

Table S2: Detailed calculations of carrier rates, likelihood of RDVs in two JBTS genes and disease prevalence in a recessive model based on ExAC data

MAF<0.2 and CADD>=15

| | joint allele frequencies per gene (~q) | CC2D2A | CEP290 | AHI1 | TMEM67 | c5orf42 | RPGRIP1L | INPP5E | CSPP1 | TMEM216 | TCTN1 | TCTN2 | TTC21B | KIF7 | TCTN3 | B9D1 | B9D2 | ARL13B | OFD1 | CEP41 | MKS1 | IFT172 | TMEM138 | TMEM231 | TMEM237 |
|--------------|--|--------------------|---------|----------|----------|----------|----------|-----------|----------|-----------|----------|----------|----------|--------|--------|----------|----------|----------|--------|--------|----------|----------|-----------|-----------|-----------|
| | | 0.0089 | 0.026 | 0.0072 | 0.006 | 0.0155 | 0.0125 | 0.0059 | 0.0111 | 0.002 | 0.006 | 0.0049 | 0.0151 | 0.023 | 0.0036 | 0.0086 | 0.0025 | 0.0079 | 0.0042 | 0.0029 | 0.0108 | 0.0235 | 0.0015 | 0.00877 | 0.0052 |
| | | carrier rate (~2q) | 0.0178 | 0.052 | 0.0144 | 0.012 | 0.031 | 0.025 | 0.0118 | 0.0222 | 0.004 | 0.012 | 0.0098 | 0.0302 | 0.046 | 0.0072 | 0.0172 | 0.005 | 0.0158 | 0.0084 | 0.0058 | 0.0216 | 0.047 | 0.003 | 0.01754 |
| CC2D2A | 0.0089 | 0.0178 | 0.00093 | 0.0003 | 0.00021 | 0.000552 | 0.000445 | 0.00021 | 0.0004 | 0.0000712 | 0.0002 | 0.0002 | 0.000538 | 8E-04 | 0.0001 | 0.0003 | 9E-05 | 0.000281 | 0.0001 | 0.0001 | 0.0004 | 0.0008 | 5.34E-05 | 0.000312 | 0.0001851 |
| CEP290 | 0.026 | 0.052 | 0.0007 | 0.00062 | 0.001612 | 0.0013 | 0.00061 | 0.0012 | 0.000208 | 0.0006 | 0.0005 | 0.00157 | 0.002 | 0.0004 | 0.0009 | 0.0003 | 0.000822 | 0.0004 | 0.0003 | 0.0011 | 0.0024 | 0.000156 | 0.000912 | 0.0005408 | |
| AHI1 | 0.0072 | 0.0144 | 0.00017 | 0.000446 | 0.00036 | 0.00017 | 0.0003 | 0.0000576 | 0.0002 | 0.0001 | 0.000435 | 7E-04 | 0.0001 | 0.0002 | 7E-05 | 0.000228 | 0.0001 | 8E-05 | 0.0003 | 0.0007 | 4.32E-05 | 0.000253 | 0.0001498 | | |
| TMEM67 | 0.006 | 0.012 | | 0.000372 | 0.0003 | 0.00014 | 0.0003 | 0.000048 | 0.0001 | 0.0001 | 0.000362 | 6E-04 | 9E-05 | 0.0002 | 6E-05 | 0.00019 | 0.0001 | 7E-05 | 0.0003 | 0.0006 | 0.000036 | 0.00021 | 0.0001248 | | |
| CSORF42 | 0.0155 | 0.031 | | | 0.000775 | 0.00037 | 0.0007 | 0.000124 | 0.0004 | 0.0003 | 0.000936 | 0.001 | 0.0002 | 0.0005 | 0.0002 | 0.00049 | 0.0003 | 0.0002 | 0.0007 | 0.0015 | 0.000093 | 0.000544 | 0.0003224 | | |
| RPGRIP1L | 0.0125 | 0.025 | | | | 0.0003 | 0.0006 | 0.0001 | 0.0003 | 0.0002 | 0.000755 | 0.001 | 0.0002 | 0.0004 | 0.0001 | 0.000395 | 0.0002 | 0.0001 | 0.0005 | 0.0012 | 0.000075 | 0.000439 | 0.00026 | | |
| INPP5E | 0.0059 | 0.0118 | | | | | 0.0003 | 0.0000472 | 0.0001 | 0.0001 | 0.000356 | 5E-04 | 8E-05 | 0.0002 | 6E-05 | 0.000186 | 1E-04 | 7E-05 | 0.0003 | 0.0006 | 3.54E-05 | 0.000207 | 0.0001227 | | |
| CSPP1 | 0.0111 | 0.0222 | | | | | | 0.0000888 | 0.0003 | 0.0002 | 0.00067 | 0.001 | 0.0002 | 0.0004 | 0.0001 | 0.000351 | 0.0002 | 0.0001 | 0.0005 | 0.001 | 6.66E-05 | 0.000389 | 0.0002309 | | |
| TMEM216 | 0.002 | 0.004 | | | | | | | 5E-05 | 4E-05 | 0.000121 | 2E-04 | 3E-05 | 7E-05 | 2E-05 | 6.32E-05 | 3E-05 | 2E-05 | 9E-05 | 0.0002 | 0.000012 | 7.02E-05 | 0.0000416 | | |
| TCTN1 | 0.006 | 0.012 | | | | | | | | | 0.0001 | 0.000362 | 6E-04 | 9E-05 | 0.0002 | 6E-05 | 0.00019 | 0.0001 | 7E-05 | 0.0003 | 0.0006 | 0.000036 | 0.00021 | 0.0001248 | |
| TCTN2 | 0.0049 | 0.0098 | | | | | | | | | | 0.000296 | 5E-04 | 7E-05 | 0.0002 | 5E-05 | 0.000155 | 8E-05 | 6E-05 | 0.0002 | 0.0005 | 2.94E-05 | 0.000172 | 0.0001019 | |
| TTC21B | 0.0151 | 0.0302 | | | | | | | | | | | 0.001 | 0.0002 | 0.0005 | 0.0002 | 0.000477 | 0.0003 | 0.0002 | 0.0007 | 0.0014 | 9.06E-05 | 0.00053 | 0.0003141 | |
| KIF7 | 0.023 | 0.046 | | | | | | | | | | | | 0.0003 | 0.0008 | 0.0002 | 0.000727 | 0.0004 | 0.0003 | 0.001 | 0.0022 | 0.000138 | 0.000807 | 0.0004784 | |
| TCTN3 | 0.0036 | 0.0072 | | | | | | | | | | | | | 0.0001 | 4E-05 | 0.000114 | 6E-05 | 4E-05 | 0.0002 | 0.0003 | 2.16E-05 | 0.000126 | 7.488E-05 | |
| B9D1 | 0.0086 | 0.0172 | | | | | | | | | | | | | | 9E-05 | 0.000272 | 0.0001 | 1E-04 | 0.0004 | 0.0008 | 5.16E-05 | 0.000302 | 0.0001789 | |
| B9D2 | 0.0025 | 0.005 | | | | | | | | | | | | | | | 0.000079 | 4E-05 | 3E-05 | 0.0001 | 0.0002 | 0.000015 | 8.77E-05 | 0.000052 | |
| ARL13B | 0.0079 | 0.0158 | | | | | | | | | | | | | | | | 0.0001 | 9E-05 | 0.0003 | 0.0007 | 4.74E-05 | 0.000277 | 0.0001643 | |
| OFD1 | 0.0042 | 0.0084 | | | | | | | | | | | | | | | | | 5E-05 | 0.0002 | 0.0004 | 2.52E-05 | 0.000147 | 8.736E-05 | |
| CEP41 | 0.0029 | 0.0058 | | | | | | | | | | | | | | | | | | 0.0001 | 0.0003 | 1.74E-05 | 0.000102 | 6.032E-05 | |
| MKS1 | 0.0108 | 0.0216 | | | | | | | | | | | | | | | | | | | 0.001 | 6.48E-05 | 0.000379 | 0.0002246 | |
| IFT172 | 0.0235 | 0.047 | | | | | | | | | | | | | | | | | | | | 0.000141 | 0.000824 | 0.0004888 | |
| TMEM138 | 0.0015 | 0.003 | | | | | | | | | | | | | | | | | | | | | 5.26E-05 | 0.0000312 | |
| TMEM231 | 0.00877 | 0.01754 | | | | | | | | | | | | | | | | | | | | | | 0.0001824 | |
| TMEM237 | 0.0052 | 0.0104 | | | | | | | | | | | | | | | | | | | | | | | |
| SUM per gene | | | 0.00093 | 0.001 | 0.00101 | 0.002982 | 0.00318 | 0.0018 | 0.0036 | 0.0007448 | 0.0023 | 0.002 | 0.006402 | 0.011 | 0.0021 | 0.0051 | 0.0016 | 0.005018 | 0.0028 | 0.002 | 0.0075 | 0.0174 | 0.001249 | 0.007353 | 0.0045421 |

