.2399C>T | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| ECT2L | ENST00000367682 | Yes | p.T727T | c.2181C>T | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| EP300 | ENST00000263253 | Yes | p.R1356* | c.4066C>T | Previously Reported | Heterozygous | Unverified | Substitution - Nonsense |
| FLT3 | ENST00000241453 | Yes | p.C368Y | c.1103G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| FLT3 | ENST00000241453 | Yes | p.C368S | c.1102T>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| GATA3 | ENST00000379328 | Yes | p.D336fs*17 | c.1006_1007insG | Previously Reported | Heterozygous | Unverified | Insertion - Frameshift |
| MAP3K13 | ENST00000424227 | Yes | p.D380N | c.1138G>A | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| MYH9 | ENST00000216181 | Yes | p.K682_L687delKLDPHL | c.2044_2061del18 | Unknown | Heterozygous | Unverified | Deletion - In frame |
| NBN | ENST00000265433 | Yes | p.R43* | c.127C>T | Unknown | Heterozygous | Unverified | Substitution - Nonsense |
| PALB2 | ENST00000261584 | Yes | p.V560L | c.1678G>C | Unknown | Heterozygous | Unverified | Substitution - Missense |
| PIK3CA | NM_006218.1 | Yes | p.E545K | c.1633G>A | Confirmed Somatic | Heterozygous | Verified | Substitution - Missense |

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| Non-Coding mutation | CNV & Expr | Methylation | | | | | | |
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| Gene | Transcript | Census Gene | AA Mutation | CDS Mutation | Somatic status | Zygosity | Validated | Type |
| AFF3 | ENST00000409579 | Yes | p.N983N | c.2949C>T | Unknown | Heterozygous | Verified | Substitution - coding silent |
| AFF3 | ENST00000409579 | Yes | p.S1112* | c.3335C>G | Unknown | Heterozygous | Unverified | Substitution - Nonsense |
| ARHGFE12 | ENST00000397843 | Yes | p.Q933E | c.2797C>G | Unknown | Heterozygous | Unverified | Substitution - Missense |
| ATM | ENST00000278616 | Yes | p.E2468K | c.7402G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| AXIN1 | ENST00000262320 | Yes | p.? | c.2186+8C>T | Unknown | Heterozygous | Unverified | Unknown |
| BCOR | ENST00000397354 | Yes | p.H457P | c.1370A>C | Unknown | Homozygous | Unverified | Substitution - Missense |
| BRCA2 | ENST00000380152 | Yes | p.S3094* | c.9281C>A | Unknown | Heterozygous | Unverified | Substitution - Nonsense |
| BRIP1 | ENST00000259008 | Yes | p.S230L | c.689C>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| BUB1B | ENST00000287598 | Yes | p.S521F | c.1562C>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| CARD11 | ENST00000396946 | Yes | p.L15L | c.45G>A | Previously Reported | Heterozygous | Unverified | Substitution - coding silent |
| CCND1 | ENST00000227507 | Yes | p.L217L | c.651G>C | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| CIC | ENST00000160740 | Yes | p.L1467V | c.4399C>G | Unknown | Heterozygous | Unverified | Substitution - Missense |
| COL2A1 | ENST00000337299 | Yes | p.G669S | c.2005G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| EGFR | ENST00000275493 | Yes | p.D1014D | c.3042C>T | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| EPS15 | ENST00000371733 | Yes | p.G522E | c.1565G>A | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| FANCA | ENST00000389301 | Yes | p.L684L | c.2052G>A | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| FANCA | ENST00000389301 | Yes | p.S1114fs*3 | c.3340delT | Unknown | Heterozygous | Unverified | Deletion - Frameshift |
| FLT4 | ENST00000393347 | Yes | p.? | c.3431+7C>T | Unknown | Heterozygous | Unverified | Unknown |
| FLT4 | ENST00000393347 | Yes | p.A634T | c.1900G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| GNA11 | ENST00000078429 | Yes | p.I167I | c.501C>T | Unknown | Homozygous | Unverified | Substitution - coding silent |
| HIST1H3B | ENST00000244661 | Yes | p.R135T | c.404G>C | Unknown | Heterozygous | Unverified | Substitution - Missense |
| HNF1A | ENST00000257555 | Yes | p.Q495* | c.1483C>T | Unknown | Heterozygous | Unverified | Substitution - Nonsense |
| HOXA13 | ENST00000222753 | Yes | p.S314L | c.941C>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| KIF5B | ENST00000302418 | Yes | p.Q734E | c.2200C>G | Unknown | Heterozygous | Unverified | Substitution - Missense |
| KMT2D | ENST00000301067 | Yes | p.E1350K | c.4048G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| NCOR1 | ENST00000268712 | Yes | p.? | c.6734-3C>T | Unknown | Heterozygous | Unverified | Unknown |
| NSD1 | ENST00000439151 | Yes | p.S1654C | c.4961C>G | Unknown | Heterozygous | Unverified | Substitution - Missense |
| NSD1 | ENST00000439151 | Yes | p.L2063V | c.6187C>G | Unknown | Heterozygous | Unverified | Substitution - Missense |
| NUMA1 | ENST00000393695 | Yes | p.E1479Q | c.4435G>C | Unknown | Homozygous | Unverified | Substitution - Missense |
| PAX3 | ENST00000392069 | Yes | p.P373P | c.1119G>A | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| PDE4DIP | ENST00000369354 | Yes | p.D371N | c.1111G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| PIK3CA | NM_006218.1 | Yes | p.K111N | c.333G>C | Confirmed Somatic | Heterozygous | Verified | Substitution - Missense |



