

**Supplementary Table 3 – Summary of mutations published in *Human Mutation* sampled in HGMD at three time points**

		May 2004			May 2005			January 2007			January 2007 (commercial)		
	Issue	1	2	3	1	2	3	1	2	3	1	2	3
12/2006	27 (12)	-	-	-	-	-	-	-	-	-	-	-	-
	27 (11)	-	-	-	-	-	-		▼			▼	✓
	27 (10)	-	-	-	-	-	-		○		✓	○	✓
	27 (9)	-	-	-	-	-	-		○		✓	○	○
	27 (8)	-	-	-	-	-	-				✓	✓	✓
	27 (7)	-	-	-	-	-	-				✓		✓
	27 (6)	-	-	-	-	-	-		○		✓	○	✓
	27 (5)	-	-	-	-	-	-	-	-	-	-	-	-
	27 (4)	-	-	-	-	-	-				✓	✓	✓
	27 (3)	-	-	-	-	-	-				✓	✓	✓
	27 (2)	-	-	-	-	-	-			▼	✓	✓	▼
1/2006	27 (1)	-	-	-	-	-	-	○	○		○	○	✓
	26 (6)	-	-	-	-	-	-				✓	✓	✓
	26 (5)	-	-	-	-	-	-				✓		✓
	26 (4)	-	-	-	-	-	-		▼		✓	▼	✓
	26 (3)	-	-	-	-	-	-		✓		✓	✓	✓
	26 (2)	-	-	-	-	-	-		•		✓	•	✓
	26 (1)	-	-	-	-	-	-			•	✓	✓	•
	25 (6)	-	-	-	-	-	-	•	✓	✓	•	✓	✓
	25 (5) S	-	-	-	-	-	-				✓	✓	✓
	25 (4)	-	-	-	▼			-	-	-	-	-	-
	25 (3)	-	-	-				-	-	-	-	-	-
	25 (2)	-	-	-				-	-	-	-	-	-
1/2005	25 (1)	-	-	-	▼			-	-	-	-	-	-

	24 (6)	-	-	-			▼	-	-	-	-	-	-
	24 (5)	-	-	-				-	-	-	-	-	-
	24 (4)	-	-	-				-	-	-	-	-	-
	24 (3)	-	-	-				-	-	-	-	-	-
	24 (2)	-	-	-				-	-	-	-	-	-
	24 (1)	-	-	-				-	-	-	-	-	-
	23 (6)	-	-	-	▼	•		-	-	-	-	-	-
	23 (5) S	-	-	-				-	-	-	-	-	-
	23 (4)	-	-	-				-	-	-	-	-	-
	23 (3)	-	-	-	✓	✓		-	-	-	-	-	-
	23 (2)	-	-	-	•	✓		-	-	-	-	-	-
1/2004	23 (1)				✓	✓		-	-	-	-	-	-
	22 (6)				✓	✓		-	-	-	-	-	-
	22 (5)				✓	✓	✓	-	-	-	-	-	-
	22 (4)					✓	✓	-	-	-	-	-	-
	22 (3)		-	-	✓	-	-	-	-	-	-	-	-
	22 (2)				✓	✓	✓	-	-	-	-	-	-
	22 (1)	-	-	-	✓			-	-	-	-	-	-
	21 (6)			-	✓	✓	-	-	-	-	-	-	-
	21 (5)				✓	✓	✓	-	-	-	-	-	-
	21 (4)				✓		✓	-	-	-	-	-	-
	21 (3)	-	-	-	-	-	-	-	-	-	-	-	-
	21 (2)		✓	✓		✓	✓	-	-	-	-	-	-
1/2003	21 (1)	✓	-	-	✓	-	-	-	-	-	-	-	-
	20 (6)	✓		○	✓	✓	○	-	-	-	-	-	-
	20 (5)	○	✓	✓	○	✓	✓	-	-	-	-	-	-
	20 (4)	○	○		○	○		-	-	-	-	-	-
	20 (3)	✓	✓	○	✓	✓	○	-	-	-	-	-	-
	20 (2)	✓	✓	✓	✓	✓	✓	-	-	-	-	-	-
	20 (1)	-	-	-	✓	✓	○	-	-	-	-	-	-

	19 (6)	-	-	-	✓	✓	-	-	-	-	-	-	-
	19 (5)	-	-	-	✓		▼	-	-	-	-	-	-
	19 (4)	-	-	-	✓	✓	✓	-	-	-	-	-	-
	19 (3)	-	-	-	✓	✓		-	-	-	-	-	-
	19 (2)	-	-	-	•	✓	✓	-	-	-	-	-	-
1/2002	19 (1)	-	-	-	✓	✓	✓	-	-	-	-	-	-

-	Not surveyed
○	Unable to determine due to inconsistency between nomenclature
▼	Gene not present in HGMD
•	Variant present but cited source different from <i>Human Mutation</i>
✓	Variant present
	Variant not present
S	Survey date