Whole Exome report

Date: 10/05/2020

Cell line: RPZ27911

Method Summary:

DNA was extracted from RPZ27911 using Qiagen all prep protocol. gDNA libraries from 500ng DNA was prepared and exons were captured using Agilent SureselectXT mouse all exon protocol. Libraries were quantitated via Agilent TapeStation. Sequencing was performed on the Illumina Novaseq6000. Raw FastQ files were mapped using the Dragen protocols for somatic mutation detection and using Pipeliner protocol for germline mutation detection. Single Nucleotide variants and insertions or deletions were identified, and a subset of calls filtered by quality, depth of coverage, and allele frequency were compiled into a variant list. A subset of the compiled variants was reviewed manually using the Integrated Genome Viewer software from the Broad Institute.

*FastQ files* are stored at smb://at-s-is2.ncifcrf.gov/ras-intl/static/Genomics/ 20200924\_6/

*BAM files from Dragen calling* are stored at smb://at-s-is2.ncifcrf.gov/ras-intl/static/Genomics/ 20200924\_6/

Please find the whole exome analysis workflow [here](file:///Volumes/ras-intl/static/Genomics/20200218_4RPZ_2Mras/reports/WES_analysis_workflow_updata20200115.html).

Summary of findings:

Indels 1 high impact

SNV 2 High impact, 53 Moderate deleterious impact

Listed Genes

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| GENE | #CHROM | POS | AF | IMPACT | AMINO ACID CHANGE | CLASS |
| Acsm2 | chr7 | 119591126 | 0.466 | HIGH | Arg80Ter | I-B |
| Pycr1 | chr11 | 120641427 | 0.525 | HIGH | Spicing variant | I-B |
| Rad51c | chr11 | 87404420 | 0.394 | HIGH | Met1? | I-B |
| Adam20 | chr8 | 40796308 | 0.256 | MODERATE | Cys485Tyr | II-A |
| Alad | chr4 | 62510200 | 0.296 | MODERATE | Ser279Tyr | II-A |
| Apaf1 | chr10 | 91036950 | 0.459 | MODERATE | Asp755Ala | II-A |
| Apc2 | chr10 | 80302397 | 0.533 | MODERATE | Glu106Gln | II-A |
| Avp | chr2 | 130581073 | 0.477 | MODERATE | Gly106Ala | II-B |
| C3 | chr17 | 57211712 | 0.439 | MODERATE | Asp1224Tyr | II-A |
| Catsperb | chr12 | 101624705 | 0.511 | MODERATE | Tyr1011Asn | II-A |
| Cdh9 | chr15 | 16849120 | 0.446 | MODERATE | Arg476Ile | II-A |
| Cspg4 | chr9 | 56887160 | 0.462 | MODERATE | Trp726Cys | II-A |
| Ddb1 | chr19 | 10628451 | 0.438 | MODERATE | Lys1081Met | II-A |
| Edem1 | chr6 | 108851214 | 0.471 | MODERATE | Thr500Ser | II-B |
| Egln2 | chr7 | 27165187 | 0.371 | MODERATE | Cys104Trp | II-A |
| F10 | chr8 | 13039651 | 0.301 | MODERATE | Thr89Ser | II-B |
| Gemin5 | chr11 | 58155179 | 0.51 | MODERATE | Trp434Cys | II-A |
| Grin3b | chr10 | 79976555 | 0.335 | MODERATE | Ser876Arg | II-A |
| Hdc | chr2 | 126604297 | 0.297 | MODERATE | Pro181Ala | II-A |
| Hoxd1 | chr2 | 74763679 | 0.462 | MODERATE | Ser193Tyr | II-A |
| Igsf8 | chr1 | 172316507 | 0.239 | MODERATE | Ser139Cys | II-A |
| Ilk | chr7 | 105742207 | 0.52 | MODERATE | Ala360Gly | II-B |
| Kbtbd8 | chr6 | 95122344 | 0.482 | MODERATE | Leu278Pro | II-A |
| Lrrc10b | chr19 | 10456885 | 0.397 | MODERATE | Gly144Ala | II-B |
| Ltn1 | chr16 | 87379716 | 0.482 | MODERATE | Ala1734Gly | II-B |
| Mfsd12 | chr10 | 81362733 | 0.498 | MODERATE | Trp353Gly | II-A |
| Midn | chr10 | 80155224 | 0.457 | MODERATE | Ala356Gly | II-B |
| Ndrg4 | chr8 | 95706979 | 0.291 | MODERATE | Ala173Gly | II-B |
| Nolc1 | chr19 | 46081340 | 0.474 | MODERATE | Gly223Ala | II-B |
| Ntsr2 | chr12 | 16659820 | 0.467 | MODERATE | Lys367Asn | II-A |
| Oit3 | chr10 | 59425442 | 0.398 | MODERATE | Ser468Pro | II-A |
| Olfr402 | chr11 | 74155835 | 0.524 | MODERATE | Ala227Val | II-A |
| Olfr482 | chr7 | 108095126 | 0.474 | MODERATE | Val148Asp | II-A |
| Olfr715 | chr7 | 107129045 | 0.5 | MODERATE | Ala116Asp | II-A |
| Olfr890 | chr9 | 38143240 | 0.49 | MODERATE | Leu35Pro | II-A |
| Plxna2 | chr1 | 194749333 | 0.508 | MODERATE | Cys543Trp | II-A |
| Ppfibp2 | chr7 | 107738278 | 0.487 | MODERATE | Leu607Val | II-A |
| Ppp1r3c | chr19 | 36733702 | 0.419 | MODERATE | Pro223Ala | II-A |
| Prpf38b | chr3 | 108904173 | 0.452 | MODERATE | Glu459Lys | II-A |
| Pth2r | chr1 | 65388465 | 0.202 | MODERATE | Ile432Met | II-A |
| Rad50 | chr11 | 53650612 | 0.435 | MODERATE | Val1266Leu | II-A |
| Rasgef1b | chr5 | 99232100 | 0.453 | MODERATE | Ala307Pro | II-A |
| Serpinb3b | chr1 | 107154410 | 0.483 | MODERATE | Thr375Ala | II-A |
| Slc35a2 | chrX | 7889700 | 1 | MODERATE | Leu110Ile | II-B |
| Slc38a1 | chr15 | 96579383 | 0.515 | MODERATE | Leu350Ile | II-B |
| Smc3 | chr19 | 53621814 | 0.308 | MODERATE | Ala211Pro | II-A |
| Tctex1d4 | chr4 | 117128306 | 0.275 | MODERATE | Glu109Gln | II-A |
| Tekt4 | chr17 | 25474778 | 0.504 | MODERATE | Gln296His | II-A |
| Tpo | chr12 | 30092752 | 0.44 | MODERATE | Phe658Val | II-A |
| Tprgl | chr4 | 154160168 | 0.549 | MODERATE | Cys123Trp | II-A |
| Trav7n-4 | chr14 | 53091856 | 0.556 | MODERATE | Phe108Val | II-A |
| Trmt112 | chr19 | 6910228 | 0.476 | MODERATE | Gly15Ala | II-B |
| Vmn2r23 | chr6 | 123742220 | 0.54 | MODERATE | Arg844Ile | II-A |
| Vps51 | chr19 | 6070971 | 0.49 | MODERATE | Thr304Ser | II-B |
| Wbp2nl | chr15 | 82308579 | 0.451 | MODERATE | Tyr155His | II-A |
| Zfp647 | chr15 | 76911722 | 0.449 | MODERATE | Ala246Gly | II-B |