Circos Genome Browser Overview **Mutations** Fusions Mutation Spectrum Sequence Context Heatmap Breakpoints
 Non-Coding mutation CNV & Expr Methylation

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| Gene | Transcript | Census Gene | AA Mutation | CDS Mutation | Somatic status | Zygoty | Validated | Type |
|--------------------------|---------------------------------|-------------|-----------------------------|--------------------------------|---------------------|--------------|------------|------------------------------|
| PLAG1 | ENST00000316981 | Yes | p.D14H | c.40G>C | Unknown | Heterozygous | Unverified | Substitution - Missense |
| PML | ENST00000268058 | Yes | p.L869R | c.2605T>G | Unknown | Heterozygous | Unverified | Substitution - Missense |
| PMS1 | ENST00000441310 | Yes | p.K156R | c.467A>G | Unknown | Heterozygous | Unverified | Substitution - Missense |
| PPFIBP1 | ENST00000228425 | Yes | p.V122M | c.364G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| PPP2R1A | ENST00000322088 | Yes | p.S143F | c.428C>T | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| PTPRK | ENST00000368226 | Yes | p.D884N | c.2650G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| RAP1GDS1 | ENST00000453712 | Yes | p.H122Y | c.364C>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| RECQL4 | ENST00000617875 | Yes | p.M989R | c.2966T>G | Unknown | Heterozygous | Unverified | Substitution - Missense |
| RHQA | ENST00000418115 | Yes | p.G17E | c.50G>A | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| RNF213 | ENST00000336301 | Yes | p.V1085V | c.3255C>T | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| RNF213 | ENST00000336301 | Yes | p.L1176L | c.3528C>G | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| ROS1 | ENST00000368508 | Yes | p.V1916L | c.5746G>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| SMARCB1 | ENST00000263121 | Yes | p.F328F | c.984C>T | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| SS18 | ENST00000415083 | Yes | p.D293N | c.877G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| SSX1 | ENST00000376919 | Yes | p.E149G | c.446A>G | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| STAT3 | ENST00000264657 | Yes | p.F174L | c.522C>G | Unknown | Heterozygous | Unverified | Substitution - Missense |
| TFRC | ENST00000360110 | Yes | p.V108V | c.324G>T | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| TFRC | ENST00000360110 | Yes | p.? | c.1405-9C>G | Unknown | Heterozygous | Unverified | Unknown |
| TP53 | ENST00000269305 | Yes | p.E285K | c.853G>A | Confirmed Somatic | Homozygous | Verified | Substitution - Missense |
| TPM3 | ENST00000368533 | Yes | p.R208fs*10 | c.623_624delGA | Unknown | Heterozygous | Unverified | Deletion - Frameshift |
| WRN | ENST00000298139 | Yes | p.E764G | c.2291A>G | Unknown | Heterozygous | Unverified | Substitution - Missense |
| WRN | ENST00000298139 | Yes | p.H945N | c.2833C>A | Unknown | Heterozygous | Unverified | Substitution - Missense |