Total SUM
0.0936
9.3616 %

disease prevalence calculation in a recessive mode

| | % disease (q ²) [h _{mz} or comp h _{tz}] |
|--------------------|---|
| CC2D2A | 0.00007921 |
| CEP290 | 0.000676 |
| AHI1 | 0.00005184 |
| TMEM67 | 0.000036 |
| CSORF42 | 0.00024025 |
| RPGRIP1L | 0.00015625 |
| INPP5E | 0.00003481 |
| CSPP1 | 0.00012321 |
| TMEM216 | 0.000004 |
| TCTN1 | 0.000036 |
| TCTN2 | 0.00002401 |
| TTC21B | 0.00022801 |
| KIF7 | 0.000529 |
| TCTN3 | 0.00001296 |
| B9D1 | 0.00007396 |
| B9D2 | 0.00000625 |
| ARL13B | 0.00006241 |
| OFD1 | 0.00001764 |
| CEP41 | 0.00000841 |
| MKS1 | 0.00011664 |
| IFT172 | 0.00055225 |
| TMEM138 | 0.00000225 |
| TMEM231 | 7.69129E-05 |
| TMEM237 | 0.00002704 |
| disease prevalence | 0.003175313 = approx 1/333 |

MAF<0.2 and CADD>=20

| | | CC2D2A | CEP290 | AHI1 | TMEM67 | c5orf42 | RPGRIP1L | INPP5E | CSPP1 | TMEM216 | TCTN1 | TCTN2 | TTC21B | KIF7 | TCTN3 | B9D1 | B9D2 | ARL13B | OFD1 | CEP41 | MKS1 | IFT172 | TMEM138 | TMEM231 | TMEM237 |
|--------------|--|--------|----------|-----------|-----------|------------|------------|------------|----------|------------|----------|--------|----------|---------|---------|------------|----------|------------|------------|------------|------------|------------|------------|------------|------------|
| | joint allele frequencies per gene (~q) | 0.0071 | 0.022 | 0.0045 | 0.0055 | 0.0117 | 0.01 | 0.0047 | 0.0089 | 0.0007 | 0.0053 | 0.003 | 0.012 | 0.02 | 0.0024 | 0.0051 | 0.002 | 0.0069 | 0.003 | 0.0023 | 0.007 | 0.015 | 0.0006 | 0.0075 | 0.0047 |
| | carrier rate (~2q) | 0.0142 | 0.044 | 0.009 | 0.011 | 0.0234 | 0.02 | 0.0094 | 0.0178 | 0.0014 | 0.0106 | 0.006 | 0.024 | 0.04 | 0.0048 | 0.0102 | 0.004 | 0.0138 | 0.006 | 0.0046 | 0.014 | 0.03 | 0.0012 | 0.015 | 0.0094 |
| CC2D2A | 0.0071 | 0.0142 | 0.000312 | 0.0000639 | 0.0000781 | 0.000332 | 0.000142 | 6.7E-05 | 0.000126 | 0.00000994 | 7.5E-05 | 4E-05 | 0.00017 | 3E-04 | 3E-05 | 7E-05 | 3E-05 | 9.8E-05 | 4E-05 | 0.00003266 | 9.9E-05 | 0.00021 | 0.00000852 | 0.000213 | 0.00006674 |
| CEP290 | 0.022 | 0.044 | | 0.000198 | 0.000242 | 0.00103 | 0.00044 | 0.00021 | 0.000392 | 0.0000308 | 0.00023 | 0.0001 | 0.000528 | 9E-04 | 0.0001 | 0.0002 | 9E-05 | 0.0003 | 0.0001 | 0.0001012 | 0.00031 | 0.00066 | 0.0000264 | 0.00066 | 0.0002068 |
| AHI1 | 0.0045 | 0.009 | | | 0.0000495 | 0.000211 | 0.00009 | 4.2E-05 | 8.01E-05 | 0.0000063 | 4.8E-05 | 3E-05 | 0.000108 | 2E-04 | 2E-05 | 5E-05 | 2E-05 | 6.2E-05 | 3E-05 | 0.0000207 | 6.3E-05 | 0.00014 | 0.0000054 | 0.000135 | 0.0000423 |
| TMEM67 | 0.0055 | 0.011 | | | 0.000257 | 0.00011 | 0.00011 | 5.2E-05 | 9.79E-05 | 0.0000077 | 5.8E-05 | 3E-05 | 0.000132 | 2E-04 | 3E-05 | 6E-05 | 2E-05 | 7.6E-05 | 3E-05 | 0.0000253 | 7.7E-05 | 0.00017 | 0.0000066 | 0.000165 | 0.0000517 |
| CSORF42 | 0.0117 | 0.0234 | | | 0.000234 | 0.00011 | 0.000208 | 0.00001638 | 0.00012 | 7E-05 | 0.000281 | 5E-04 | 6E-05 | 0.0001 | 5E-05 | 0.00016 | 7E-05 | 0.00005382 | 0.00016 | 0.00005382 | 0.00016 | 0.00035 | 0.00001404 | 0.000351 | 0.00010998 |
