| Author, year         | Nucleotide change | Exon | Age | (Refs.) |
|----------------------|-------------------|------|-----|---------|
| Yang et al, 2016     | c.6992G>A         | 56   | 35  | (23)    |
|                      | c.1285C>T         | 10   | 47  |         |
|                      | c.1715-1G>A       | 13-14| 31  |         |
|                      | c.6037+2T>G       | 48-49| 60  |         |
|                      | c.2963G>A         | 24   | 35  |         |
|                      | c.8038delC        | 63   | 45  |         |
|                      | c.6452G>T         | 52   | 31  |         |
|                      | c.3995delA        | 32   | 16  |         |
|                      | c.5434T>C         | 44   | 45  |         |
|                      | c.7180C>T         | 57   | 25  |         |
|                      | c.3463+1G>T       | 27-28| 40  |         |
|                      | c.6388G>A         | 52   | 11  |         |
| Chung et al, 2009    | c.64delA          | 1    | 30  | (22)    |
|                      | c.169A>G          | 2    | 16  |         |
|                      | c.299G>A          | 3    | 13  |         |
|                      | c.386G>A          | 4    | 19  |         |
|                      | c.1441_1442delCA  | 11   | 12  |         |
|                      | c.2581 C>T        | 21   | 25  |         |
|                      | c.2728G>C         | 22   | 18  |         |
|                      | c.2763T>A         | 23   | 21  |         |
|                      | c.3389A>C         | 27   | 8   |         |
|                      | c.3658delIG       | 29   | 38  |         |
|                      | c.3667_3670delTGTC| 29   | 19  |         |
|                      | c.5368C>T        | 43   | 28  |         |
|                      | c.5434T>C        | 33   | 10  |         |
|                      | c.5477A>G        | 33   | 16  |         |
|                      | c.6252C>G        | 50   | 46  |         |
|                      | c.6388G>A        | 52   | 15  |         |
|                      | c.6431A>G        | 52   | 5   |         |
|                      | c.6892A>T        | 56   | 19  |         |
|                      | c.7565G>A        | 60   | 11  |         |
|                      | c.7826dupA       | 63   | 20  |         |
|                      | c.7975delIG      | 63   | 6   |         |
|                      | c.8123A>G        | 64   | 22  |         |