Supp. Table S1. All *MYH7* mutations associated with skeletal muscle myopathy

Exon	DNA variation*	Codon change	Protein variation	Heptad position	Clinical Diagnosis	Reference(s)
14	c.1322C>T	ACG>ATG	p.Thr441Met	N/A	MPD1, HCM	Darin et al, Neurology 2007
16	c.1816G>A	GTG>ATG	p.Val606Met	N/A	MPD1, HCM	Overeem et al, Neuromusc Disord 2007
21	c.2348G>C	CGC>CCC	p.Arg783Pro	N/A	MPD1, DCM	Homayoun et al, Neuromusc Disord 2011
30	c.4315G>C	GCT>CCT	p.Ala1439Pro	a	MPD1	Park et al, Neuromusc Disord 2013
32	c.4399C>G	CTG>GTG	p.Leu1467Val	a	Congenital myopathy	Cullup, Neuromusc Disord 2012
32	c.4442T>C	CTC>CCC	p.Leu1481Pro	a	MPD1	Current study
32	c.4499G>C	CGG>CCG	p.Arg1500Pro	f	MPD1	Meredith et al, Am J Hum Genet 2004
33	c.4522_4524delGAG	delGAG	p.Glu1508del	g	MPD1	Udd, Neuromusc Disord 2009 Dubourg et al, J Neurol 2011 Current Study
33	c.4622A>C	CAG>CCG	p.Gln1541Pro	e	MPD1	Current study
34	c.4763G>C	CGC>CCC	p.Arg1588Pro	b	MPD1	Cullup, Neuromusc Disord 2012
34	c.4772T>C	CTG>CCG	p.Leu1591Pro	e	MPD1	Tasca et al, Neuromusc Disord 2012

