

| serie number | mutation<br>type_cat | studies reported | <i>n</i> cases | exon    |
|--------------|----------------------|------------------|----------------|---------|
| 1            | A990V                | 1                | qualitative    | 20      |
| 2            | A1046                | 1                | qualitative    | 20      |
| 3            | A533T                | 1                | qualitative    | 9       |
| 4            | A994A                | 1                | 1              | 20      |
| 5            | C2985T               | 1                | 1              | 9       |
| 6            | C535P                | 1                | qualitative    | 9       |
| 7            | D1056G               | 1                | qualitative    | 20      |
| 8            | D549                 | 1                | qualitative    | 9       |
| 9            | E542G                | 1                | 1              | 9       |
| 10           | E542K                | 9                | 21             | 9       |
| 11           | E522V                | 1                | qualitative    | 9       |
| 12           | E545K                | 15               | 31             | 9       |
| 13           | E545G                | 1                | qualitative    | 9       |
| 14           | E545Q                | 1                | qualitative    | 9       |
| 15           | F1016L               | 1                | qualitative    | 20      |
| 16           | F550L                | 1                | 1              | 9       |
| 17           | F998S                | 1                | qualitative    | 20      |
| 18           | G1049                | 3                | 2              | 20      |
| 19           | H1047L               | 2                | qualitative    | 20      |
| 20           | H1047R               | 9                | 16             | 20      |
| 21           | H1047Y               | 1                | qualitative    | 20      |
| 22           | H1048                | 1                | qualitative    | 20      |
| 23           | H1065R               | 1                | qualitative    | 20      |
| 24           | H994Y                | 1                | qualitative    | 20      |
| 25           | I1062T               | 1                | qualitative    | 20      |
| 26           | M1040V               | 1                | 1              | 20      |
| 27           | M1043I               | 2                | 8              | 20      |
| 28           | M1043V               | 1                | qualitative    | 20      |
| 29           | N1068S               | 1                | 1              | 20      |
| 30           | p.A1066T             | 0                | qualitative    | 20      |
| 31           | Q1033                | 0                | qualitative    | 20      |
| 32           | Q1064                | 0                | qualitative    | 20      |
| 33           | Q546L                | 0                | qualitative    | 9       |
| 34           | Q981                 | 0                | qualitative    | 20      |
| 35           | R516K                | 1                | 1              | 9       |
| 36           | S1008P               | 0                | qualitative    | 20      |
| 37           | S1015S               | 0                | qualitative    | 20      |
| 38           | S541S                | 1                | 1              | 9       |
| 39           | T1025T               | 2                | 16             | 20      |
| 40           | T1053A               | 1                | 1              | 20      |
| 41           | T1061A               | 0                | qualitative    | 20      |
| 42           | T5441I               | 0                | qualitative    | 9       |
| 43           | W1057                | 0                | qualitative    | 20      |
| 44           | Y1021Y               | 1                | 1              | 20      |
| 45           | Y343C                | 1                | 1              | no data |