| RPGRIP1L | 0.01 | 0.02 | | | | 0.000178 | 0.000014 | 0.00011 | 6E-05 | 0.00024 | 4E-04 | 5E-05 | 0.0001 | 4E-05 | 0.00014 | 6E-05 | 0.000046 | 0.00014 | 0.0003 | 0.000012 | 0.0003 | 0.000012 | 0.0003 | 0.000094 | |
| INPP5E | 0.0047 | 0.0094 | | | | 8.37E-05 | 0.00000658 | 5E-05 | 3E-05 | 0.000113 | 2E-04 | 2E-05 | 5E-05 | 2E-05 | 2E-05 | 6.5E-05 | 3E-05 | 0.00002162 | 6.6E-05 | 0.00014 | 0.00000564 | 0.000141 | 0.00004418 | | |
| CSPP1 | 0.0089 | 0.0178 | | | | 0.00002492 | 9.4E-05 | 5E-05 | 0.000214 | 4E-04 | 4E-05 | 9E-05 | 4E-05 | 0.00012 | 5E-05 | 0.00004094 | 0.00012 | 0.000027 | 0.00001068 | 0.000267 | 0.00008366 | | | | |
| TMEM216 | 0.0007 | 0.0014 | | | | | 7.4E-06 | 4E-06 | 1.68E-05 | 3E-05 | 3E-06 | 7E-06 | 3E-06 | 9.7E-06 | 4E-06 | 0.00000322 | 9.8E-06 | 2.1E-05 | 0.00000084 | 0.000021 | 0.0000658 | | | | |
| TCTN1 | 0.0053 | 0.0106 | | | | | | 3E-05 | 0.000127 | 2E-04 | 3E-05 | 5E-05 | 2E-05 | 7.3E-05 | 3E-05 | 0.00002438 | 7.4E-05 | 0.000016 | 0.00000636 | 0.000159 | 0.00004982 | | | | |
| TCTN2 | 0.003 | 0.006 | | | | | | | 0.000072 | 1E-04 | 1E-05 | 3E-05 | 1E-05 | 4.1E-05 | 2E-05 | 0.0000138 | 4.2E-05 | 0.00009 | 0.0000036 | 0.00009 | 0.0000282 | | | | |
| TTC21B | 0.012 | 0.024 | | | | | | | | | | 5E-04 | 6E-05 | 0.0001 | 5E-05 | 0.00017 | 7E-05 | 0.0000552 | 0.00017 | 0.00036 | 0.000144 | 0.00036 | 0.0001128 | | |
| KIF7 | 0.02 | 0.04 | | | | | | | | | | | 1E-04 | 0.0002 | 8E-05 | 0.00028 | 0.0001 | 0.000092 | 0.00028 | 0.0006 | 0.000024 | 0.0006 | 0.000188 | | |
| TCTN3 | 0.0024 | 0.0048 | | | | | | | | | | | | | 2E-05 | 1E-05 | 3.3E-05 | 1E-05 | 0.00001104 | 3.4E-05 | 7.2E-05 | 0.0000288 | 0.000072 | 0.00002256 | |
| B9D1 | 0.0051 | 0.0102 | | | | | | | | | | | | | | 2E-05 | 7E-05 | 3E-05 | 0.00002346 | 7.1E-05 | 0.00015 | 0.00000612 | 0.000153 | 0.00004794 | |
| B9D2 | 0.002 | 0.004 | | | | | | | | | | | | | | | 2.8E-05 | 1E-05 | 0.0000092 | 2.8E-05 | 0.00006 | 0.0000024 | 0.00006 | 0.0000188 | |
| ARL13B | 0.0069 | 0.0138 | | | | | | | | | | | | | | | | 4E-05 | 0.00003174 | 9.7E-05 | 0.00021 | 0.00000828 | 0.000207 | 0.00006486 | |
| OFD1 | 0.003 | 0.006 | | | | | | | | | | | | | | | | | 0.0000138 | 4.2E-05 | 0.00009 | 0.0000036 | 0.00009 | 0.0000282 | |
| CEP41 | 0.007 | 0.0046 | | | | | | | | | | | | | | | | | | | 3.2E-05 | 6.9E-05 | 0.00000276 | 0.000069 | 0.00002162 |
| MKS1 | 0.0023 | 0.014 | | | | | | | | | | | | | | | | | | | 0.00021 | 0.0000084 | 0.00021 | 0.0000658 | |
| IFT172 | 0.015 | 0.03 | | | | | | | | | | | | | | | | | | | | 0.000018 | 0.00045 | 0.000141 | |
| TMEM138 | 0.0006 | 0.0012 | | | | | | | | | | | | | | | | | | | | | 0.000018 | 0.00000564 | |
| TMEM231 | 0.0075 | 0.015 | | | | | | | | | | | | | | | | | | | | | | 0.0000705 | |
| TMEM237 | 0.0047 | 0.0094 | | | | | | | | | | | | | | | | | | | | | | | |
| SUM per gene | | | 0.000312 | 0.0002619 | 0.0003696 | 0.00183 | 0.001016 | 0.00057 | 0.001166 | 0.00011662 | 0.0008 | 0.0005 | 0.002002 | 0.004 | 0.0006 | 0.0012 | 0.0005 | 0.00172 | 0.0008 | 0.00062008 | 0.00192 | 0.00432 | 0.00019092 | 0.004791 | 0.00157168 |