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| Overview | Mutations | Fusions | Non-Mutant Genes | CNV & Expr | Methylation | Studies | References | | |
|----------------------------------------------------------------------------------------------------------|-------------------------------------------------------------------------------------------------------------------|-------------|-------------------------------------|----------------------------------|---------------------|--------------|------------|---------------------|--|
| Gene | Transcript | Census Gene | AA Mutation | CDS Mutation | Somatic status | Zygoty | Validated | Type | |
| EGFR  | ENST00000275493  | Yes | p.E746_A750delELREA | c.2235_2249del15 | Previously Reported | Heterozygous | Unverified | Deletion - In frame | |

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Circos Genome Browser Overview Mutations Fusions Mutation Spectrum Sequence Context Heatmap Breakpoints
 Non-Coding mutation CNV & Expr Methylation

| Gene | Transcript | Census Gene | AA Mutation | CDS Mutation | Somatic status | Zygosity | Validated | Type |
|-------------------------|---------------------------------|-------------|-------------------------------------|-------------------------------------|---------------------|--------------|------------|---------------------------------|
| ACSL6 | ENST00000296869 | Yes | p.? | c.1507+1G>T | Unknown | Heterozygous | Unverified | Unknown |
| ALK | ENST00000389048 | Yes | p.L1035L | c.3105G>A | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| ARID1B | ENST00000275248 | Yes | p.A1917A | c.5751G>T | Unknown | Homozygous | Unverified | Substitution - coding silent |
| ARID2 | ENST00000334344 | Yes | p.M452V | c.1354A>G | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| ASXL1 | ENST00000375687 | Yes | p.Y1087H | c.3259T>C | Unknown | Heterozygous | Unverified | Substitution - Missense |
| ATM | ENST00000278616 | Yes | p.Q1919P | c.5756A>C | Unknown | Homozygous | Unverified | Substitution - Missense |
| AXIN2 | ENST00000307078 | Yes | p.T586T | c.1758G>A | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| BCR | ENST00000305877 | Yes | p.R567H | c.1700G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| BRIP1 | ENST00000259008 | Yes | p.E1054A | c.3161A>C | Unknown | Heterozygous | Unverified | Substitution - Missense |
| CCNE1 | ENST00000262643 | Yes | p.D83N | c.247G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| CIC | ENST00000160740 | Yes | p.S1031S | c.3093G>C | Unknown | Homozygous | Unverified | Substitution - coding silent |
| CSF1R | ENST00000286301 | Yes | p.R710R | c.2130C>T | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| CSF1R | ENST00000286301 | Yes | p.P202S | c.604C>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| DDX6 | NM_004397.3 | Yes | p.? | c.1141+5G>A | Unknown | Heterozygous | Unverified | Unknown |
| DICER1 | ENST00000343455 | Yes | p.E732E | c.2196A>G | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| DNMT3A | ENST00000321117 | Yes | p.G223* | c.667G>T | Previously Reported | Heterozygous | Unverified | Substitution - Nonsense |
| EBF1 | ENST00000313708 | Yes | p.P339Q | c.1016C>A | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| FGFR2 | ENST00000358487 | Yes | p.Y328S | c.983A>C | Unknown | Homozygous | Unverified | Substitution - Missense |
| FLT3 | ENST00000241453 | Yes | p.P738H | c.2213C>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| GMPS | ENST00000496455 | Yes | p.V613L | c.1837G>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| GMPS | ENST00000496455 | Yes | p.P612P | c.1836G>T | Previously Reported | Heterozygous | Unverified | Substitution - coding silent |
| GPC3 | ENST00000370818 | Yes | p.V289L | c.865G>T | Previously Reported | Homozygous | Unverified | Substitution - Missense |
| KAT6B | ENST00000287239 | Yes | p.G1134C | c.3400G>T | Previously Reported | Homozygous | Unverified | Substitution - Missense |
| KMT2D | ENST00000301067 | Yes | p.G3195V | c.9584G>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| KRAS | ENST00000311936 | Yes | p.G12C | c.34G>T | Confirmed Somatic | Heterozygous | Verified | Substitution - Missense |
| NBN | ENST00000265433 | Yes | p.V153I | c.457G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| NIN | ENST00000382041 | Yes | p.P1987S | c.5959C>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| NTRK3 | ENST00000394480 | Yes | p.? | c.2293-10A>T | Unknown | Heterozygous | Unverified | Unknown |
| PDE4DJP | ENST00000369354 | Yes | p.V1261L | c.3781G>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| PDGFRA | ENST00000257290 | Yes | p.P250H | c.749C>A | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| PMS2 | ENST00000265849 | Yes | p.E491K | c.1471G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| PTRR8 | NM_002837.2 | Yes | p.T1280M | c.3839C>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| RANBP2 | ENST00000283195 | Yes | p.D387N | c.1159G>A | Unknown | Heterozygous | Unverified | Substitution - Missense |
| SETBP1 | ENST00000282030 | Yes | p.Q1244R | c.3731A>G | Unknown | Homozygous | Unverified | Substitution - Missense |
| SMARCA4 | ENST00000429416 | Yes | p.K1566_E1567>N* | c.4698_4699GG>TT | Unknown | Homozygous | Verified | Complex - compound substitution |
| STK11 | ENST00000326873 | Yes | p.W332* | c.996G>A | Confirmed Somatic | Homozygous | Verified | Substitution - Nonsense |
| TP53 | ENST00000269305 | Yes | p.M246I | c.738G>C | Confirmed Somatic | Homozygous | Verified | Substitution - Missense |
| TRRAP | ENST00000355540 | Yes | p.Y2267Y | c.6801C>T | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| USP6 | NM_004505.1 | Yes | p.T693T | c.2079A>G | Unknown | Heterozygous | Unverified | Substitution - coding silent |

