

Supplementary Table 1. Germline mutations identified in *MSH6* by country of probands, the number of families found to carry each mutation, the nucleotide change, the predicted protein change and the mutation type\*

Country of recruitment (No. of families)	No. of families with mutation	<i>MSH6</i> DNA change	<i>MSH6</i> mutation in protein	Mutation type
Netherlands (5) USA (1)	6	c.651_652insT	p.K218fs	Frameshift
USA (1) Scotland (2) Australia (2)	5	c.3261insC	p.F1088fs	Frameshift
Netherlands (4)	4	c.1784delT	p.L595fs	Frameshift
USA (2) Netherlands (2)	4	c.3261delC	p.F1088fs	Frameshift
USA (3) Netherlands (1)	4	c.467C>G	p.S156X	Protein truncation
USA (1) Australia (2)	3	c.3939_3957dup19	p.A1320fs	Frameshift
Scotland (3)	3	c.3939_3958dup19	p.A1320fs	Frameshift
Netherlands (2)	2	1614–1615delTCinsG	p.Y538X	Protein truncation
Netherlands (1) USA (1)	2	c.1444C>T	p.R482X	Protein truncation
Netherlands (2)	2	c.2191C>T	p.Q731X	Protein truncation
Netherlands (1) Scotland (1)	2	c.2569_2572delGATT	p.D857fs	Frameshift
Netherlands (2)	2	c.2719_2720delGT	p.V907fs	Frameshift
USA (1) Scotland (1)	2	c.2731C>T	p.R911X	Nonsense
Australia (2)	2	c.3202C>T	p.R1068X	Protein truncation
USA (2)	2	c.3439-2A>G	N/A	Splice site
Ontario (2)	2	c.3514_3515insA	p.R1172fs	Frameshift
USA (2)	2	c.3577G>A	p.E1193K	Missense
Netherlands (2)	2	c.3729_3732dupATTA	p.F1245fs	Frameshift
USA (2)	2	c.3768T>G	p.Y1256X	Nonsense
Netherlands (1) USA (1)	2	c.3987_3988insGTCA	p.L1330fs	Frameshift
Netherlands (2)	2	c.4001G>A	p.R1334Q	Splice donor defect
USA (1) Scotland (1)	2	Deletion Exon 5-6	N/A	Large rearrangement
Australia (1)	1	c. 3725_3737delGTACATTATTTTC	p.R1242fs	Frameshift
USA (1)	1	c.10C>T	p.Q4X	Protein truncation
Netherlands (1)	1	c.1190_1191delAT	p.Y397fs	Frameshift
Netherlands (1)	1	c.1267delT	p.C426fs	Frameshift
USA (1)	1	c.1430dupG	p.Y478fs	Frameshift
Australia (1)	1	c.1519_1520insTCCAAGTATGATA	p.R507fs	Frameshift
Netherlands (1)	1	c.1615_1616delCT	p.L539fs	Frameshift

Australia (1)	1	c.1628-1629delAA	p.K543fs	Frameshift
Scotland (1)	1	c.1634_1637delAAGA	p.K545fs	Frameshift
USA (1)	1	c.1637_1638delAG	p.E546fs	Frameshift
Scotland (1)	1	c.1637delAG	E546fs	Frameshift
Scotland (1)	1	c.1786T>A c.3724_3726delCGT	p.F596I, p.D1242del	Missense inframe deletion
Netherlands (1)	1	c.1869delC	p.G624fs	Frameshift
Scotland (1)	1	c.2061T>A	p.C687X	Protein truncation
USA (1)	1	c.2062_2063delGT	p.V688fs	Frameshift
Australia (1)	1	c.2150_2153delTCAG	p.V717fs	Frameshift
Australia (1)	1	c.2194C>T	p.R732X	Protein truncation
USA (1)	1	c.220G>T	p.G74X	Nonsense
Canada (1)	1	c.2504delA	p.Q835fs	Frameshift
USA (1)	1	c.2690_2691insA	p.N897fs	Frameshift
Australia (1)	1	c.2714T>A	p.L905X	Protein truncation
USA (1)	1	c.3078_3079insT	p.V1027fs	Frameshift
USA (1)	1	c.3155_3156delAG	p.E1052fs	Frameshift
Netherlands (1)	1	c.3182delT	p.L1061fs	Frameshift
Scotland (1)	1	c.3253dupA	p.T1085fs	Frameshift
Netherlands (1)	1	c.3273dupT	p.K1092fs	Frameshift
Canada (1)	1	c.3312delT	p.F1104fs	Frameshift
Canada (1)	1	c.3335_3336insATGA	p.D1112fs	Frameshift
USA (1)	1	c.3438+2_3438+3insT	N/A	Aberrant splicing
USA (1)	1	c.3516_3517delAG	p.R1172fs	Frameshift
Scotland (1)	1	c.3516_3527delAGTGTACTAG	p.R1176del	Inframe deletion
Scotland (1)	1	c.3518_3519insA	p.F1174fs	Frameshift
Netherlands (1)	1	c.3523dupA	p.T1175fs	Frameshift
USA (1)	1	c.3528_3532delACTTG	p.R1176fs	Frameshift
Australia (1)	1	c.3559_3562delGAAA	p.E1187fs	Frameshift
Australia (1)	1	c.3572-3573insT (c.3573dupT)	p.V1192fs	Frameshift
Netherlands (1)	1	c.3772C>T	p.Q1258X	Frameshift
Scotland (1)	1	c.3799_3800delAT	p.M1267fs	Frameshift
Netherlands (1)	1	c.3838C>T	p.Q1280X	Protein truncation
Scotland (1)	1	c.3840_3846delGGAGACT	p.E1280fs	Frameshift
USA (1)	1	c.3850_3851insATTA	p.T1284fs	Frameshift
USA (1)	1	c.3863_3866dupAATT	p.F1289fs	Frameshift
USA (1)	1	c.3920_3923dupATCT	p.P1309fs	Frameshift
Scotland (1)	1	c.3946_3958del13	p.G1316fs	Frameshift
USA (1)	1	c.3956_3959delAAGC	p.A1320fs	Frameshift
Scotland (1)	1	c.3980_3983dupATCA	p.Q1328fs	Frameshift

Scotland (1)	1	c.644dupT	p.V215fs	Frameshift
Netherlands (1)	1	c.649-650insT	p.D271fs	Frameshift
Netherlands (1)	1	c.742C>T	p.R248X	Protein truncation
Scotland (1)	1	c.866_867delGinsAA	p.G289E	Missense
Netherlands (1)	1	c.del 11 kB promotor area	N/A	N/A
Australia (1)	1	c.del exon 2-6	N/A	Large rearrangement
Netherlands (1)	1	c.exon 4 Nucleotide Substitutes	N/A	N/A
Netherlands (1)	1	Deletion exon 3	N/A	Large rearrangement

\* N/A, not applicable or not known.