Genomics Research Centre Diagnostics Clinic Genotyping Analysis Report

Patient: DG0000A

RUNID: {Number derived from IonTorrent}

Analysis Performed: {to show what parameters were used, includes version number of curated files?}

Date Analysis Performed:

{Perhaps we can also have a table of summary data here from the stuff that Miles demonstrated, as well as some quality metrics from the Ion Server, to give an idea of overall quality?}

{The report should be ideally editable, at least in parts, to allow marking of false positives or other human note taking, or to remove data that’s extraneous for sequencing customers}

**Tier 1 Annotations – Disease Specific Genes** {FHM, Ataxia, etc. For epilepsy and others, perhaps tiers could have specific names as there may be more tiers than the “standard” arrangement and some may be more likely to check for a specific patient }

Tier 1.1 – Top 10 Most Damaging Mutations/Variants {Amino acid changing variant, no rs# OR Not amino acid changing and has no rs#; amino acid changing, has rs#, but MAF <1% - All sorted by the number of predictors showing “Damaging” or the equivalent}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus**\*** | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A2 | Exon | - | - | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN1A | Exon | - | - | NM\_007242.1 | c.166G>A | p.Thr55Met | 2.33 | 0.42 | Disease Causing | 0 | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN1A | Exon | - | - | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR2 | Exon | - | - | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH3 | Exon | - | - | NM\_0033503.2 | c.3166G>A | p.Ala1056Thr | -0.56 | 0.36 | Polymorphism | 0 | 20 | G=10, A=10 |

{**\*** - Primary sort in table by Locus}

**{#**- Because Mutation Taster (and maybe other reports) have a lot of information, the listing here might need to be a link to a second document or something appended to the end of this report to show all consequences}

Tier 1.2 – Mutations {Amino acid changing variant, no rs#}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus**\*** | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A2 | Exon | - | - | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN1A | Exon | - | - | NM\_007242.1 | c.166G>A | p.Thr55Met | 2.33 | 0.42 | Disease Causing | 0 | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN1A | Exon | - | - | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR2 | Exon | - | - | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH3 | Exon | - | - | NM\_0033503.2 | c.3166G>A | p.Ala1056Thr | -0.56 | 0.36 | Polymorphism | 0 | 20 | G=10, A=10 |

{**\*** - Primary sort in table by Locus}

**{#**- Because Mutation Taster (and maybe other reports) have a lot of information, the listing here might need to be a link to a second document or something appended to the end of this report to show all consequences}

Tier 1.3 – Variants {Not amino acid changing and has no rs#; amino acid changing, has rs#, but MAF <1%}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A2 | Exon | 0.001 | rs33874396 | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN1A | 3\_UTR | - | - | NM\_007242.1 | c.-166G>A | - | - | - | Disease Causing | - | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN1A | Exon | 0.01 | rs897976 | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR2 | Exon | 0.005 | rs785858 | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH3 | Intron | - | - | NM\_0033503.2 | c.3166+13G>A | - | - | - | Polymorphism | - | 20 | G=10, A=10 |

Tier 1.4 – Rare SNPs {any variant with rs# and MAF >1%<10%}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A2 | Exon | 0.05 | rs33874396 | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN1A | 3\_UTR | 0.02 | rs344645765 | NM\_007242.1 | c.-166G>A | - | - | - | Disease Causing | - | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN1A | Exon | 0.09 | rs897976 | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR2 | Exon | 0.08 | rs785858 | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH3 | Intron | 0.03 | rs3464 | NM\_0033503.2 | c.3166+13G>A | - | - | - | Polymorphism | - | 20 | G=10, A=10 |

**Tier 2 Annotations – Pathway Specific Genes** {CNS, Muscle, Cardiac, etc. Drawn from KEGG or similar database?}

Tier 2.1 – Top 10 Most Damaging Mutations/Variants {Amino acid changing variant, no rs# OR Not amino acid changing and has no rs#; amino acid changing, has rs#, but MAF <1% - All sorted by the number of predictors showing “Damaging” or the equivalent}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus**\*** | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A6 | Exon | - | - | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN4A | Exon | - | - | NM\_007242.1 | c.166G>A | p.Thr55Met | 2.33 | 0.42 | Disease Causing | 0 | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN4A | Exon | - | - | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR6 | Exon | - | - | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH1 | Exon | - | - | NM\_0033503.2 | c.3166G>A | p.Ala1056Thr | -0.56 | 0.36 | Polymorphism | 0 | 20 | G=10, A=10 |

Tier 2.2 – Mutations {Amino acid changing variant, no rs#}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus**\*** | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A6 | Exon | - | - | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN4A | Exon | - | - | NM\_007242.1 | c.166G>A | p.Thr55Met | 2.33 | 0.42 | Disease Causing | 0 | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN4A | Exon | - | - | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR6 | Exon | - | - | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH1 | Exon | - | - | NM\_0033503.2 | c.3166G>A | p.Ala1056Thr | -0.56 | 0.36 | Polymorphism | 0 | 20 | G=10, A=10 |