Total SUM
0.0309
3.0917 %

disease prevalence calculation in a recessive mode

| | disease (q2) [hmz or comp htz] |
|----------|--------------------------------|
| CC2D2A | 0.00005041 |
| CEP290 | 0.000484 |
| AHI1 | 0.00002025 |
| TMEM67 | 0.00003025 |
| CSORF42 | 0.00013689 |
| RPGRIP1L | 0.0001 |
| INPP5E | 0.00002209 |
| CSPP1 | 0.00007921 |
| TMEM216 | 0.00000049 |
| TCTN1 | 0.00002809 |
| TCTN2 | 0.000009 |
| TTC21B | 0.000144 |
| KIF7 | 0.0004 |
| TCTN3 | 0.00000576 |
| B9D1 | 0.00002601 |
| B9D2 | 0.000004 |
| ARL13B | 0.00004761 |
| OFD1 | 0.000009 |
| CEP41 | 0.000049 |
| MKS1 | 0.00000529 |
| IFT172 | 0.000225 |
| TMEM138 | 0.00000036 |
| TMEM231 | 0.00005625 |
| TMEM237 | 0.00002209 |

disease prevalence 0.00195505 = approx 1/500

MAF<0.2 and CADD>=25

| | | CC2D2A | CEP290 | AH1 | TMEM67 | c5orf42 | RPGRIP1L | INPP5E | CSPP1 | TMEM216 | TCTN1 | TCTN2 | TTC21B | KIF7 | TCTN3 | B9D1 | B9D2 | ARL13B | OFD1 | CEP41 | MKS1 | IFT172 | TMEM138 | TMEM231 | TMEM237 |
|--|--------|--------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|------------|
| joint allele frequencies per gene (~q) | | 0.0037 | 0.015 | 0.0027 | 0.0032 | 0.006 | 0.0054 | 0.0023 | 0.0049 | 0.0004 | 0.0033 | 0.0012 | 0.0076 | 0.012 | 0.0011 | 0.0015 | 0.0009 | 0.0017 | 0.0017 | 0.0016 | 0.0049 | 0.0096 | 0.0003 | 0.0039 | 0.0019 |
| carrier rate (~2q) | | 0.0074 | 0.03 | 0.0054 | 0.0064 | 0.012 | 0.0108 | 0.0046 | 0.0098 | 0.0008 | 0.0066 | 0.0024 | 0.0152 | 0.024 | 0.0022 | 0.003 | 0.0018 | 0.0034 | 0.0034 | 0.0032 | 0.0098 | 0.0192 | 0.0006 | 0.0078 | 0.0038 |
| CC2D2A | 0.0037 | 0.0074 | 0.000111 | 2E-05 | 0.00002368 | 8.88E-05 | 4E-05 | 1.7E-05 | 3.63E-05 | 2.96E-06 | 2.44E-05 | 8.88E-06 | 5.62E-05 | 8.88E-05 | 8.14E-06 | 1.11E-05 | 6.66E-06 | 1.26E-05 | 1.26E-05 | 1.18E-05 | 3.63E-05 | 7.1E-05 | 2.22E-06 | 5.77E-05 | 0.00002812 |
| CEP290 | 0.015 | 0.03 | 0.000081 | 0.000096 | 0.000036 | 0.000162 | 0.000069 | 0.000147 | 0.000012 | 0.000099 | 0.000036 | 0.000228 | 0.00036 | 0.000033 | 0.000045 | 0.000027 | 0.000051 | 0.000051 | 0.000048 | 0.000147 | 0.000288 | 0.000009 | 0.000234 | 0.000114 | 0.000114 |
| AH1 | 0.0027 | 0.0054 | 0.00001728 | 6.48E-05 | 2.92E-05 | 1.24E-05 | 2.65E-05 | 2.16E-06 | 1.78E-05 | 6.48E-06 | 4.1E-05 | 6.48E-05 | 5.94E-06 | 8.1E-06 | 4.86E-06 | 9.18E-06 | 9.18E-06 | 8.64E-06 | 2.65E-05 | 5.18E-05 | 1.62E-06 | 4.21E-05 | 0.00002052 | 0.00002052 | 0.00002052 |
| TMEM67 | 0.0032 | 0.0064 | 7.68E-05 | 3.46E-05 | 1.47E-05 | 3.14E-05 | 2.56E-06 | 2.11E-05 | 7.68E-06 | 4.86E-05 | 7.68E-05 | 7.04E-06 | 9.6E-06 | 5.76E-06 | 1.09E-05 | 1.09E-05 | 1.02E-05 | 3.14E-05 | 6.14E-05 | 1.92E-06 | 4.99E-05 | 0.00002432 | 0.00002432 | 0.00002432 | 0.00002432 |
| CSORF42 | 0.006 | 0.012 | 6.48E-05 | 2.76E-05 | 5.88E-05 | 4.8E-06 | 3.96E-05 | 1.44E-05 | 9.12E-05 | 0.000144 | 1.32E-05 | 0.000018 | 1.08E-05 | 2.04E-05 | 2.04E-05 | 1.92E-05 | 5.88E-05 | 0.000115 | 3.6E-06 | 9.36E-05 | 0.0000456 | 0.0000456 | 0.0000456 | 0.0000456 | 0.0000456 |
| RPGRIP1L | 0.0054 | 0.0108 | 2.48E-05 | 5.29E-05 | 4.32E-06 | 3.56E-05 | 1.3E-05 | 8.21E-05 | 0.00013 | 1.19E-05 | 1.62E-05 | 9.72E-06 | 1.84E-05 | 1.84E-05 | 1.73E-05 | 5.29E-05 | 0.000104 | 3.24E-06 | 8.42E-05 | 0.00004104 | 0.00004104 | 0.00004104 | 0.00004104 | 0.00004104 | 0.00004104 |
| INPP5E | 0.0023 | 0.0046 | 1.84E-06 | 1.52E-05 | 5.52E-06 | 3.5E-05 | 5.52E-05 | 5.06E-06 | 6.9E-06 | 4.14E-06 | 7.82E-06 | 7.36E-06 | 2.25E-05 | 4.42E-05 | 1.38E-06 | 3.59E-05 | 0.00001748 | 0.00001748 | 0.00001748 | 0.00001748 | 0.00001748 | 0.00001748 | 0.00001748 | 0.00001748 | 0.00001748 |
