Supp. Table S1. Summary of previously misreported mutations and corrections

cDNA	Protein	Error Type	Error	Correction	Initial Report	Correction
c.205T>G	F69V	cDNA position	c.206T>G	c.205T>G	Tomatsu 2004c, Tomatsu 2005	Typo
c.498C>G	H166Q	cDNA position	c.478C>G	c.498C>G	Tomatsu 2004c	Tomatsu 2005
c.491A>C	N164T	cDNA position	c.500A>C	c.491A>C	Tomatsu 2004c	Tomatsu 2005
c.689G>A	W230X	cDNA position	c.690G>A	c.689G>A	Tomatsu 2005	Tomatsu 2005 incorrectly cites Montaño 2003; Montaño 2003 correctly lists c.689G>A
c.969C>T	L323L	Mutation effect	Deleterious	Synonymous	Tomatsu 1997	Tomatsu 2005
c.1023C>G	S341R	cDNA position	c.1022C>G	c.1023C>G	Tomatsu 2004c	Tomatsu 2005
c.1028T>G	M343R	cDNA position and protein	c.1027A>T (p.M343L)	c.1028T>G (p.M343R)	Ogawa 1995	Tomatsu 2005
c.1157G>A	R386H	cDNA position	c.1158G>A	c.1157G>A	Tomatsu 2004c	Tomatsu 2005
c.1417C>T	Q473X	cDNA position	c.1472C>T	c.1417C>T	Terzioglu 2002	Tomatsu 2005
c.1483A>T	N495Y	cDNA position	c.1472A>T	c.1483A>T	Pajares 2012	Author correspondence
c.1568A>G	X523W	Protein notation	Y523W	X523W	Lee 2012	Typo

Supp. Table S2. Reported *GALNS* genotypes of patients with Morquio A

DNA and protein sequence numbering is based on the *GALNS* cDNA sequence (GenBank entry NM_000512.4), and the DNA sequence position "1" corresponds to the A of the initial ATG in the reference sequence.

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.1A>G	p.M1V	c.1A>G	p.M1V	Severe	Italian	M	Tomatsu	2004b
c.1A>G	p.M1V	c.1A>G	p.M1V	Severe	Italian	M	Tomatsu	2004b
c.1A>G	p.M1V	c.1A>G	p.M1V	Severe	Italian	F	Tomatsu	2004b
c.1A>G	p.M1V	c.1A>G	p.M1V	Severe	Italian	M	Tomatsu	2004b
c.1A>G	p.M1V	c.1450C>T	p.P484S		Japanese	M	Montaño	2007
not given	p.M1I	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.3G>A	p.M1I	c.257T>C	p.L86P	Severe	Finnish	M	Dung	2013
c.29G>A	p.W10Ter	c.29G>A	p.W10Ter			M	Bunge	1997
c.29G>A	p.W10Ter	c.29G>A	p.W10Ter	Severe	Italian	M	Tomatsu	2004b
c.29G>A	p.W10Ter	c.448C>T	p.H150Y	Intermediate	Italian	F	Tomatsu	2004b
c.29G>A	p.W10Ter	c.850_852delTTC	p.F284del	Severe	Italian	F	Tomatsu	2004b
c.43C>A	p.L15M	n/a, reported as allele	n/a, reported as allele	Attenuated	Scottish		Tomatsu	2005
c.43C>A	p.L15M	unknown	unknown	Attenuated			Tomatsu	2005
c.47T>A	p.V16E	c.47T>A	p.V16E		Brazilian		Morrone	2014
c.67G>C	p.G23R	c.1175C>T	p.A392V	Mild	Italian	F	Tomatsu	2004b
c.67G>C	p.G23R	c.1175C>T	p.A392V	Attenuated			Tomatsu	2005
not given	p.N32D	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.106_111del	p.L36_L37del	c.953T>G	p.M318R		Taiwanese		Yang	2001

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.106_111del	p.L36_L37del	c.1097T>C	p.L366P		Chinese	M	Wang	2010
c.106_111del	p.L36_L37del	c.1097T>C	p.L366P		Chinese		Guo	2012
c.107T>G	p.L36R	c.107T>G	p.L36R		Asian-multiethnic	F	Morrone	2014
c.107T>G	p.L36R	c.107T>G	p.L36R		Asian-multiethnic		Morrone	2014
c.107T>G	p.L36R	c.107T>G	p.L36R		Asian-multiethnic	F	Morrone	2014
c.107T>C	p.L36P	c.567-131ins42	c.567-131ins42	Attenuated			Tomatsu	2005
c.107T>C	p.L36P	n/a, reported as allele	n/a, reported as allele	Severe	Mexican		Tomatsu	2005
c.118G>A	p.D40N	c.118G>A	p.D40N		Asian-multiethnic	F	Morrone	2014
c.120+1G>A	c.120+1G>A	c.120+1G>A	c.120+1G>A		Tunisian	F	Laradi	2006
c.120+1G>A	c.120+1G>A	c.120+1G>A	c.120+1G>A		Tunisian	M	Laradi	2006
c.120+1G>A	c.120+1G>A	c.120+1G>A	c.120+1G>A		Tunisian	F	Laradi	2006
c.120+1G>A	c.120+1G>A	c.120+1G>A	c.120+1G>A		Tunisian	M	Laradi	2006
c.120+1G>A	c.120+1G>A	c.120+1G>A	c.120+1G>A		Tunisian	F	Laradi	2006
c.120+1G>A	c.120+1G>A	c.120+1G>A	c.120+1G>A			F	Carraresi	2008
c.120+1G>A	c.120+1G>A	c.120+1G>A	c.120+1G>A		Tunisian	M	Khedhiri	2011
c.120+1G>A	c.120+1G>A	c.120+1G>A	c.120+1G>A		Tunisian	F	Khedhiri	2011
c.120+1G>A	c.120+1G>A	c.120+1G>A	c.120+1G>A		Tunisian	M	Khedhiri	2011
c.120+1G>A	c.120+1G>A	c.120+1G>A	c.120+1G>A		Tunisian	F	Khedhiri	2011
c.120+1G>A	c.120+1G>A	c.120+1G>A	c.120+1G>A		Tunisian	F	Khedhiri	2011
c.120+1G>C	c.120+1G>C	c.120+1G>C	c.120+1G>C		Middle Eastern		Morrone	2014
c.120_121ins158	p.D40fsTer20	n/a, reported as allele	n/a, reported as allele	Severe	Mexican		Tomatsu	2005