{**\*** - Primary sort in table by Locus}

**{#**- Because Mutation Taster (and maybe other reports) have a lot of information, the listing here might need to be a link to a second document or something appended to the end of this report to show all consequences}

Tier 2.3 – Variants {Not amino acid changing and has no rs#; amino acid changing, has rs#, but MAF <1%}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A6 | Exon | 0.001 | rs33874396 | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN4A | 3\_UTR | - | - | NM\_007242.1 | c.-166G>A | - | - | - | Disease Causing | - | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN4A | Exon | 0.01 | rs897976 | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR6 | Exon | 0.005 | rs785858 | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH1 | Intron | - | - | NM\_0033503.2 | c.3166+13G>A | - | - | - | Polymorphism | - | 20 | G=10, A=10 |

Tier 2.4 – Rare SNPs {any variant with rs# and MAF >1%<10%}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A6 | Exon | 0.05 | rs33874396 | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN4A | 3\_UTR | 0.02 | rs344645765 | NM\_007242.1 | c.-166G>A | - | - | - | Disease Causing | - | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN4A | Exon | 0.09 | rs897976 | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR6 | Exon | 0.08 | rs785858 | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH1 | Intron | 0.03 | rs3464 | NM\_0033503.2 | c.3166+13G>A | - | - | - | Polymorphism | - | 20 | G=10, A=10 |

**Tier 3 Annotations – All Other Genes** {Anything not included in the previous sets. I would suggest perhaps moving the Rare SNPs for this tier down to Tier 4, as there will be too many to work with. That may also need to be done for the Epilepsy or other panels where there’s more than 30 or so targeted genes in a tier}.

Tier 3.1 – Top 10 Most Damaging Mutations/Variants {Amino acid changing variant, no rs# OR Not amino acid changing and has no rs#; amino acid changing, has rs#, but MAF <1% - All sorted by the number of predictors showing “Damaging” or the equivalent}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus**\*** | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A6 | Exon | - | - | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN4A | Exon | - | - | NM\_007242.1 | c.166G>A | p.Thr55Met | 2.33 | 0.42 | Disease Causing | 0 | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN4A | Exon | - | - | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR6 | Exon | - | - | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH1 | Exon | - | - | NM\_0033503.2 | c.3166G>A | p.Ala1056Thr | -0.56 | 0.36 | Polymorphism | 0 | 20 | G=10, A=10 |

{**\*** - Primary sort in table by Locus}

**{#**- Because Mutation Taster (and maybe other reports) have a lot of information, the listing here might need to be a link to a second document or something appended to the end of this report to show all consequences}

Tier 3.2 – Mutations {Amino acid changing variant, no rs#}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus**\*** | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A6 | Exon | - | - | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN4A | Exon | - | - | NM\_007242.1 | c.166G>A | p.Thr55Met | 2.33 | 0.42 | Disease Causing | 0 | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN4A | Exon | - | - | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR6 | Exon | - | - | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH1 | Exon | - | - | NM\_0033503.2 | c.3166G>A | p.Ala1056Thr | -0.56 | 0.36 | Polymorphism | 0 | 20 | G=10, A=10 |

{**\*** - Primary sort in table by Locus}

**{#**- Because Mutation Taster (and maybe other reports) have a lot of information, the listing here might need to be a link to a second document or something appended to the end of this report to show all consequences}

Tier 3.3 – Variants {Not amino acid changing and has no rs#; amino acid changing, has rs#, but MAF <1%}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A6 | Exon | 0.001 | rs33874396 | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN4A | 3\_UTR | - | - | NM\_007242.1 | c.-166G>A | - | - | - | Disease Causing | - | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN4A | Exon | 0.01 | rs897976 | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR6 | Exon | 0.005 | rs785858 | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH1 | Intron | - | - | NM\_0033503.2 | c.3166+13G>A | - | - | - | Polymorphism | - | 20 | G=10, A=10 |

*Tier 3.4 – Rare SNPs {any variant with rs# and MAF >1%<10%}*

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| *Locus* | *Genotype* | *Ref* | *Gene* | *Location* | *MAF* | *dbSNP* | *Transcript* | *Coding* | *Amino Acid* | *PhyloP* | *Sift* | *Mutation Taster****#*** | *PolyPhen* | *Coverage* | *Allele Coverage* |
| *chr1:160090674* | *C/C* | *T* | *ATP1A6* | *Exon* | *0.05* | *rs33874396* | *NM\_007203.3* | *c.3166G>A* | *p.Ala1056Thr* | *-0.78* | *0.5* | *Disease Causing* | *0* | *26* | *T=0, C=26* |
| *chr1:160090681* | *T/C* | *C* | *SCN4A* | *3\_UTR* | *0.02* | *rs344645765* | *NM\_007242.1* | *c.-166G>A* | *-* | *-* | *-* | *Disease Causing* | *-* | *25* | *T=12, C=13* |
| *chr1:160093222* | *CCT/CCCT* | *CCT* | *SCN4A* | *Exon* | *0.09* | *rs897976* | *NM\_007242.1* | *c.3166G>A* | *p.Ala1056Thr* | *-0.1* | *0.6* | *Polymorphism* | *0* | *36* | *CCT=15, CCCT=21* |
| *chr1:160109567* | *CG/CG* | *C* | *SCLR6* | *Exon* | *0.08* | *rs785858* | *NM\_007245.3* | *c.366C>CG* | *p.Pro156X* | *0.76* | *0.54* | *Disease Causing* | *1* | *49* | *C=0, CG=49* |
| *chr2:166846016* | *G/A* | *A* | *NOTCH1* | *Intron* | *0.03* | *rs3464* | *NM\_0033503.2* | *c.3166+13G>A* | *-* | *-* | *-* | *Polymorphism* | *-* | *20* | *G=10, A=10* |

