|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Location** | **Variant allele** | **Consequence** | **Gene** | **dbSNP ID, HGMD ID** | **SIFT** | **PolyPhen** | **CADD Phred score** | **Clinical significance in ClinVar** |
| 1:33430102-33430102 | G | missense\_variant | *RNF19B* | rs113840389 | tolerated\_low\_confidence(0.29) | benign(0) | 6.343 | - |
| 1:94884114-94884114 | T | missense\_variant | *ABCD3* | rs554914466 | tolerated(0.64) | benign(0.027) | 9.050 | - |
| **11:31816238-31816238** | **A** | **missense\_variant** | ***PAX6*** | **rs757259413,CM930572** | **deleterious(0)** | **probably\_damaging(1)** | **29.3** | **pathogenic** |
| 16:70884524-70884524 | G | missense\_variant | *HYDIN* | rs1798314 | deleterious(0.01) | benign(0.255) | 25.0 | - |
| 19:55246731-55246731 | T | missense\_variant | *KIR3DL3* | - | deleterious(0.05) | benign(0.042) | 11.26 | - |
| 19:55246741-55246741 | C | missense\_variant | *KIR3DL3* | rs662386 | tolerated(0.09) | benign(0.024) | 0.147 | - |
| 19:55253552-55253552 | A | missense\_variant | *KIR2DL3* | rs78713511 | tolerated(0.94) | benign(0.007) | 0.001 | - |
| 19:55255377-55255377 | T | missense\_variant | *KIR2DL3* | - | tolerated(0.19) | benign(0.005) | 11.20 | - |
| 19:55358655-55358655 | G | missense\_variant | *KIR2DS4* | - | tolerated\_low\_confidence(0.2) | benign(0) | 0.324 | - |
| 21:14982716-14982716 | C | missense\_variant | *POTED* | rs201206142 | tolerated\_low\_confidence(1) | benign(0) | 0.001 | - |
| 3:75714298-75714298 | G | missense\_variant | *FRG2C* | rs74714110 | tolerated(0.64) | benign(0) | 0.001 | - |
| 3:75714337-75714337 | G | missense\_variant | *FRG2C* | rs73840323 | tolerated(1) | benign(0) | 0.001 | - |
| 3:75714702-75714702 | G | missense\_variant | *FRG2C* | - | tolerated(1) | benign(0) | 0.001 | - |
| 3:75786379-75786379 | C | missense\_variant | *ZNF717* | rs80170761 | tolerated(0.09) | benign(0.047) | 0.017 | - |
| 3:75787224-75787224 | T | missense\_variant | *ZNF717* | rs112847134 | tolerated(0.54) | benign(0.001) | 3.560 | - |
| 3:75787240-75787240 | G | missense\_variant | *ZNF717* | rs76111663 | deleterious(0) | probably\_damaging(0.999) | 23.8 | - |
| 3:75787405-75787405 | T | missense\_variant | *ZNF717* | rs73843014 | tolerated(0.09) | benign(0.137) | 23.6 | - |
| 3:75787464-75787464 | T | missense\_variant | *ZNF717* | rs74861398 | tolerated(0.92) | benign(0) | 1.979 | - |
| 3:75787927-75787927 | G | missense\_variant | *ZNF717* | rs75388892 | tolerated(0.1) | benign(0.141) | 9.841 | - |
| 3:75787996-75787996 | T | missense\_variant | *ZNF717* | rs78640256 | tolerated(0.37) | probably\_damaging(0.991) | 23.3 | - |
| 3:75788115-75788115 | A | missense\_variant | *ZNF717* | rs62246570 | tolerated(0.09) | benign(0.023) | 0.142 | - |
| 3:75788366-75788366 | A | missense\_variant | *ZNF717* | rs80085410 | tolerated(0.1) | benign(0.189) | 11.53 | - |
| X:50054209-50054209 | G | missense\_variant | *CCNB3* | - | tolerated(1) | benign(0) | 0.001 | - |
| X:55172687-55172687 | C | missense\_variant | *FAM104B* | rs1047037 | tolerated(0.15) | benign(0.013) | 0.019 | - |
| X:55172708-55172708 | G | missense\_variant | *FAM104B* | rs1047034 | deleterious(0.02) | benign(0.068) | 9.740 | - |

**Appendix B**: List of prioritized variants that had a minor allele frequency of less than 1% in public frequency databases and were present in a heterozygous state in all six of the affected individuals that underwent whole genome sequencing