Genes with yellow background have variants detected by Novaseq but missed by previous Highseq.

I-A             Mutation call is well supported and there is a high probability that the variant will impact RAS dependent proliferation in this cell (e.g. Homozygous mutation of Trp53)

*Variants called by one of variant calling methods and visualized by IGV. Single variant on a gene, AF>0.9 with high impact. Or multiple variants on a gene, 0.9>AF>0.2 with high impact.*

*Variants called by one of variant calling methods and visualized by IGV. Single variant on a gene, AF>0.9 with moderate deleterious impact. Or multiple variants on a gene, 0.9>AF>0.2 with moderate deleterious impact.*

I-B             Mutation call is well supported, the variant is probably damaging, the variant may impact cell growth or genetic stability

*Variants called by one of variant calling methods and visualized by IGV. Single variant on a gene, 0.9>AF>0.2 with high impact.*

II-A        Call is well supported, variant is non-synonymous and likely impact protein function

*Variants called by one of variant calling methods and visualized by IGV. Single variant on a gene, 0.9>AF>0.2 with moderate deleterious impact.*

II-B           Call is well supported, non-synonymous change in amino acids with minor change in amino acid properties (e.g. Serine to Threonine, Ile to Leu, or Glycine to Alanine) that may alter protein function

*Variants called by one of variant calling methods and visualized by IGV. Single variant on a gene, AF>0.2 with moderate deleterious impact* and *with minor change in amino acid properties (e.g. Serine to Threonine, Ile to Leu, or Glycine to Alanine).*

II-C Mutation call is well supported but the change is unlikely to impact protein function.