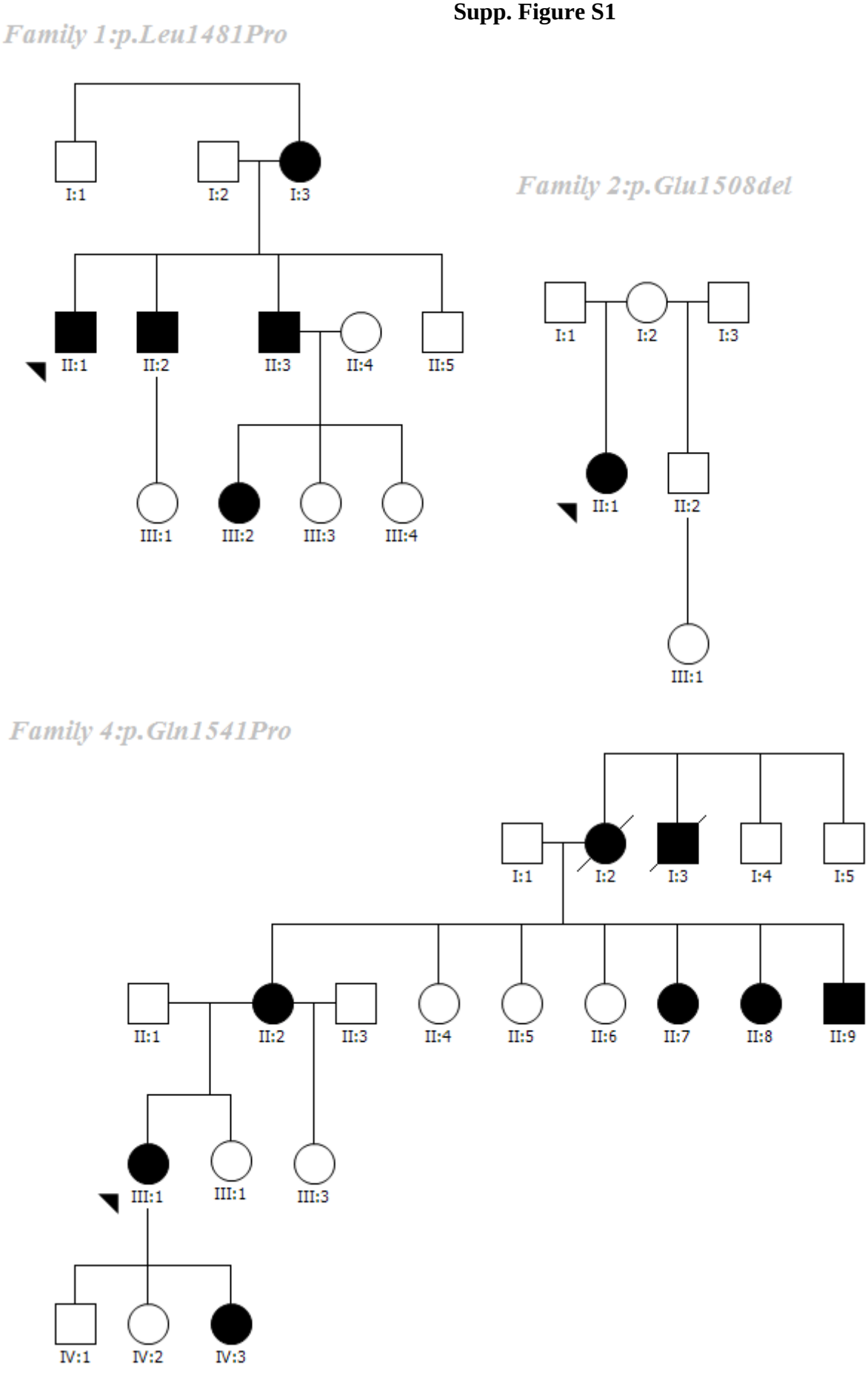
34	c.4790T>G	CTG>CGG	p.Leu1597Arg	d	Axial myopathy, Contractural myopathy	Clarke et al, Neuromusc Disord 2013
34	c.4795A>C	ACC>CCC	p.Thr1599Pro	f	MPD1	Current study
34	c.4807G>C	GCA>CCA	p.Ala1603Pro	c	MPD1	Udd, Neuromusc Disord 2009
34	c.4823G>C	CGC>CCC	p.Arg1608Pro	a	Congenital myopathy, Cardiomyopathy	Current study
34	c.4835T>C	CTG>CCG	p.Leu1612Pro	e	MPD1	Current Study
34	c.4850_4852delAGA	delAAG	p.Lys1617del	c	MPD1, DCM	Meredith et al, Am J Hum Genet 2004 Lamont et al, JNNP 2006 Current study
34	c.4906G>C	GCC>CCC	p.Ala1636Pro	a	MPD1	Current study
34	c.4937T>C	CTC>CCC	p.Leu1646Pro	d	MPD1	Current study
35	c.4985G>C	CGT>CCT	p.Arg1662Pro	f	MPD1	Current study
35	c.4987G>C	GCC>CCC	p.Ala1663Pro	g	MPD1	Meredith et al, Am J Hum Genet 2004
35	c.5005_5007delGAG	delGAG	p.Glu1669del		MPD1	Current study
35	c.5117T>C	CTG>CCG	p.Leu1706Pro	g	MPD1	Meredith et al, Am J Hum Genet 2004
36	c.5186_5188delAGA	delAAG	p.Lys1729del	c	MPD1, DCM, Scapuloperoneal myopathy	Meredith et al, Am J Hum Genet 2004 Muelas et al, Neurology 2010
36	c.5186_5188dupAGA	dupAAG	p.Lys1729dup	c	MPD1	Current study
37	c.5336T>C	CTG>CCG	p.Leu1779Pro	d	MSM	Chai et al, Neuromusc Disord 2007
37	c.5352_5354delGAA	delGAA	p.Lys1784del	b	MPD1, MSM	Stalpers et al, Neuromusc Disord 2011 Tasca et al, Neuromusc Disord 2012

37	c.5378_5380delTGC	delCTG	p.Leu1793del	d	MPD1, DCM	Current study
37	c.5378T>C	CTG>CCG	p.Leu1793Pro	d	MPD1, MSM, HCM	Dye et al, Neuromusc Disord 2006 Uro-Coste et al, Neuromusc Disord 2009
37	c.5401G>A	GAG>AAG	p.Glu1801Lys	e	MPD1, DCM, HCM	Udd , Neuromusc Disord 2009 Current study
37	c.5533C>T	CGG>TGG	p.Arg1845Trp	f	MSM	Tajsharghi et al, Ann Neurol 2003 Pegoraro et al, Neuromusc Disord 2007
38	c.5566G>A	GAG>AAG	p.Glu1856Lys	c	MPD1, Cardiomyopathy	Udd, Neuromusc Disord 2009
38	c.5647G>A	GAG>AAG	p.Glu1883Lys	b	MSM, HCM	Tajsharghi et al, Neurology 2007
39	c.5702A>T	CAC>CTC	p.His1901Leu	f	MSM	Bohlega et al, Neurol 2004
39	c.5740G>A	GAG>AAG	p.Glu1914Lys	e	DCM, Congenital myopathy	Current study
40	c.5807A>G	TAG>TGG	p.*1936Trpext*31	N/A	Proximal myopathy with CFTD	Ortolano et al, Neuromusc Disord 2011