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.120_121ins158	p.D40fsTer20	n/a, reported as allele	n/a, reported as allele	Severe	Japanese		Tomatsu	2005
c.120+1G>A	c.120+1G>A	n/a, reported as allele	n/a, reported as allele		Tunisian		Khedhiri	2012
c.121-1G>A	c.121-1G>A	c.121-1G>A	c.121-1G>A	Severe	Middle Eastern	F	Morrone	2014
c.121-1G>A	c.121-1G>A	c.121-1G>A	c.121-1G>A	Severe	Middle Eastern	M	Morrone	2014
c.121-2A>G	c.121-2A>G	c.121-2A>G	c.121-2A>G	Severe	Japanese	F	Ogawa	1995
c.121-2A>G	c.121-2A>G	c.121-2A>G	c.121-2A>G	Severe	Japanese	M	Ogawa	1995
c.121A>T	p.M41L	c.121A>T	p.M41L	Mild	Acadian	M	Tomatsu	2004c
c.121A>T	p.M41L	c.121A>T	p.M41L	Severe	Acadian	M	Tomatsu	2004c
c.121A>T	p.M41L	c.121A>T	p.M41L	Attenuated			Tomatsu	2005
c.121A>T	p.M41L	c.121A>T	p.M41L	Attenuated	American Caucasian, French	M	Dung	2013
c.121-1G>A	p.M41RfsTer46	c.337A>T	p.I113F	Severe	American Caucasian	M	Dung	2013
c.121-?_244+?del	p.M41_P81>RfsTer47	n/a, reported as allele	n/a, reported as allele	Severe	Japanese		Tomatsu	2005
c.121-?_244+?del	p.M41_P81>RfsTer47	n/a, reported as allele	n/a, reported as allele	Severe	Japanese		Tomatsu	2005
c.121-1G>C	p.M41RfsTer46	n/a, reported as allele	n/a, reported as allele	Severe	Ukrainian		Tomatsu	2005
c.122T>A	p.M41K	c.122T>A	p.M41K	Severe	American Caucasian	F	Dung	2013
c.125G>A	p.G42E	c.374C>T	p.P125L	Severe	Italian	M	Dung	2013
c.125G>A	p.G42E	n/a, reported as allele	n/a, reported as allele	Severe	Italian		Tomatsu	2005
c.139G>A	p.G47R	c.139G>A	p.G47R	Severe	Polish	F	Bunge	1997
c.139G>A	p.G47R	c.139G>A	p.G47R	Severe	Honduran	M	Tomatsu	2004c
c.139G>A	p.G47R	c.139G>A	p.G47R	Severe	Polish	M	Dung	2013
c.139G>A	p.G47R	c.1142delC	p.P381LfsTer9	Rapidly progressing	Spanish	F	Pajares	2012
c.143T>G	p.V48G	c.600delG	p.E202KfsTer115		British	M	Morrone	2014

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.151G>A	p.E51K	c.151G>A	p.E51K		Brazilian		Morrone	2014
c.158C>T	p.S53F	n/a, reported as allele	n/a, reported as allele	Undefined	Japanese		Tomatsu	2005
c.178G>A	p.D60N	c.689G>A	p.W230Ter	Mild	Finnish	M	Montaño	2003
c.178G>A	p.D60N	c.689G>A	p.W230Ter	Mild	Finnish	M	Montaño	2003
c.178G>A	p.D60N	c.689G>A	p.W230Ter	Attenuated			Tomatsu	2005
c.178G>A	p.D60N	c.871G>A	p.A291T	Mild	Finnish	M	Bunge	1997
c.178G>A	p.D60N	c.871G>A	p.A291T	Intermediate	Finnish	M	Montaño	2003
c.178G>A	p.D60N	c.871G>A	p.A291T	Intermediate	Finnish	M	Montaño	2003
c.178G>A	p.D60N	c.871G>A	p.A291T	Attenuated			Tomatsu	2005
c.178G>A	p.D60N	c.1319delT	p.L440RfsTer54	Attenuated			Tomatsu	2005
c.178G>A	p.D60N	c.1319delT	p.L440RfsTer54	Intermediate	Finnish	F	Montaño	2003
not given	p.D60E	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.181C>T	p.R61W	c.498delC	p.H166fsTer32	Attenuated			Tomatsu	2005
c.181C>T	p.R61W	c.498delC	p.H166fsTer32	Attenuated	American Caucasian	M	Dung	2013
c.181C>T	p.R61W	c.498delC	p.H166fsTer32	Attenuated	American Caucasian	F	Dung	2013
c.181C>T	p.R61W	n/a, reported as allele	n/a, reported as allele	Attenuated	American		Tomatsu	2005
c.196G>A	p.G66R	c.196G>A	p.G66R		Tunisian	F	Laradi	2006
c.196G>A	p.G66R	c.196G>A	p.G66R		Tunisian	F	Khedhiri	2011
not given	p.G66R	n/a, reported as allele	n/a, reported as allele		Tunisian		Khedhiri	2012
c.199C>A, c.1226G>C	p.L67M, p.W409S	c.199C>A, c.1226G>C	p.L67M, p.W409S	Intermediate	Dutch	M	Bunge	1997
c.205T>G	p.F69V	c.901G>T	p.G301C	Severe	Colombian	M	Tomatsu	2004c
c.205T>G	p.F69V	unknown	unknown	Severe	Colombian	M	Kato	1997

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.209_220del	p.N71_S74delinsT	n/a, reported as allele	n/a, reported as allele	Severe	Irish		Tomatsu	2005
c.218A>G	p.Y73C	c.725C>G	p.S242C	Intermediate	Korean		Lee	2012
c.218A>G	p.Y73C	c.725C>G	p.S242C		Korean	F	Park	2013
c.221C>T	p.S74F	c.423_566del	Exon 5 skipping	Slowly progressing	Spanish	M	Pajares	2012
c.224C>G	p.A75G	c.224C>G	p.A75G	Severe	Colombian	F	Tomatsu	2004a
c.224C>G	p.A75G	c.224C>G	p.A75G	Severe	Colombian	M	Tomatsu	2004a
c.230C>G	p.P77R	c.230C>G	p.P77R	Severe	Indian	M	Tomatsu	1995
c.230C>G	p.P77R	c.230C>G	p.P77R	Severe	Caucasian	M	Tomatsu	2004c
c.230C>G	p.P77R	c.230C>G	p.P77R	Severe	Saudi Arabia	F	Dung	2013
c.230C>G	p.P77R	c.230C>G	p.P77R		Asian-multiethnic		Morrone	2014
c.230C>G	p.P77R	c.230C>G	p.P77R		Middle Eastern		Morrone	2014
c.235_236delTG	p.C79LfsTer78	unknown	unknown	Intermediate	Dutch	F	Bunge	1997
c.236G>A	p.C79Y	c.236G>A	p.C79Y		Italian	M	Catarzi	2012
c.236G>A	p.C79Y	c.415G>A	p.G139S	Severe	Argentine	M	Tomatsu	2004a
c.239C>T	p.S80L	c.740G>A	p.G247D		British	F	Morrone	2014
c.239C>T	p.S80L	n/a, reported as allele	n/a, reported as allele	Severe	British		Tomatsu	2005
c.242C>T	p.P81L	c.242C>T	p.P81L		Turkish	M	Morrone	2014
c.242C>T	p.P81L	c.1156C>T	p.R386C		Other	M	Morrone	2014
c.244T>C	p.S82P	c.244T>C	p.S82P	Attenuated	Canadian	F	Dung	2013
c.245C>T	p.S82L	c.498delC	p.H166fsTer32	Severe	American Caucasian	M	Dung	2013
c.251C>A	p.A84E	c.251C>A	p.A84E	Rapidly progressing	Portuguese	M	Morrone	2014