**Tier 4 Annotations – Polymorphisms** {Polymorphisms for all Tier }.

Tier 4.1 – Polymorphisms for Tier 1 Genes {any variant with rs# and MAF >10%}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A2 | Exon | 0.55 | rs33874396 | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN1A | 3\_UTR | 0.2 | rs344645765 | NM\_007242.1 | c.-166G>A | - | - | - | Disease Causing | - | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN1A | Exon | 0.1 | rs897976 | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR2 | Exon | 0.3 | rs785858 | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH3 | Intron | 0.34 | rs3464 | NM\_0033503.2 | c.3166+13G>A | - | - | - | Polymorphism | - | 20 | G=10, A=10 |

Tier 4.2 – Polymorphisms for Tier 2 Genes {any variant with rs# and MAF >10%}

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Locus | Genotype | Ref | Gene | Location | MAF | dbSNP | Transcript | Coding | Amino Acid | PhyloP | Sift | Mutation Taster**#** | PolyPhen | Coverage | Allele Coverage |
| chr1:160090674 | C/C | T | ATP1A6 | Exon | 0.55 | rs33874396 | NM\_007203.3 | c.3166G>A | p.Ala1056Thr | -0.78 | 0.5 | Disease Causing | 0 | 26 | T=0, C=26 |
| chr1:160090681 | T/C | C | SCN4A | 3\_UTR | 0.2 | rs344645765 | NM\_007242.1 | c.-166G>A | - | - | - | Disease Causing | - | 25 | T=12, C=13 |
| chr1:160093222 | CCT/CCCT | CCT | SCN4A | Exon | 0.1 | rs897976 | NM\_007242.1 | c.3166G>A | p.Ala1056Thr | -0.1 | 0.6 | Polymorphism | 0 | 36 | CCT=15, CCCT=21 |
| chr1:160109567 | CG/CG | C | SCLR6 | Exon | 0.3 | rs785858 | NM\_007245.3 | c.366C>CG | p.Pro156X | 0.76 | 0.54 | Disease Causing | 1 | 49 | C=0, CG=49 |
| chr2:166846016 | G/A | A | NOTCH1 | Intron | 0.34 | rs3464 | NM\_0033503.2 | c.3166+13G>A | - | - | - | Polymorphism | - | 20 | G=10, A=10 |

*Tier 4.3 –* Polymorphisms for Tier 3 Genes *{any variant with rs# and MAF >10%}*

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| *Locus* | *Genotype* | *Ref* | *Gene* | *Location* | *MAF* | *dbSNP* | *Transcript* | *Coding* | *Amino Acid* | *PhyloP* | *Sift* | *Mutation Taster****#*** | *PolyPhen* | *Coverage* | *Allele Coverage* |
| *chr1:160090674* | *C/C* | *T* | *ATP1A6* | *Exon* | *0.55* | *rs33874396* | *NM\_007203.3* | *c.3166G>A* | *p.Ala1056Thr* | *-0.78* | *0.5* | *Disease Causing* | *0* | *26* | *T=0, C=26* |
| *chr1:160090681* | *T/C* | *C* | *SCN4A* | *3\_UTR* | *0.2* | *rs344645765* | *NM\_007242.1* | *c.-166G>A* | *-* | *-* | *-* | *Disease Causing* | *-* | *25* | *T=12, C=13* |
| *chr1:160093222* | *CCT/CCCT* | *CCT* | *SCN4A* | *Exon* | *0.1* | *rs897976* | *NM\_007242.1* | *c.3166G>A* | *p.Ala1056Thr* | *-0.1* | *0.6* | *Polymorphism* | *0* | *36* | *CCT=15, CCCT=21* |
| *chr1:160109567* | *CG/CG* | *C* | *SCLR6* | *Exon* | *0.3* | *rs785858* | *NM\_007245.3* | *c.366C>CG* | *p.Pro156X* | *0.76* | *0.54* | *Disease Causing* | *1* | *49* | *C=0, CG=49* |
| *chr2:166846016* | *G/A* | *A* | *NOTCH1* | *Intron* | *0.34* | *rs3464* | *NM\_0033503.2* | *c.3166+13G>A* | *-* | *-* | *-* | *Polymorphism* | *-* | *20* | *G=10, A=10* |