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| Gene | Transcript | Census Gene | AA Mutation | CDS Mutation | Somatic status | Zygosity | Validated | Type |
|-------------------------|---------------------------------|-------------|-------------|---------------------------------|---------------------|--------------|------------|------------------------------|
| ATM | ENST00000278616 | Yes | p.? | c.4237-9A>G | Unknown | Heterozygous | Unverified | Unknown |
| ATM | ENST00000278616 | Yes | p.N100SI | c.3014A>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| AXIN2 | ENST00000307078 | Yes | p.? | c.1713-8C>T | Unknown | Heterozygous | Unverified | Unknown |
| BRAF | ENST00000288602 | Yes | p.G464V | c.1391G>T | Confirmed Somatic | Heterozygous | Verified | Substitution - Missense |
| CASP8 | ENST00000264275 | Yes | p.L132L | c.396A>G | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| CD79A | ENST00000221972 | Yes | p.C106Y | c.317G>A | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| CLTC1 | ENST00000263200 | Yes | p.D1485N | c.4453G>A | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| CRT3 | ENST00000268184 | Yes | p.P578A | c.1732C>G | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| EXT2 | ENST00000533608 | Yes | p.E672E | c.2016G>A | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| GAS7 | ENST00000432992 | Yes | p.? | c.1218+8G>T | Unknown | Homozygous | Unverified | Unknown |
| GNAS | ENST00000371085 | Yes | p.N6N | c.18C>T | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| HOXD13 | ENST00000249505 | Yes | p.P260P | c.780G>A | Unknown | Heterozygous | Unverified | Substitution - coding silent |
| KRAS | ENST00000311936 | Yes | p.G13D | c.38G>A | Confirmed Somatic | Heterozygous | Verified | Substitution - Missense |
| NFI | ENST00000358273 | Yes | p.T467fs*3 | c.1398_1399insC | Unknown | Heterozygous | Unverified | Insertion - Frameshift |
| NF2 | ENST00000338641 | Yes | p.E231* | c.691G>T | Confirmed Somatic | Homozygous | Verified | Substitution - Nonsense |
| PBRM1 | ENST00000337303 | Yes | p.I228V | c.682A>G | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| PCSK7 | ENST00000320934 | Yes | p.P52R | c.155C>G | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| PDGFRA | ENST00000257290 | Yes | p.Y172F | c.515A>T | Previously Reported | Heterozygous | Unverified | Substitution - Missense |
| PER1 | ENST00000317276 | Yes | p.E189D | c.567G>T | Unknown | Homozygous | Unverified | Substitution - Missense |
| SLC45A3 | ENST00000367145 | Yes | p.R251W | c.751C>T | Unknown | Heterozygous | Unverified | Substitution - Missense |
| TCEA1 | ENST00000360389 | Yes | p.? | c.126+7A>T | Unknown | Heterozygous | Unverified | Unknown |
| TP53 | ENST00000269305 | Yes | p.R280K | c.839G>A | Confirmed Somatic | Heterozygous | Verified | Substitution - Missense |