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.253G>A	p.A85T	c.253G>A	p.A85T		Tunisian	F	Khedhiri	2011
c.268C>T	p.R90W	c.268C>T	p.R90W	Severe	Pakistani	M	Tomatsu	1995
c.268C>T	p.R90W	c.268C>T	p.R90W		Asian-multiethnic	M	Morrone	2014
c.268C>T	p.R90W	c.268C>T	p.R90W		Turkish		Morrone	2014
c.268C>T	p.R90W	c.1019G>A	p.G340D	Severe	Austrian	F	Tomatsu	2004c
c.272T>C	p.L91P	c.272T>C	p.L91P		Portuguese	F	Morrone	2014
c.278T>A	p.I93N	unknown	unknown	Severe	American Caucasian, Albanian	M	Dung	2013
c.280C>T	p.R94C	c.280C>T	p.R94C	Severe	Middle Eastern	M	Morrone	2014
c.280C>T	p.R94C	c.280C>T	p.R94C	Severe	Middle Eastern	M	Morrone	2014
c.280C>T	p.R94C	c.280C>T	p.R94C		Middle Eastern		Morrone	2014
c.280C>T	p.R94C	c.289T>G	p.F97V	Attenuated	Canadian	F	Cole	1996
c.280C>T	p.R94C	c.289T>G	p.F97V	Attenuated			Tomatsu	2005
c.280C>T	p.R94C	c.608C>T	p.A203V	Severe	Brazilian	F	Dung	2013
c.280C>T	p.R94C	c.608C>T	p.A203V	Severe	Brazilian	M	Dung	2013
c.280C>G	p.R94G	c.612C>G	p.N204K	Intermediate	Japanese	M	Ogawa	1995
c.280C>G	p.R94G	c.612C>G	p.N204K	Mild	Japanese	M	Tomatsu	2004c
c.280C>G	p.R94G	c.612C>G	p.N204K	Attenuated			Tomatsu	2005
c.280C>G	p.R94G	c.776G>A	p.R259Q	Attenuated			Tomatsu	2005
c.280C>T	p.R94C	unknown	unknown	Unknown/atypical	Caucasian	M	Ogawa	1995
c.281G>T	p.R94L	c.281G>T	p.R94L	Rapidly progressing	Portuguese	M	Morrone	2014
c.281G>T	p.R94L	n/a, reported as allele	n/a, reported as allele	Severe	Turkish		Tomatsu	2005

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.287G>T	p.G96V	c.287G>T	p.G96V	Severe	Irish	M	Tomatsu	1995
c.287G>T	p.G96V	unknown	unknown	Severe	Irish	F	Yamada	1998
c.317A>G	p.N106S	c.553delG	p.E185Rfs14	Attenuated	Korean	M	Lachman	2014
c.319G>A	p.A107T	c.263C>T	p.T88I		Chinese	F	Wang	2010
c.319G>A	p.A107T	c.319G>A	p.A107T	Severe	Italian	M	Tomatsu	2004b
c.319G>A	p.A107T	c.1162G>A	p.D388N	Intermediate	Italian	M	Tomatsu	2004b
c.319G>A	p.A107T	unknown	unknown	Severe	Chinese	M	Wang	2010
c.320–1G>T	p.A107GfsTer54	unknown	unknown	Attenuated	Chilean	M	Dung	2013
c.331C>T	p.Q111Ter	c.337A>T	p.I113F		British	F	Morrone	2014
c.332A>G	p.Q111R	c.332A>G	p.Q111R	Intermediate	Dutch	F	Bunge	1997
c.332A>G	p.Q111R	c.332A>G	p.Q111R	Attenuated			Tomatsu	2005
c.334delG	p.E112RfsTer15	c.334delG	p.E112RfsTer15		Turkish		Terzioglu	2002
c.334delG	p.E112RfsTer15	c.334delG	p.E112RfsTer15		Turkish		Terzioglu	2002
c.334delG	p.E112RfsTer15	c.334delG	p.E112RfsTer15		Turkish		Terzioglu	2002
c.334delG	p.E112RfsTer15	c.334delG	p.E112RfsTer15		Turkish		Terzioglu	2002
c.337A>T	p.I113F	c.319G>A	p.A107T		British	F	Morrone	2014
c.337A>T	p.I113F	c.337A>T	p.I113F		British	F	Tomatsu	1995
c.337A>T	p.I113F	c.337A>T	p.I113F		British	F	Morrone	2014
c.337A>T	p.I113F	c.337A>T	p.I113F		British	M	Morrone	2014
c.337A>T	p.I113F	c.337A>T	p.I113F		Irish	F	Morrone	2014
c.337A>T	p.I113F	c.337A>T	p.I113F		Irish	F	Morrone	2014