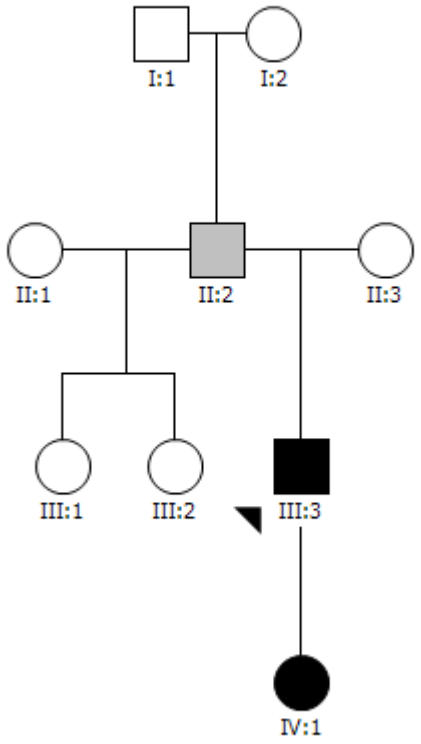
All *MYH7* mutations associated with a defined skeletal myopathy, with or without cardiomyopathy. All mutations are heterozygous dominant except p.Glu1883Lys which is recessive.

*MYH7 cDNA Accession number used = NM_000257.2. DNA variation is given according to the Human Genome Variation Society notation as obtained from Mutalyzer (https://mutalyzer.nl)
CFTD = congenital fibre type disproportion, DCM = dilated cardiomyopathy, HCM = hypertrophic cardiomyopathy, MPD1 = distal myopathy type 1, MSM = Myosin storage myopathy

