|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  |  |  |  | **Strong GoF**  |  | **Other** |  |  |
|  |  |  Total  alleles |  |  L223P |  Δ229-235 |  Δ249-252 |  R267W |  V331E |  E340K |  T385A |  Combined |  |  G35E |  T39M |  L76fs |  V88M |  V112I |  R131G |  A210V |  A240E |  R244S |  A250T |  A275V |  A301S |  R310H |  N337D |  A341V |  Combined |  |  **P332Q** † |
| **Cases** |  MinE DF1 | 8,732 |  | 1 | 2 | 1 | - | 1 | 1 | 1 | 7 |  | - | - | - | - | 1 | - | - | 1 | 1 | 1 | 1 | 3 | 1 | 3 | 2 | 14 |  | 107 |
|  Answer ALS | 1,412 |  | - | 1 | - | 1 | - | - | - | 2 |  | 1 | - | - | - | - | 1 | - | - | - | - | - | 1 | - | - | - | 3 |  | 16 |
|  MinE DF2+\* | 3,176 |  | - | - | - | - | - | - | 1 | 1 |  | - | - | - | - | - | - | - | - | - | - | - | - | 1 | - | - | 1 |  | 40 |
|  GENESIS | 5,936 |  | - | 2 | - | - | - | - | - | 2 |  | - | - | - | - | - | - | - | - | - | - | - | - | - | - | 1 | 1 |  | 15 |
|  UCL | 1,366 |  | - | - | - | 1 | - | - | - | 1 |  | - | - | - | - | - | - | - | - | - | - | - | 2 | - | - | - | 2 |  | 5 |
|  HSP study | 1,612 |  | - | - | - | - | - | - | - | 0 |  | - | - | - | - | - | - | - | - | - | - | - | - | - | - | - | 0 |  | 15 |
|  Total | **22,234** |  | 1 | 5 | 1 | 2 | 1 | 1 | 2 | **13** |  | 1 | 0 | 0 | 0 | 1 | 1 | 0 | 1 | 1 | 1 | 1 | 6 | 2 | 3 | 3 | **21** |  | 198 |
|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| **Controls** |  MinE DF1 | 3,664 |  | - | - | - | - | - | - | - | 0 |  | - | 1 | 1 | 1 | - | - | - | - | - | - | - | - | - | 1 | - | 4 |  | 47 |
|  Answer ALS | 184 |  | - | - | - | - | - | - | - | 0 |  | - | - | - | - | - | - | 1 | - | - | - | - | - | - | - | - | 1 |  | 3 |
|  MinE DF2+\* | 812 |  | - | - | - | - | - | - | - | 0 |  | - | - | - | - | - | - | - | - | - | - | - | - | - | - | 1 | 1 |  | 15 |
|  GENESIS | 7,330 |  | - | - | - | - | - | - | - | 0 |  | - | - | - | - | - | - | - | - | - | - | 1 | - | - | 1 | 1 | 3 |  | 58 |
|  UCL | 3,616 |  | - | - | - | - | - | - | - | 0 |  | - | - | - | - | - | - | - | - | - | - | 1 | 1 | 2 | - | - | 4 |  | 38 |
|  HSP study | 2,490 |  | - | - | - | - | - | - | - | 0 |  | - | - | - | - | - | - | - | - | - | - | - | - | - | - | - | 0 |  | 19 |
|  LBC | 2,708 |  | - | - | - | - | - | - | - | 0 |  | - | - | - | - | - | - | - | - | - | - | - | - | - | 2 | - | 2 |  | 26 |
|  Total | **20,804** |  | 0 | 0 | 0 | 0 | 0 | 0 | 0 | **0** |  | 0 | 1 | 1 | 1 | 0 | 0 | 1 | 0 | 0 | 0 | 2 | 1 | 2 | 4 | 2 | **15** |  | 206 |