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.337A>T	p.I113F	c.337A>T	p.I113F		Irish	F	Morrone	2014
c.337A>T	p.I113F	c.337A>T	p.I113F		Irish	F	Morrone	2014
c.337A>T	p.I113F	c.337A>T	p.I113F		Irish	F	Morrone	2014
c.337A>T	p.I113F	c.467T>G	p.F156C	Severe	British/Irish	M	Yamada	1998
c.337A>T	p.I113F	c.467T>G	p.F156C		Australian		Morrone	2014
c.337A>T	p.I113F	c.719A>G	p.Y240C		Other	M	Morrone	2014
c.337A>T	p.I113F	c.841_867del	p.T281_N289del		Irish	F	Morrone	2014
c.337A>T	p.I113F	c.841_867del	p.T281_N289del		British	M	Tomatsu	1995
c.337A>T	p.I113F	c.935C>G	p.T312S	Intermediate	Irish	M	Yamada	1998
c.337A>T	p.I113F	c.935C>G	p.T312S	Mild	Irish	F	Yamada	1998
c.337A>T	p.I113F	c.935C>G	p.T312S		Irish	F	Morrone	2014
c.337A>T	p.I113F	c.935C>G	p.T312S		Irish	F	Morrone	2014
c.337A>T	p.I113F	c.935C>G	p.T312S		Irish	M	Morrone	2014
c.337A>T	p.I113F	c.935C>G	p.T312S		Irish	M	Morrone	2014
c.337A>T	p.I113F	c.1082G>T	p.R361G	Severe	British/German	M	Tomatsu	2004c
c.337A>T	p.I113F	c.1157G>A	p.R386H		Other	M	Morrone	2014
c.337A>T	p.I113F	c.1171A>G	p.M391V		British	M	Tomatsu	1995
c.337A>T	p.I113F	c.1171A>G	p.M391V		Other	M	Morrone	2014
c.337A>T	p.I113F	c.1183C>G	p.L395V	Severe	Irish	M	Yamada	1998
c.337A>T	p.I113F	c.1319delT	p.L440RfsTer54		British	F	Tomatsu	1995

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.337A>T	p.I113F	c.1319delT	p.L440RfsTer54		Irish	F	Morrone	2014
c.337A>T	p.I113F	unknown	unknown		British	M	Tomatsu	1995
c.337A>T	p.I113F	unknown	unknown		Irish	M	Tomatsu	1995
c.337A>T	p.I113F	unknown	unknown		Irish	M	Tomatsu	1995
c.337A>T	p.I113F	unknown	unknown		Irish	F	Tomatsu	1995
c.337A>T	p.I113F	unknown	unknown		Irish	M	Tomatsu	1995
c.337A>T	p.I113F	unknown	unknown	Intermediate	British/Irish	F	Yamada	1998
c.337A>T	p.I113F	unknown	unknown	Mild	Irish	M	Yamada	1998
c.337A>T	p.I113F	unknown	unknown	Severe	British/Irish	M	Yamada	1998
c.337A>T	p.I113F	unknown	unknown	Severe	Irish	M	Yamada	1998
c.337A>T	p.I113F	unknown	unknown		British	M	Morrone	2014
c.337A>T	p.I113F	unknown	unknown		British	M	Morrone	2014
c.337A>T	p.I113F	unknown	unknown		British	F	Morrone	2014
c.337A>T	p.I113F	unknown	unknown		Irish	M	Morrone	2014
c.346G>A	p.G116S	c.346G>A	p.G116S	Severe	Brazilian	M	Tomatsu	2004a
c.346G>A	p.G116S	c.346G>A	p.G116S	Severe	Brazilian	M	Tomatsu	2004a
c.346G>A	p.G116S	c.346G>A	p.G116S	Rapidly progressing	Portuguese	F	Morrone	2014
c.346G>A	p.G116S	c.452C>T	p.P151L		Asian-multiethnic		Morrone	2014
c.346G>A	p.G116S	c.860C>T	p.S287L	Severe	Turkish	F	Dung	2013
c.346G>A	p.G116S	c.860C>T	p.S287L		Turkish	M	Morrone	2014

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.346G>A	p.G116S	c.1156C>T	p.R386C	Severe	American Caucasian	F	Dung	2013
c.346G>A	p.G116S	c.1156C>T	p.R386C	Severe	American Caucasian, Italian	F	Dung	2013
c.346G>A	p.G116S	c.1485C>G	p.N495K	Severe	American Caucasian	F	Dung	2013
c.347G>T	p.G116V	c.347G>T	p.G116V		Asian-multiethnic		Morrone	2014
c.347G>T	p.G116V	c.347G>T	p.G116V		Asian-multiethnic	F	Morrone	2014
c.347G>T	p.G116V	c.347G>T	p.G116V		Asian-multiethnic	M	Morrone	2014
c.347G>T	p.G116V	c.347G>T	p.G116V		Asian-multiethnic	F	Morrone	2014
c.347G>T	p.G116V	c.347G>T	p.G116V		Asian-multiethnic	F	Morrone	2014
c.347G>T	p.G116V	c.347G>T	p.G116V		Asian-multiethnic	M	Morrone	2014
c.347G>T	p.G116V	c.347G>T	p.G116V		Norwegian	M	Morrone	2014
c.363G>C	p.E121D	c.363G>C	p.E121D	Moderate	African-Varied	M	Pajares	2012
c.374C>T	p.P125L	c.374C>T	p.P125L	Severe	Japanese		Tomatsu	1997
c.374C>T	p.P125L	c.374C>T	p.P125L	Severe	Italian	M	Tomatsu	2004b
c.374C>T	p.P125L	c.1019G>A	p.G340D		Chinese		He	2013
c.374C>T	p.P125L	unknown	unknown		Chinese		He	2013
c.374C>T	p.P125L	unknown	unknown		Chinese		He	2013
c.376G>T	p.E126Ter	c.865A>G	p.N289D		Australian		Morrone	2014
c.385A>T	p.K129Ter	c.899-1G>C	c.899-1G>C			F	Carraresi	2008
c.405_422+1del	p.S135fsTer94	c.1480A>G	p.M494V	Severe	Canadian Caucasian	M	Dung	2013
c.405C>G	p.S135R	unknown	unknown	Severe		M	Yamada	1998
c.409_420del12	c.409_c.420del12	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.413T>C	p.V138A	c.121-2779_567-248, c.1140-730_1365-1530	double gene deletion		Japanese	M	Tomatsu	1996
c.413T>C	p.V138A	c.121-2779_567-248, c.1140-730_1365-1530	double gene deletion	Attenuated	Japanese	M	Tomatsu	2005
c.415G>A	p.G139S	c.245C>T	p.S82L	Severe	Chinese	M	Wang	2010
c.415G>A	p.G139S	c.415G>A	p.G139S		Asian-multiethnic		Morrone	2014
c.415G>A	p.G139S	c.415G>A	p.G139S		Asian-multiethnic		Morrone	2014
c.415G>A	p.G139S	c.415G>A	p.G139S		South-East Asian		Morrone	2014
c.415G>A	p.G139S	c.491A>C	p.N164T	Severe	Brazilian	F	Tomatsu	2004a
c.415G>A	p.G139S	c.740G>A	p.G247D	Mild	Irish/German	M	Tomatsu	2004c
c.415G>A	p.G139S	c.901G>T	p.G301C	Rapidly progressing	Spanish	M	Pajares	2012
c.415G>A	p.G139S	c.901G>T	p.G301C	Severe	Spanish	M	Dung	2013
c.415G>A	p.G139S	c.1219A>C	p.N407H	Severe	American black, American Caucasian	M	Dung	2013
c.421T>A	p.W141R	c.421T>A	p.W141R	Severe	Turkish	M	Bunge	1997
c.421T>A	p.W141R	c.421T>A	p.W141R		Turkish	M	Morrone	2014
c.422G>A	p.W141Ter	c.422G>A	p.W141Ter		British	M	Morrone	2014
c.422+1G>A	c.422+1G>A	n/a, reported as allele	n/a, reported as allele	Severe	Italian		Tomatsu	1997
c.422+2_+8del	p.W141CfsTer29	n/a, reported as allele	n/a, reported as allele	Severe	Canadian		Tomatsu	2005
c.423-1G>A	c.423-1G>A	c.423-1G>A	c.423-1G>A		Afghan	F	Fukuda	1997
c.423-1G>A	c.423-1G>A	c.423-1G>A	c.423-1G>A	Severe	Afghan	F	Yamada	1998
c.433C>T	p.H145Y	c.433C>T	p.H145Y		Canadian		Morrone	2014
c.442C>T	p.Q148Ter	c.121-2779_567-248, c.1140-730_1365-1530	double gene deletion	Severe	Japanese	M	Tomatsu	1996
c.442C>T	p.Q148Ter	c.121-2779_567-248, c.1140-730_1365-1530	double gene deletion		Japanese	M	Tomatsu	1996