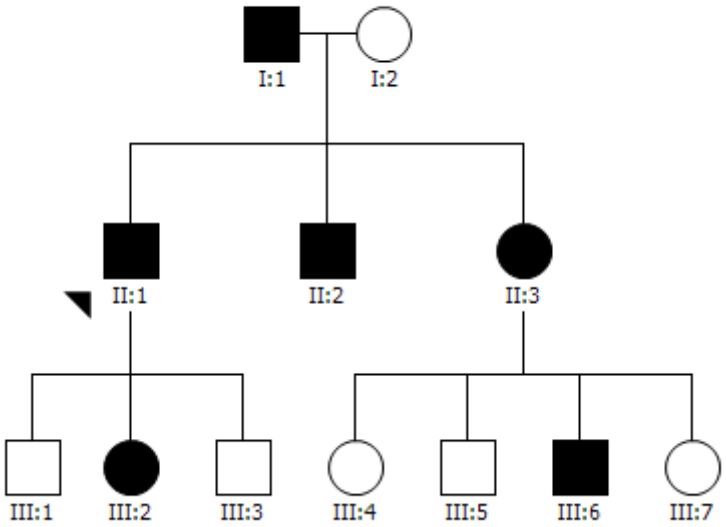
Supp. Figure S1



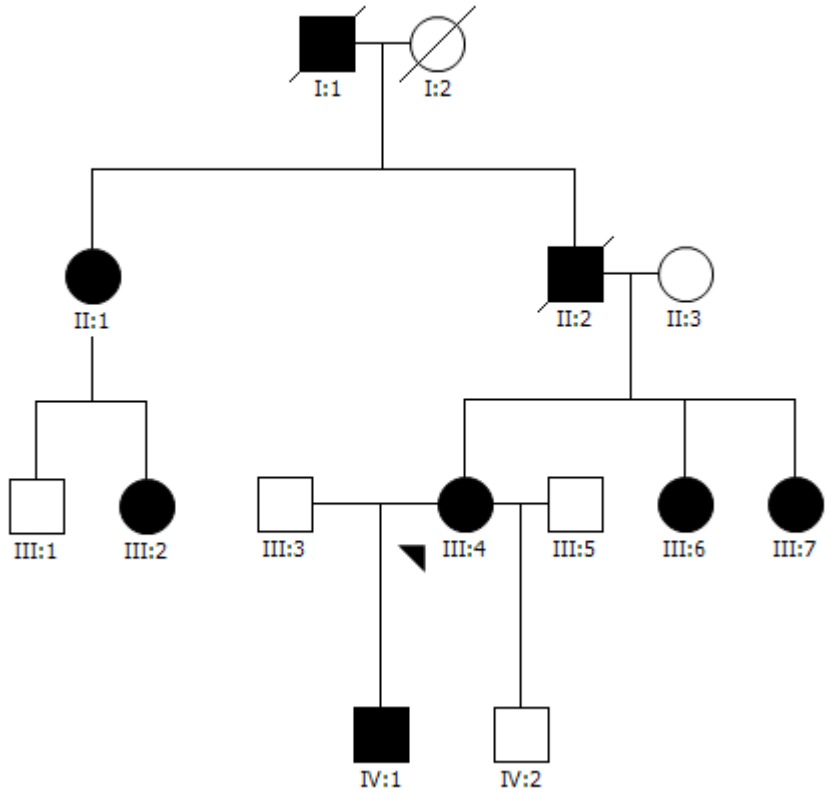
Family 6:p.Arg1608Pro



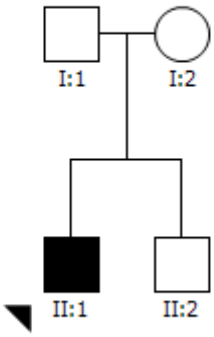
Family 5:p.Thr1599Pro



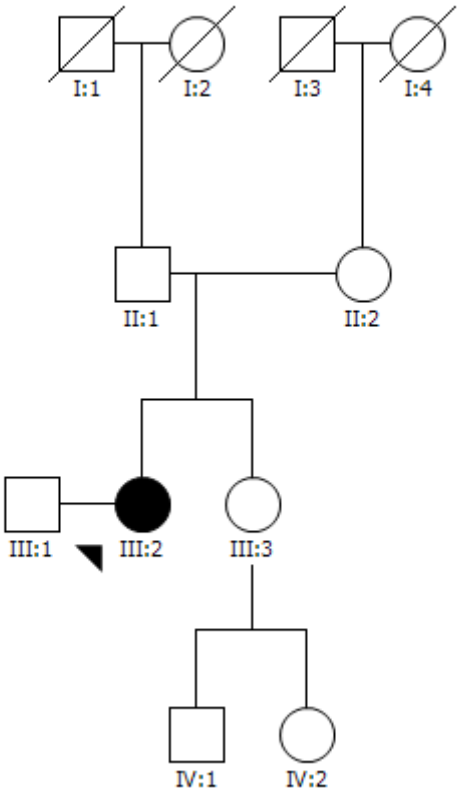
Family 9:p.Lys1617del



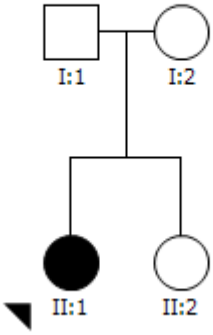
Family 7:p.Leu1612Pro