| CSPP1 | 0.0049 | 0.0098 | 7.84E-06 | 3.23E-05 | 1.18E-05 | 7.45E-05 | 0.000118 | 1.08E-05 | 1.47E-05 | 8.82E-06 | 1.67E-05 | 1.67E-05 | 1.57E-05 | 4.8E-05 | 9.41E-05 | 2.94E-06 | 7.64E-05 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 |
| TMEM216 | 0.0004 | 0.0008 | 2.64E-06 | 9.6E-07 | 6.08E-06 | 9.6E-06 | 8.8E-07 | 1.2E-06 | 7.2E-07 | 1.36E-06 | 1.36E-06 | 1.28E-06 | 3.92E-06 | 7.68E-06 | 2.4E-07 | 6.24E-06 | 0.0000304 | 0.0000304 | 0.0000304 | 0.0000304 | 0.0000304 | 0.0000304 | 0.0000304 | 0.0000304 | 0.0000304 |
| TCTN1 | 0.0033 | 0.0066 | 7.92E-06 | 5.02E-05 | 7.92E-05 | 7.26E-06 | 9.9E-06 | 5.94E-06 | 1.12E-05 | 1.12E-05 | 1.06E-05 | 3.23E-05 | 6.34E-05 | 1.98E-06 | 5.15E-05 | 0.00002508 | 0.00002508 | 0.00002508 | 0.00002508 | 0.00002508 | 0.00002508 | 0.00002508 | 0.00002508 | 0.00002508 | 0.00002508 |
| TCTN2 | 0.0012 | 0.0024 | 1.82E-05 | 2.88E-05 | 2.64E-06 | 3.6E-06 | 2.16E-06 | 4.08E-06 | 4.08E-06 | 3.84E-06 | 1.18E-05 | 2.3E-05 | 7.2E-07 | 1.87E-05 | 0.0000912 | 0.0000912 | 0.0000912 | 0.0000912 | 0.0000912 | 0.0000912 | 0.0000912 | 0.0000912 | 0.0000912 | 0.0000912 | 0.0000912 |
| TTC21B | 0.0076 | 0.0152 | 0.000182 | 1.67E-05 | 2.28E-05 | 1.37E-05 | 2.58E-05 | 2.43E-05 | 7.45E-05 | 0.000146 | 4.56E-06 | 0.000119 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 |
| KIF7 | 0.012 | 0.024 | 0.000182 | 1.67E-05 | 2.28E-05 | 1.37E-05 | 2.58E-05 | 2.43E-05 | 7.45E-05 | 0.000146 | 4.56E-06 | 0.000119 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 |
| TCTN3 | 0.0011 | 0.0022 | 2.64E-05 | 0.000036 | 2.16E-05 | 2.58E-05 | 2.43E-05 | 7.45E-05 | 0.000146 | 4.56E-06 | 0.000119 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 | 0.00005776 |
| B9D1 | 0.0015 | 0.003 | 3.3E-06 | 1.98E-06 | 3.74E-06 | 3.74E-06 | 3.52E-06 | 1.08E-05 | 3.74E-06 | 3.52E-06 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 |
| B9D2 | 0.0009 | 0.0018 | 3.3E-06 | 1.98E-06 | 3.74E-06 | 3.74E-06 | 3.52E-06 | 1.08E-05 | 3.74E-06 | 3.52E-06 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 | 1.08E-05 |
| ARL13B | 0.0017 | 0.0034 | 3.06E-06 | 3.06E-06 | 2.88E-06 | 8.82E-06 | 1.73E-05 | 5.4E-07 | 1.4E-05 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 | 0.0000684 |
| OFD1 | 0.0017 | 0.0034 | 5.78E-06 | 5.44E-06 | 1.67E-05 | 3.26E-05 | 1.02E-06 | 2.65E-05 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 |
| CEP41 | 0.0049 | 0.0098 | 5.44E-06 | 1.67E-05 | 3.26E-05 | 1.02E-06 | 2.65E-05 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 | 0.0001292 |
| MKS1 | 0.0016 | 0.0032 | 1.57E-05 | 3.07E-05 | 9.6E-07 | 2.5E-05 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 | 0.00001216 |
| IFT172 | 0.0096 | 0.0192 | 9.41E-05 | 2.94E-06 | 7.64E-05 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 | 0.00003724 |
| TMEM138 | 0.0003 | 0.0006 | 5.76E-06 | 0.00015 | 5.76E-06 | 4.68E-06 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 |
| TMEM231 | 0.0039 | 0.0078 | 4.68E-06 | 0.0000228 | 4.68E-06 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 | 0.0000228 |
| TMEM237 | 0.0019 | 0.0038 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 | 0.00002964 |
| SUM per gene | | | 0.000111 | 0.000101 | 0.00013696 | 0.00059 | 0.00033 | 0.000166 | 0.000375 | 3.85E-05 | 0.000288 | 0.000113 | 0.000731 | 0.001337 | 0.000149 | 0.000206 | 0.000127 | 0.000242 | 0.000248 | 0.000239 | 0.000747 | 0.001557 | 5.44E-05 | 0.00142 | 0.00072124 |