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.451C>T	p.P151S	c.121-2779_567-248, c.1140-730_1365-1530	double gene deletion	Severe	Japanese	M	Tomatsu	1996
c.451C>T	p.P151S	c.121-2779_567-248, c.1140-730_1365-1530	double gene deletion		Japanese	F	Tomatsu	1996
c.451C>A	p.P151T	c.415G>A	p.G139S	Severe	Korean		Lee	2012
c.451C>A	p.P151T	c.415G>A	p.G139S	Severe	Korean		Lee	2012
c.451C>A	p.P151T	c.752G>A	p.R251Q		Korean	F	Park	2013
c.451C>A	p.P151T	c.1000C>T	p.Q334Ter	Severe	Korean		Lee	2012
c.451C>A	p.P151T	c.1000C>T	p.Q334Ter	Severe	Korean		Lee	2012
c.451C>A	p.P151T	c.1000C>T	p.Q334Ter		Korean	F	Park	2013
c.451C>A	p.P151T	c.1000C>T	p.Q334Ter		Korean	F	Park	2013
c.451C>A	p.P151T	c.1243-1G>A	c.1243-1G>A		Korean	F	Park	2013
c.452C>T	p.P151L	c.121-2779_567-248, c.1140-730_1365-1530	double gene deletion	Severe	Japanese	F	Tomatsu	1996
c.452C>T	p.P151L	c.121-2779_567-248, c.1140-730_1365-1530	double gene deletion		Japanese	M	Tomatsu	1996
c.452C>T	p.P151L	c.452C>T	p.P151L	Severe	Pakistani	F	Tomatsu	1995
c.452C>T	p.P151L	c.452C>T	p.P151L	Severe	Pakistani	M	Tomatsu	1995
c.452C>T	p.P151L	c.452C>T	p.P151L	Severe	Pakistani	M	Dung	2013
c.452C>T	p.P151L	c.452C>T	p.P151L		Asian-multiethnic	M	Morrone	2014
c.452C>T	p.P151L	c.452C>T	p.P151L		Asian-multiethnic	M	Morrone	2014
c.452C>T	p.P151L	c.452C>T	p.P151L		Asian-multiethnic	M	Morrone	2014
c.463G>A	p.G155R	c.423_566del	Exon 5 skipping	Moderate	Spanish	M	Pajares	2012
c.463G>A	p.G155R	c.463G>A	p.G155R	Severe	Bulgarian		Bunge	1997
c.463G>A	p.G155R	unknown	unknown	Severe	Chinese	M	Wang	2010

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.464G>A	p.G155E	unknown	unknown	Severe	Dutch/Irish/Scottish	F	Tomatsu	2004c
c.466T>C	p.F156L	c.466T>C	p.F156L	Severe	Middle Eastern	F	Morrone	2014
c.467T>C	p.F156S	c.872C>A	p.A291D	Attenuated			Tomatsu	2005
c.477G>T	p.W159C	c.477G>T	p.W159C		Saudi Arabia	F	Qubbaj	2008
c.477G>T	p.W159C	c.477G>T	p.W159C		Saudi Arabia	F	Qubbaj	2008
c.477G>T	p.W159C	c.477G>T	p.W159C		Saudi Arabia	F	Qubbaj	2008
c.477G>T	p.W159C	c.477G>T	p.W159C		Middle Eastern		Morrone	2014
c.477G>T	p.W159C	c.477G>T	p.W159C		Middle Eastern		Morrone	2014
c.477G>A	p.W159Ter	c.477G>A	p.W159Ter	Severe	American Caucasian	F	Dung	2013
not given	p.G161E	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.485C>T	p.S162F	c.485C>T	p.S162F		Colombian	F	Dung	2013
c.485C>T	p.S162F	c.485C>T	p.S162F	Severe	Colombian	M	Kato	1997
c.485C>T	p.S162F	c.485C>T	p.S162F	Severe	Colombian	M	Kato	1997
c.485C>T	p.S162F	unknown	unknown	Severe	Colombian	F	Kato	1997
c.488C>A	p.P163H	c.567-1G>T	c.567-1G>T	Intermediate	Chinese	M	Wang	2010
not given	p.P163H	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.489delC	p.N164TfsTer34	c.740G>A	p.G247D		Other	M	Morrone	2014
c.491A>C	p.N164T	c.491A>C	p.N164T	Severe	Brazilian	M	Tomatsu	2004a
c.494G>A	p.C165Y	c.494G>A	p.C165Y		Brazilian	F	Kubaski	2013
c.494G>A	p.C165Y	c.494G>A	p.C165Y		Brazilian		Morrone	2014
c.497A>G	p.H166R	c.497A>G	p.H166R		British	F	Morrone	2014