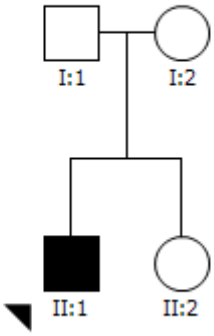
Family 10:p.Lys1617del



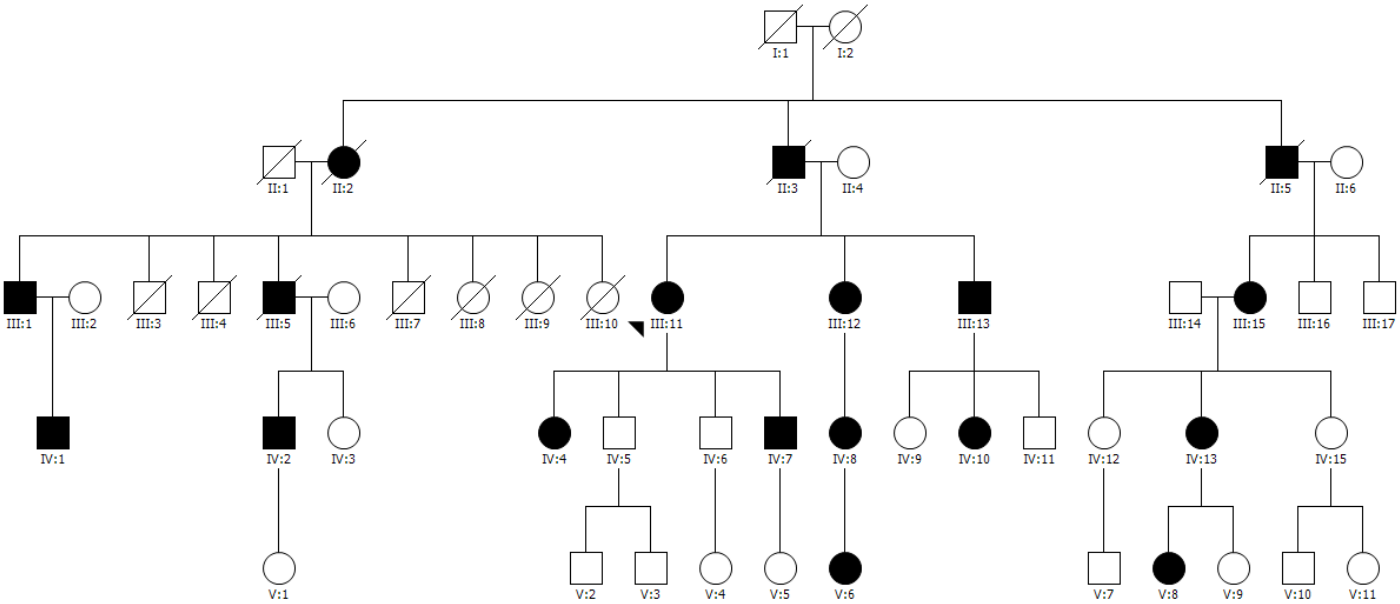
Family 11:p.Lys1617del



Family 12:p.Lys1517del

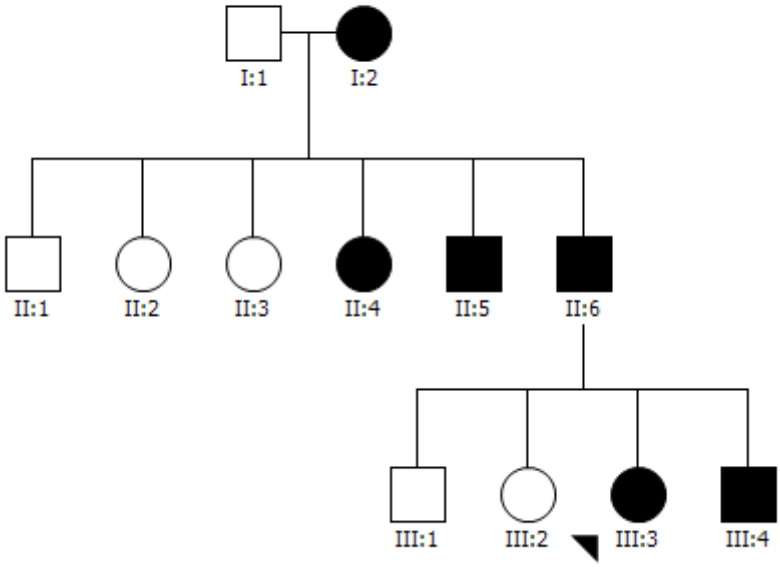


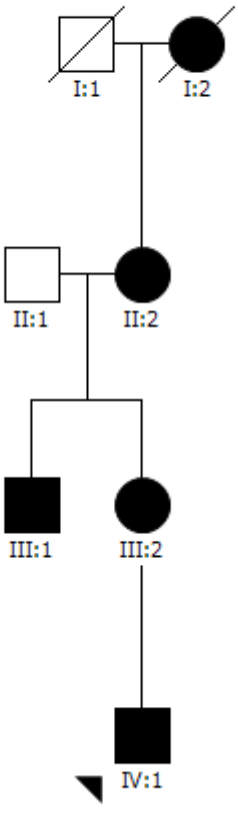
Family 13:p.Ala1636Pro



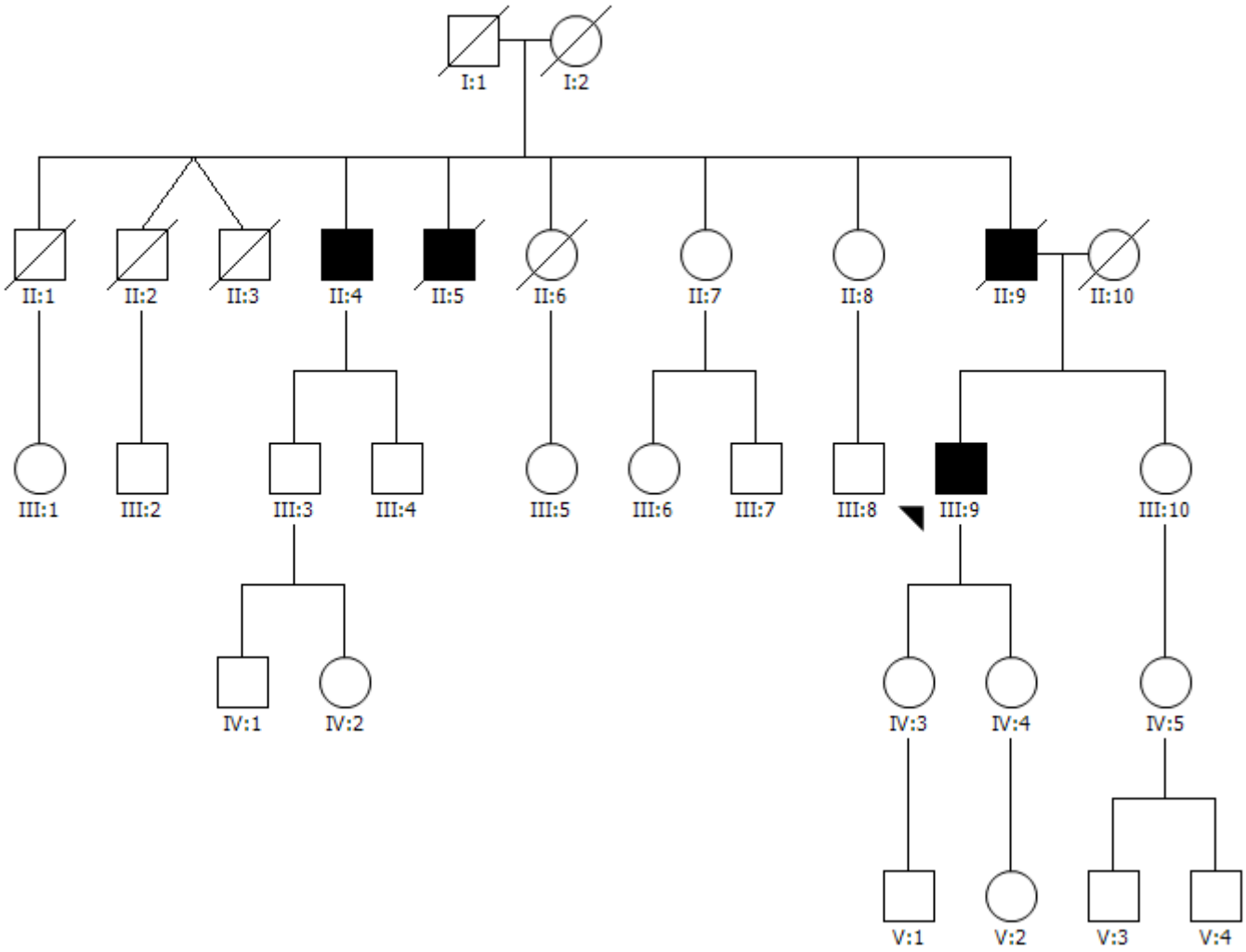
