

**S3 Table**

<b>F13a</b>	<b>TGM1</b>		<b>TGM5</b>	<b>TGM6</b>
g.210_211del, (homozygous)	c.4640del (homozygous)	p.Asn288Thr	p.Gly113Cys (homozygous)	p.Leu517Trp
p.Arg681His (homozygous)	g.3366A>G (homozygous)	p.Arg306Trp	p.Lys445Asn (homozygous)	p.Asp327Gly
p.Arg171Ter	p.Ser42Tyr	p.Gly217Ser	p.Trp255Arg	p.Asp510His
p.Tyr441Ter	p.Arg142Cys	g.1303_1307del	p.Leu41Pro	p.Arg111Cys
p.Asn60Lys	p.Arg323Gln	p.Gln582Ter	c.640del	c.1722_1724del
p.Gly501Arg	p.Arg141His (homozygous)	g.86C>T	c.1811_1815delinsT CCTTCA	
p.Gly562Arg (homozygous)	p.Arg142His (homozygous)	p.Asp101Val		
p.Val414Phe (homozygous)	p.Val378Leu	p.Leu204Gln		
p.Arg260His (homozygous)	p.Arg395Leu	p.Tyr276Asn (homozygous)		
p.Val34Leu (homozygous)	p.Val382Met (homozygous)	p.Arg126Cys		
p.Arg326Gln	p.Arg388His	p.Arg315Cys		
p.Val316Phe	g.9008del	p.Arg315His (homozygous)		
p.Tyr283Cys	p.Asp490Gly	p.Arg315Leu (homozygous)		
c.1286_1287insC (homozygous)	p.Gly278Arg	p.Val359Met		
g.[5G>A;15C>T]	p.Gly392Asp	p.Arg396His		
p.Arg703Trp	p.Arg142Pro	g.1922_1928del		
p.Arg661Ter	p.Arg286Gln	g.1331_1332insA		
p.Met242Thr	p.Val518Met	p.Arg670Ter (homozygous)		
c.602_605del	p.Ser160Cys	g.1223_1227del		
n.1+12C>A	p.Gly94Asp			