TOTAL SUM
0.010027
1.002716 %

disease prevalence calculation in a recessive mode

| | disease (q2) [hmz or comp htz] |
|----------|--------------------------------|
| CC2D2A | 0.00001369 |
| CEP290 | 0.000225 |
| AH1 | 0.00000729 |
| TMEM67 | 0.00001024 |
| CSORF42 | 0.000036 |
| RPGRIP1L | 0.00002916 |
| INPP5E | 0.0000529 |
| CSPP1 | 0.00002401 |
| TMEM216 | 0.00000016 |
| TCTN1 | 0.00001089 |
| TCTN2 | 0.00000144 |
| TTC21B | 0.00005776 |
| KIF7 | 0.000144 |
| TCTN3 | 0.00000121 |
| B9D1 | 0.00000225 |
| B9D2 | 0.00000081 |
| ARL13B | 0.00000289 |
| OFD1 | 0.00000289 |
| CEP41 | 0.00002401 |
| MKS1 | 0.00000256 |
| IFT172 | 0.00009216 |
| TMEM138 | 0.00000009 |
| TMEM231 | 0.00001521 |
| TMEM237 | 0.00000361 |

disease prevalence 0.00071262 = approx 1/1500

MAF<0.2 and truncating RDVs (stop-gain, frameshift, canonical splice)