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.498delC	p.H166fsTer32	n/a, reported as allele	n/a, reported as allele	Severe	American		Tomatsu	2005
c.498C>G	p.H166Q	n/a, reported as allele	n/a, reported as allele	Severe			Tomatsu	2005
c.498C>G	p.H166Q	unknown	unknown	Severe	Irish/Danish	F	Tomatsu	2004c
c.499T>G	p.F167V	c.1140G>T	p.R380S	Attenuated			Tomatsu	2005
c.499T>G	p.F167V	c.1140G>T	p.R380S		Italian	F	Montaño	2007
c.499T>G	p.F167V	c.1140G>T	p.R380S		Italian	M	Montaño	2007
c.499T>G	p.F167V	n/a, reported as allele	n/a, reported as allele	Attenuated	Italian		Tomatsu	2005
c.499T>G	p.F167V	n/a, reported as allele	n/a, reported as allele	Attenuated	Italian		Tomatsu	2005
c.501_502insT	p.G168WfsTer3	unknown	unknown	Severe	German	M	Bunge	1997
c.502_503delGGinsTT	p.G168L	c.423_566del	Exon 5 skipping	Severe	Chinese	M	Wang	2010
not given	p.G168L	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.512A>C	p.D171A	c.512A>C	p.D171A	Attenuated			Tomatsu	2005
not given	p.D171A	not given	p.D171A	Mild			Sukegawa	2000
c.532_533delAT	p.I178PfsTer13	unknown	unknown		Lebanese	M	Bunge	1997
c.535C>T	p.P179S	c.1156C>T	p.R386C		Turkish	M	Morrone	2014
c.535C>T	p.P179S	unknown	unknown		Turkish		Terzioglu	2002
c.536C>A	p.P179H	c.536C>A	p.P179H	Severe			Bunge	1997
c.536C>T	p.P179L	c.536C>T	p.P179L	Severe		M	Yamada	1998
c.554A>G	p.E185G	n/a, reported as allele	n/a, reported as allele	Severe	British		Tomatsu	2005
c.[566+?_1003-?dup; 1139+?_1140-?del]	complex rearrangement: duplication from intron 5 to intron 9, deletion in intron 10	unknown	unknown	Severe	Middle Eastern	F	Morrone	2014
c.567-131ins42	c.567-131ins42	n/a, reported as allele	n/a, reported as allele	Undefined	Mexican		Tomatsu	2005
c.567-131ins42	c.567-131ins42	n/a, reported as allele	n/a, reported as allele	Undefined	Mexican		Tomatsu	2005

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.600delG	p.E202KfsTer115	unknown	unknown	Severe	New Zealander	F	Tomatsu	2004c
c.602G>A	p.G201E	c.602G>A	p.G201E		Middle Eastern		Morrone	2014
c.602G>A	p.G201E	c.602G>A	p.G201E		Middle Eastern		Morrone	2014
c.602G>A	p.G201E	c.602G>A	p.G201E		Middle Eastern		Morrone	2014
c.608C>T	p.A203V	n/a, reported as allele	n/a, reported as allele	Undefined	Brazilian		Tomatsu	2005
c.612C>G	p.N204K	c.612C>G	p.N204K		Japanese	M	Fukuda	1992
c.612C>G	p.N204K	c.612C>G	p.N204K	Mild	Japanese	M	Ogawa	1995
c.612C>G	p.N204K	c.612C>G	p.N204K	Mild	Japanese	M	Ogawa	1995
c.612C>G	p.N204K	c.612C>G	p.N204K	Attenuated			Tomatsu	2005
c.612C>G	p.N204K	c.612C>G	p.N204K		American Caucasian	F	Dung	2013
c.627C>G	p.Y209Ter	c.627C>G	p.Y209Ter				Morrone	2014
c.631C>T	p.Q211Ter	c.1120C>T	p.Q374Ter	Severe	Italian	M	Tomatsu	2004b
c.634-1G>T	p.E212VfsTer9	c.860C>T	p.S287L		Greek	F	Dung	2013
c.634-1G>A	c.634-1G>A	unknown	unknown	Severe	Chinese	M	Wang	2010
c.641T>C	p.L214P	c.641T>C	p.L214P		Turkish		Morrone	2014
c.647T>C	p.F216S	c.647T>C	p.F216S		Middle Eastern		Morrone	2014
c.688T>G	p.W230G	c.688T>G	p.W230G	Severe	Pakistani	F	Tomatsu	1995
c.688T>G	p.W230G	c.688T>G	p.W230G		Asian-multiethnic	F	Morrone	2014
c.697G>A	p.D233N	c.697G>A	p.D233N		Middle Eastern		Morrone	2014
c.697G>A	p.D233N	c.697G>A	p.D233N		Middle Eastern		Morrone	2014
c.697G>A	p.D233N	c.1034T>C	p.L345P	Severe	German/Polish	M	Tomatsu	2004c
c.697G>A	p.D233N	unknown	unknown	Severe	Polish	M	Dung	2013

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.706C>G	p.H236D	c.706C>G	p.H236D	Attenuated	Chinese	F	Wang	2010
c.708delC	p.H236RfsTer25	c.708delC	p.H236RfsTer25		Turkish		Terzioglu	2002
c.708delC	p.H236RfsTer25	c.708delC	p.H236RfsTer25		Turkish		Terzioglu	2002
c.715G>T	p.V239F	c.715G>T	p.V239F	Severe	Brazilian	M	Dung	2013
c.715G>T	p.V239F	n/a, reported as allele	n/a, reported as allele	Severe	Brazilian		Tomatsu	2005
c.715G>T	p.V239F	n/a, reported as allele	n/a, reported as allele	Severe	Brazilian		Tomatsu	2005
not given	p.Y240C	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.740G>A	p.G247D	c.415G>A	p.G139S	Attenuated			Tomatsu	2005
c.740G>A	p.G247D	c.489delC	p.N164TfsTer34	Severe	Norwegian	M	Bunge	1997
c.740G>A	p.G247D	c.761A>G	p.Y254C	Attenuated	American Caucasian	F	Dung	2013
c.740G>A	p.G247D	c.901G>T	p.G301C	Severe	American Caucasian, German/Swedish	F	Dung	2013
c.740G>A	p.G247D	c.[937A>G; 977G>C]	p[T313A; W326S]		Australian		Morrone	2014
c.740G>A	p.G247D	unknown	unknown		German	f	Bunge	1997
c.751C>T	p.R251Ter	c.268C>T	p.R90W		British	F	Morrone	2014
c.752G>A	p.R251Q	c.451C>A	p.P151T	Severe	Korean		Lee	2012
c.752G>A	p.R251Q	c.1155C>A	p.Y385Ter		British	F	Morrone	2014
c.752G>A	p.R251Q	c.1155C>A	p.Y385Ter		British	M	Morrone	2014
not given	p.R251Q	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.752G>A	p.R251Q	unknown	unknown		Irish	F	Morrone	2014
c.757C>T	p.R253W	c.757C>T	p.R253W	Attenuated			Tomatsu	2005
c.757C>T	p.R253W	c.757C>T	p.R253W		Pakistani	M	Montaño	2007

