



**D**

| Allele | Mutation(s)                                  | Change in amino acid sequence (full length=96 aa)          |
|--------|--|--|
| 1-1    | Del 1229 (-1 bp)                             | starting at 21, early stop at 45                           |
| 6-1    | Del 1231-1237 (-7 bp)                        | starting at 21, early stop at 43                           |
| 4-1    | Del 1229-1233 (-5 bp)                        | starting at 20, early stop at 80                           |
| 5-1    | Del 1204-1229 (-26 bp)                       | starting at 12, early stop at 73                           |
| 5-3    | Del 1216-1233 (-18 bp), Ins 1254 C (+1 bp)   | deletion of 16-21, change starting at 30, early stop at 76 |
| 6-3    | Del 229-1238 (-1010 bp), Ins 229 CCT (+3 bp) | no protein or truncated protein missing N-terminus         |
| 6-2    | Del 229-1238 (-1010 bp)                      | no protein or truncated protein missing N-terminus         |
| 3-1    | Del 233-1240 (-1008 bp)                      | no protein or truncated protein missing N-terminus         |