| | | CC2D2A | CEP290 | AHI1 | TMEM67 | cSorF42 | RPGRIP1L | INPP5E | CSPP1 | TMEM216 | TCTN1 | TCTN2 | TTC21B | KIF7 | TCTN3 | B9D1 | B9D2 | ARL13B | OFD1 | CEP41 | MKS1 | IFT172 | TMEM138 | TMEM231 | TMEM237 |
|--------------|--|------------|----------|---------|-----------|-----------|----------|----------|-----------|-----------|----------|----------|----------|----------|----------|---------|----------|----------|---------|----------|----------|-----------|-----------|------------|------------|
| | joint allele frequencies per gene (~q) | 0.001868 | 0.003644 | 0.00068 | 0.00149 | 0.0015975 | 0.000832 | 5.86E-05 | 0.00103 | 5.996E-05 | 0.000649 | 0.000285 | 0.001015 | 0.00058 | 0.00033 | 7.7E-05 | 3E-05 | 0.00012 | 0.00415 | 0.0001 | 0.000305 | 0.00057 | 1.499E-05 | 0.0006 | 0.00042 |
| | carrier rate (~2q) | 0.003737 | 0.007288 | 0.00136 | 0.002981 | 0.003195 | 0.001664 | 0.000117 | 0.00207 | 0.0001199 | 0.001299 | 0.00057 | 0.002029 | 0.00116 | 0.00066 | 0.00015 | 6E-05 | 0.00024 | 0.0083 | 0.0003 | 0.000611 | 0.00115 | 2.998E-05 | 0.0012 | 0.00084 |
| CC2D2A | 0.00186841 | 0.00373682 | 1.36E-05 | 2.5E-06 | 5.57E-06 | 1.194E-05 | 3.11E-06 | 2.19E-07 | 3.9E-06 | 2.241E-07 | 2.43E-06 | 1.06E-06 | 3.79E-06 | 2.2E-06 | 1.23E-06 | 2.9E-07 | 1E-07 | 4.48E-07 | 1.6E-05 | 5E-07 | 1.14E-06 | 2.1E-06 | 5.601E-08 | 4.484E-06 | 3.1389E-06 |
| CEP290 | 0.00364412 | 0.00728824 | | 5E-06 | 1.09E-05 | 2.329E-05 | 6.06E-06 | 4.27E-07 | 7.5E-06 | 4.37E-07 | 4.73E-06 | 2.08E-06 | 7.4E-06 | 4.2E-06 | 2.41E-06 | 5.6E-07 | 2E-07 | 8.75E-07 | 3E-05 | 1E-06 | 2.23E-06 | 4.2E-06 | 1.093E-07 | 8.746E-06 | 6.1221E-06 |
| AHI1 | 0.00068 | 0.00136 | | | 2.03E-06 | 4.345E-06 | 1.13E-06 | 7.96E-08 | 1.4E-06 | 8.155E-08 | 8.83E-07 | 3.87E-07 | 1.38E-06 | 7.9E-07 | 4.49E-07 | 1E-07 | 4E-08 | 1.63E-07 | 5.6E-06 | 2E-07 | 4.15E-07 | 7.8E-07 | 2.039E-08 | 1.632E-06 | 1.1424E-06 |
| TMEM67 | 0.00149042 | 0.00298084 | | | 9.524E-06 | 2.48E-06 | 1.75E-07 | 3.1E-06 | 1.787E-07 | 1.94E-06 | 8.49E-07 | 3.02E-06 | 1.7E-06 | 9.84E-07 | 2.3E-07 | 9E-08 | 3.58E-07 | 1.2E-05 | 4E-07 | 9.1E-07 | 1.7E-06 | 4.468E-08 | 3.577E-06 | 2.5039E-06 | |
| CSORF42 | 0.00159748 | 0.00319496 | | | | 2.66E-06 | 1.87E-07 | 3.3E-06 | 1.916E-07 | 2.07E-06 | 9.1E-07 | 3.24E-06 | 1.9E-06 | 1.05E-06 | 2.4E-07 | 1E-07 | 3.83E-07 | 1.3E-05 | 5E-07 | 9.76E-07 | 1.8E-06 | 4.789E-08 | 3.834E-06 | 2.6838E-06 | |
| RPGRIP1L | 0.00083184 | 0.00166368 | | | | | 9.74E-08 | 1.7E-06 | 9.975E-08 | 1.08E-06 | 4.74E-07 | 1.69E-06 | 9.7E-07 | 5.49E-07 | 1.3E-07 | 5E-08 | 2E-07 | 6.9E-06 | 2E-07 | 5.08E-07 | 9.6E-07 | 2.494E-08 | 1.996E-06 | 1.3975E-06 | |
| INPP5E | 0.00005856 | 0.00011712 | | | | | | 1.2E-07 | 7.023E-09 | 7.6E-08 | 3.34E-08 | 1.19E-07 | 6.8E-08 | 3.87E-08 | 9E-09 | 4E-09 | 1.41E-08 | 4.9E-07 | 2E-08 | 3.58E-08 | 6.7E-08 | 1.756E-09 | 1.405E-07 | 9.8381E-08 | |
| CSPP1 | 0.00103333 | 0.00206666 | | | | | | | 2.478E-07 | 1.34E-06 | 5.89E-07 | 2.1E-06 | 1.2E-06 | 6.82E-07 | 1.6E-07 | 6E-08 | 2.48E-07 | 8.6E-06 | 3E-07 | 6.31E-07 | 1.2E-06 | 3.098E-08 | 2.48E-06 | 1.736E-06 | |
| TMEM216 | 0.00005996 | 0.00011992 | | | | | | | | 7.79E-08 | 3.42E-08 | 1.22E-07 | 7E-08 | 3.96E-08 | 9.2E-09 | 4E-09 | 1.44E-08 | 5E-07 | 2E-08 | 3.66E-08 | 6.9E-08 | 1.798E-09 | 1.439E-07 | 1.0073E-07 | |
| TCTN1 | 0.0006493 | 0.0012986 | | | | | | | | | 3.7E-07 | 1.32E-06 | 7.5E-07 | 4.29E-07 | 9.9E-08 | 4E-08 | 1.56E-07 | 5.4E-06 | 2E-07 | 3.97E-07 | 7.5E-07 | 1.947E-08 | 1.558E-06 | 1.0908E-06 | |
| TCTN2 | 0.00028487 | 0.00056974 | | | | | | | | | | 5.78E-07 | 3.3E-07 | 1.88E-07 | 4.4E-08 | 2E-08 | 6.84E-08 | 2.4E-06 | 8E-08 | 1.74E-07 | 3.3E-07 | 8.54E-09 | 6.837E-07 | 4.7858E-07 | |
| TTC21B | 0.00101473 | 0.00202946 | | | | | | | | | | | 1.2E-06 | 6.7E-07 | 1.6E-07 | 6E-08 | 2.44E-07 | 8.4E-06 | 3E-07 | 6.2E-07 | 1.2E-06 | 3.042E-08 | 2.435E-06 | 1.7047E-06 | |
| KIF7 | 0.00058106 | 0.00116212 | | | | | | | | | | | | 3.84E-07 | 8.9E-08 | 3E-08 | 1.39E-07 | 4.8E-06 | 2E-07 | 3.55E-07 | 6.7E-07 | 1.742E-08 | 1.395E-06 | 9.7618E-07 | |
| TCTN3 | 0.00033008 | 0.00066016 | | | | | | | | | | | | | 5.1E-08 | 2E-08 | 7.92E-08 | 2.7E-06 | 9E-08 | 2.02E-07 | 3.8E-07 | 9.896E-09 | 7.922E-07 | 5.5453E-07 | |
| B9D1 | 0.00007661 | 0.00015322 | | | | | | | | | | | | | | 5E-09 | 1.84E-08 | 6.4E-07 | 2E-08 | 4.68E-08 | 8.8E-08 | 2.297E-09 | 1.839E-07 | 1.287E-07 | |
| B9D2 | 0.00003 | 0.00006 | | | | | | | | | | | | | | | | 7.2E-09 | 2.5E-07 | 8E-09 | 1.83E-08 | 3.4E-08 | 8.994E-10 | 7.2E-08 | 5.04E-08 |
| ARL13B | 0.00012 | 0.00024 | | | | | | | | | | | | | | | | | 1E-06 | 3E-08 | 7.33E-08 | 1.4E-07 | 3.598E-09 | 2.88E-07 | 2.016E-07 |
| OFD1 | 0.00414872 | 0.00829744 | | | | | | | | | | | | | | | | | | 1E-06 | 2.53E-06 | 4.8E-06 | 1.244E-07 | 9.957E-06 | 6.9698E-06 |
| CEP41 | 0.00030539 | 0.00028312 | | | | | | | | | | | | | | | | | | | 8.65E-08 | 1.6E-07 | 4.244E-09 | 3.397E-07 | 2.3782E-07 |
| MKS1 | 0.00014156 | 0.00061078 | | | | | | | | | | | | | | | | | | | | 3.5E-07 | 9.156E-09 | 7.329E-07 | 5.1306E-07 |
| IFT172 | 0.00057409 | 0.00114818 | | | | | | | | | | | | | | | | | | | | | 1.721E-08 | 1.378E-06 | 9.6447E-07 |
| TMEM138 | 0.00001499 | 0.00002998 | | | | | | | | | | | | | | | | | | | | | | 3.598E-08 | 2.5183E-08 |
| TMEM231 | 0.0006 | 0.0012 | | | | | | | | | | | | | | | | | | | | | | | 1.008E-06 |
| TMEM237 | 0.00042 | 0.00084 | | | | | | | | | | | | | | | | | | | | | | | |
| SUM per gene | | | 1.36E-05 | 7.5E-06 | 1.85E-05 | 4.909E-05 | 1.54E-05 | 1.18E-06 | 2.1E-05 | 1.468E-06 | 1.46E-05 | 6.79E-06 | 2.48E-05 | 1.5E-05 | 9.11E-06 | 2.2E-06 | 9E-07 | 3.42E-06 | 0.00012 | 5E-06 | 1.14E-05 | 2.2E-05 | 5.852E-07 | 4.689E-05 | 3.3828E-05 |