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Circos Genome Browser Overview **Mutations** Fusions Mutation Spectrum Non-Mutant Genes Breakpoints
 Non-Coding mutation CNV & Expr Methylation Studies References

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| Gene | Transcript | Census Gene | AA Mutation | CDS Mutation | Somatic status | Zygoty | Validated | Type |
|------------------------|---------------------------------|-------------|--------------------------|-------------------------------|-------------------|--------------|------------|-------------------------|
| ALK | ENST00000389048 | Yes | p.R69Q | c.206G>A | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| ARID1A | ENST00000324856 | Yes | p.S1707G | c.5119A>G | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| ATR | NM_001184 | Yes | p.S601F | c.1802C>T | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| BRAF | ENST00000288602 | Yes | p.V600E | c.1799T>A | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| BRD3 | ENST00000303407 | Yes | p.S609F | c.1826C>T | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| CAMTA1 | ENST00000303635 | Yes | p.E205K | c.613G>A | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| CDH1 | ENST00000261769 | Yes | p.Y190H | c.568T>C | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| CDKN2A | ENST00000304494 | Yes | p.? | c.150+2T>C | Confirmed Somatic | Heterozygous | Unverified | Unknown |
| COL1A1 | ENST00000225964 | Yes | p.R312C | c.934C>T | Confirmed Somatic | Homozygous | Unverified | Substitution - Missense |
| COL2A1 | ENST00000337299 | Yes | p.R989C | c.2965C>T | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| DICER1 | ENST00000343455 | Yes | p.M1825I | c.5475G>A | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| EBF1 | ENST00000313708 | Yes | p.G107R | c.319G>A | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| ERC1 | ENST00000397203 | Yes | p.S392F | c.1175C>T | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| KMT2C | ENST00000262189 | Yes | p.E4319K | c.12955G>A | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| PTPRK | ENST00000368226 | Yes | p.H1045Y | c.3133C>T | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| RHOH | ENST00000505618 | Yes | p.R157W | c.469C>T | Confirmed Somatic | Homozygous | Unverified | Substitution - Missense |
| SEPT6 | ENST00000343984 | Yes | p.P181S | c.541C>T | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| SPEN | ENST00000375759 | Yes | p.P3345S | c.10033C>T | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| SPEN | ENST00000375759 | Yes | p.R423C | c.1267C>T | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |
| TP53 | ENST00000269305 | Yes | p.G266E | c.797G>A | Confirmed Somatic | Heterozygous | Unverified | Substitution - Missense |

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