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.757C>T	p.R253W	c.757C>T	p.R253W		Pakistani	M	Montaño	2007
c.757C>T	p.R253W	c.757C>T	p.R253W		Pakistani	M	Montaño	2007
c.757C>T	p.R253W	c.757C>T	p.R253W		Pakistani	M	Montaño	2007
c.757C>T	p.R253W	n/a, reported as allele	n/a, reported as allele	Attenuated	Pakistani		Tomatsu	2005
c.757C>T	p.R253W	n/a, reported as allele	n/a, reported as allele	Attenuated	Pakistani		Tomatsu	2005
c.757C>T	p.R253W	n/a, reported as allele	n/a, reported as allele	Attenuated	Pakistani		Tomatsu	2005
c.757C>T	p.R253W	n/a, reported as allele	n/a, reported as allele	Attenuated	Pakistani		Tomatsu	2005
c.757C>T	p.R253W	n/a, reported as allele	n/a, reported as allele	Attenuated	Pakistani		Tomatsu	2005
c.757C>T	p.R253W	n/a, reported as allele	n/a, reported as allele	Attenuated	Pakistani		Tomatsu	2005
c.757C>T	p.R253W	n/a, reported as allele	n/a, reported as allele	Attenuated	Pakistani		Tomatsu	2005
c.757C>T	p.R253W	n/a, reported as allele	n/a, reported as allele	Attenuated	Pakistani		Tomatsu	2005
c.758G>A	p.R253Q	c.922T>C	p.C308R	Attenuated	American Caucasian	M	Dung	2013
c.761A>G	p.Y254C	c.423_566del	Exon 5 skipping	Moderate	Spanish	F	Pajares	2012
c.761A>G	p.Y254C	c.761A>G	p.Y254C	Rapidly progressing	Spanish	F	Pajares	2012
c.761A>G	p.Y254C	c.761A>G	p.Y254C	Rapidly progressing	Spanish	M	Pajares	2012
c.769G>A	p.A257T	unknown	unknown	Severe	Polish	F	Yamada	1998
c.776G>A	p.R259Q	c.280C>G	p.R94G	Mild	Polish	F	Bunge	1997
c.776G>A	p.R259Q	c.280C>G	p.R94G	Mild	Polish	M	Bunge	1997
c.776G>A	p.R259Q	c.280C>G	p.R94G	Intermediate	Polish	F	Tylki-Szymanska	1998
c.776G>A	p.R259Q	c.280C>G	p.R94G	Intermediate	Polish	M	Tylki-Szymanska	1998
c.776G>A	p.R259Q	c.776G>A	p.R259Q	Mild	Polish	F	Bunge	1997
c.776G>A	p.R259Q	c.776G>A	p.R259Q	Attenuated			Tomatsu	2005

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.776G>A	p.R259Q	c.901G>T	p.G301C		Caucasian	F	Lachman	2014
c.776G>A	p.R259Q	c.1520G>T	p.C507F	Attenuated	Hispanic	F	Morrone	2014
c.778G>A	p.E260K	c.1156C>T	p.R386C	Rapidly progressing	Spanish	M	Pajares	2012
c.780G>C	p.E260D	n/a, reported as allele	n/a, reported as allele	Undefined	Danish		Tomatsu	2005
c.791G>C	p.S264T	c.791G>C	p.S264T		British	M	Morrone	2014
c.841_867del	p.T281_N289del	c.841_867del	p.T281_N289del		French Canadian		Morrone	2014
c.841_867del	p.T281_N289del	c.884C>T	p.S295F	Mild	Dutch	M	Bunge	1997
c.841_867del	p.T281_N289del	c.899-1G>C	c.899-1G>C		British	F	Morrone	2014
c.850T>G	p.F284V	c.850T>G	p.F284V	Attenuated	Turkish	F	Dung	2013
c.850T>G	p.F284V	c.1275delA	p.V427SfsTer13		British	M	Morrone	2014
c.850T>G	p.F284V	unknown	unknown	Severe		M	Yamada	1998
c.850T>G	p.F284V	unknown	unknown	Attenuated			Tomatsu	2005
c.853_855delTTC	p.F285del	unknown	unknown	Severe	American Caucasian, British/German/Portuguese	M	Dung	2013
c.860C>T	p.S287L	c.758+4A>T	c.758+4A>T		Greek	M	Morrone	2014
c.860C>T	p.S287L	c.860C>T	p.S287L	Severe	Polish	F	Bunge	1997
c.860C>T	p.S287L	c.860C>T	p.S287L	Severe	Spanish	M	Pajares	2012
c.860C>T	p.S287L	c.860C>T	p.S287L	Severe	Macedonian	M	Dung	2013
c.860C>T	p.S287L	c.860C>T	p.S287L		Middle Eastern		Morrone	2014
c.860C>T	p.S287L	c.860C>T	p.S287L		Middle Eastern	M	Morrone	2014
c.860C>T	p.S287L	c.884C>T	p.S295F	Mild	Austrian	M	Tomatsu	2004c