TOTAL SUM
0.00044
0.04436 %

disease prevalence calculation in a recessive mode

| | disease (q2) [hmz or comp htz] |
|----------|--------------------------------|
| CC2D2A | 3.49096E-06 |
| CEP290 | 1.32796E-05 |
| AHI1 | 4.624E-07 |
| TMEM67 | 2.22135E-06 |
| CSORF42 | 2.55194E-06 |
| RPGRIP1L | 6.91958E-07 |
| INPP5E | 3.42927E-09 |
| CSPP1 | 1.06777E-06 |
| TMEM216 | 3.5952E-09 |
| TCTN1 | 4.2159E-07 |
| TCTN2 | 8.11509E-08 |
| TTC21B | 1.02968E-06 |
| KIF7 | 3.37631E-07 |
| TCTN3 | 1.08953E-07 |
| B9D1 | 5.86909E-09 |
| B9D2 | 9E-10 |
| ARL13B | 1.44E-08 |
| OFD1 | 1.72119E-05 |
| CEP41 | 9.32631E-08 |
| MKS1 | 2.00392E-08 |
| IFT172 | 3.29579E-07 |
| TMEM138 | 2.247E-10 |
| TMEM231 | 0.00000036 |
| TMEM237 | 1.764E-07 |

disease prevalence 4.39646E-05 = approx 1/25'000