Table 3. Inherited Disease-Causing Mutations in *FLNA*

	FLN1 Mutation	Predicted Protein Defect
Pedigree		
P1	Point mutation C—>T at 544 bp	Truncation at amino acid 182
P2	Point mutation T—>C at exon 4+2	Missplice exon 4, probable truncation
P3	Point mutation A—>G at exon 7-2	Missplice intron 6, probable truncation
P4	Point mutation G—>A at exon 20-1	Missplice intron 20
P5	Deletion AGGAGGTG at exon 25+4	Missplice intron 25, probable truncation
P6	Point mutation A—>T at 245 bp	GLU—>VAL at amino acid 82
P7	Deletion TGTGCCCC at 7627-7632 bp	Truncation of last 94 amino acids
Sporadic Females		
F1	Deletion GGCCC at 287-291 bp	Frameshift and truncation after amino acid 96
F2	Point mutation C—>G at exon 4-3	Missplice intron 3, probable truncation
F3	Point mutation G—>A at exon 2+1	Missplice intron 2, probable truncation
F4	Point mutation G—>A at 5290 bp	ALA—>THR at amino acid 1764
F5	Point mutation C—>T at 688 bp	Truncation at amino acid 230
F6	Deletion CAGC at 6636-6639 bp	Frameshift and truncation after amino acid 2212
Sporadic Males		
M1	Point mutation C—>G at 6915 bp	Truncation at amino acid 2305
M2	Point mutation C—>T at 1966 bp	LEU—>PHE at amino acid 656

OMMBID, McGraw-Hill, New York, Chap 230. www.ommbid.com