Family 15:p.Arg1662Pro

Family 14:p.Leu1646Pro

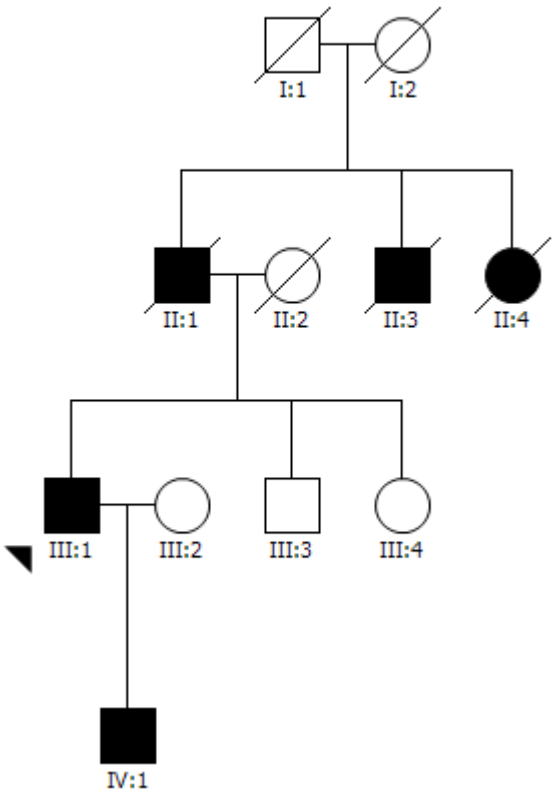




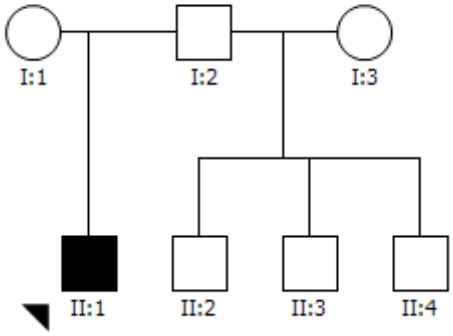
Family 16:p.Glu1669del



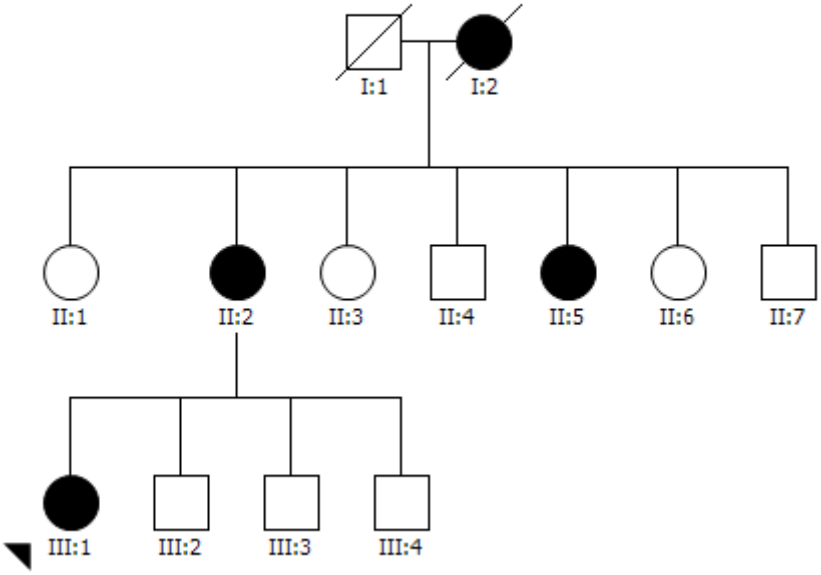
Family 17:p.Lys1729dup



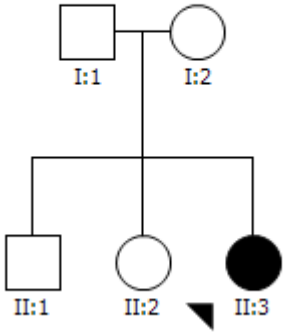
Family 18:p.Leu1739del

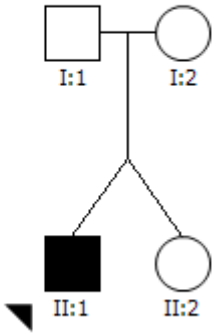


Family 19:p.Glu1801Lys



Family 20:p.Glu1914Lys





Supp. Figure S1. Pedigrees for all families, except family 8 which was archival and detailed information was not available.
Black Symbol: Affected individual clinically.
Grey symbol in family 6: Somatic mosaic for *MYH7* mutation, minimally affected clinically.