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.860C>T	p.S287L	c.1055T>C	p.L352P	Severe	Irish/Italian/Polish	F	Tomatsu	2004c
c.860C>T	p.S287L	c.1168delC	p.L390Ter		Turkish	M	Morrone	2014
c.860C>T	p.S287L	c.1240C>T	p.Q414Ter		New Zealander		Morrone	2014
c.866A>G	p.N289S	c.866A>G	p.N289S	Attenuated	Chinese	F	Wang	2010
c.868G>A	p.G290S	c.953T>G	p.M318R		Chinese		He	2013
c.868G>A	p.G290S	n/a, reported as allele	n/a, reported as allele	Severe	British		Tomatsu	2005
c.868G>A	p.G290S	n/a, reported as allele	n/a, reported as allele	Severe	Japanese		Tomatsu	2005
c.871G>A	p.A291T	c.245C>T	p.S82L	Severe	Chinese	M	Wang	2010
c.871G>A	p.A291T	c.758+1G>C	c.758+1G>C	Severe	Japanese	M	Tomatsu	2004c
c.871G>A	p.A291T	c.871G>A	p.A291T	Severe	Pakistani	M	Tomatsu	1995
c.871G>A	p.A291T	c.871G>A	p.A291T		Asian-multiethnic	M	Morrone	2014
c.871G>A	p.A291T	c.871G>A	p.A291T		Asian-multiethnic	M	Morrone	2014
c.871G>A	p.A291T	c.871G>A	p.A291T		Asian-multiethnic	F	Morrone	2014
c.871G>A	p.A291T	c.871G>A	p.A291T		Asian-multiethnic	F	Morrone	2014
c.871G>A	p.A291T	c.871G>A	p.A291T		British	M	Morrone	2014
c.871G>A	p.A291T	c.871G>A	p.A291T		British	M	Morrone	2014
c.872C>A	p.A291D	c.467T>C	p.F156S	Mild	French	F	Bunge	1997
c.874delG	p.A292PfsTer26	c.874delG	p.A292PfsTer26		Turkish		Terzioglu	2002
c.884C>T	p.S295F	c.833_859del27	p.A277_N288del	Attenuated			Tomatsu	2005
c.884C>T	p.S295F	c.860C>T	p.S287L	Attenuated			Tomatsu	2005
c.898+1G>A	p.G300DfsTer34	c.121-1G>C	p.M41RfsTer46	Severe	Ukrainian	F	Dung	2013
c.898+1G>A	c.898+1G>A	c.898+1G>A	c.898+1G>A		Lebanese		Bunge	1997

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.898+1G>C	c.898+1G>C	c.898+1G>C	c.898+1G>C	Severe	Czech/German/Irish/Polish	M	Tomatsu	2004c
c.898+1G>C	c.898+1G>C	c.898+1G>C	c.898+1G>C	Attenuated			Tomatsu	2005
c.899-2A>G	c.899-2A>G	unknown	unknown	Severe	Mexican/Lebanese	M	Tomatsu	2004c
c.901G>T	p.G301C	c.423_566del	Exon 5 skipping	Moderate	Spanish	F	Pajares	2012
c.901G>T	p.G301C	c.498C>G	p.H166Q		Australian		Morrone	2014
c.901G>T	p.G301C	c.498C>G	p.H166Q		Australian		Morrone	2014
c.901G>T	p.G301C	c.901G>T	p.G301C	Severe	Moroccan	M	Bunge	1997
c.901G>T	p.G301C	c.901G>T	p.G301C	Severe	Portugese	F	Bunge	1997
c.901G>T	p.G301C	c.901G>T	p.G301C	Severe	Colombian	M	Kato	1997
c.901G>T	p.G301C	c.901G>T	p.G301C	Severe	Colombian	M	Kato	1997
c.901G>T	p.G301C	c.901G>T	p.G301C	Severe	Colombian	F	Kato	1997
c.901G>T	p.G301C	c.901G>T	p.G301C	Severe	Colombian	F	Kato	1997
c.901G>T	p.G301C	c.901G>T	p.G301C	Severe	Colombian	M	Kato	1997
c.901G>T	p.G301C	c.901G>T	p.G301C	Severe	Colombian	M	Kato	1997
c.901G>T	p.G301C	c.901G>T	p.G301C	Severe	Colombian	F	Kato	1997
c.901G>T	p.G301C	c.901G>T	p.G301C	Severe	French	F	Tomatsu	2004c
c.901G>T	p.G301C	c.901G>T	p.G301C	Moderate	African-Varied	M	Pajares	2012
c.901G>T	p.G301C	c.901G>T	p.G301C	Severe	American Caucasian	F	Dung	2013
c.901G>T	p.G301C	c.901G>T	p.G301C	Severe	Brazilian	M	Dung	2013
c.901G>T	p.G301C	c.901G>T	p.G301C	Rapidly progressing	Portuguese	F	Morrone	2014

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.901G>T	p.G301C	c.901G>T	p.G301C	Rapidly progressing/ moderate	Portuguese	M	Morrone	2014
c.901G>T	p.G301C	c.901G>T	p.G301C	Rapidly progressing/ moderate	Portuguese	M	Morrone	2014
c.901G>T	p.G301C	c.1156C>T	p.R386C	Moderate	Spanish	F	Pajares	2012
c.901G>T	p.G301C	c.1171A>G	p.M391V		French Canadian		Morrone	2014
c.901G>T	p.G301C	c.1171A>G	p.M391V		French Canadian		Morrone	2014
c.901G>T	p.G301C	c.1180A>C	p.T394P	Rapidly progressing	Spanish	F	Pajares	2012
c.901G>T	p.G301C	unknown	unknown		Caucasian	M	Lachman	2014
c.901G>T	p.G301C	unknown	unknown	Severe	Colombian	M	Kato	1997
c.901G>T	p.G301C	unknown	unknown	Severe	French	M	Tomatsu	2004c
not given	p.G304D	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.920T>C	p.L307P	c.1023C>G	p.S341R	Severe	Brazilian	M	Tomatsu	2004a
c.920T>C	p.L307P	c.1023C>G	p.S341R	Severe	Brazilian	M	Tomatsu	2004a
c.925G>A	p.G309R	c.422G>A	p.W141Ter		British	M	Morrone	2014
c.925G>A	p.G309R	n/a, reported as allele	n/a, reported as allele	Severe	British		Tomatsu	2005
c.930G>C	p.K310N	c.1156C>T	p.R386C	Attenuated			Tomatsu	2005
c.930G>C	p.K310N	n/a, reported as allele	n/a, reported as allele	Attenuated	American		Tomatsu	2005
c.934A>G	p.T312A	c.1019G>A	p.G340D	Severe	Chinese	M	Wang	2010
c.935C>G	p.T312S	c.154_165del12	p.P52_E55del	Mild	Irish	F	Tomatsu	2004c
c.935C>G	p.T312S	c.154_165del12	p.P52_E55del	Attenuated			Tomatsu	2005
c.935C>G	p.T312S	c.209_220del	p.N71_S74delinsT	Attenuated			Tomatsu	2005
c.935C>G	p.T312S	c.337A>T	p.I113F	Attenuated			Tomatsu	2005

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.935C>G	p.T312S	c.935C>G	p.T312S	Mild	British/Irish	M	Yamada	1998
c.935C>G	p.T312S	c.935C>G	p.T312S	Attenuated			Tomatsu	2005
c.935C>G	p.T312S	c.935C>G	p.T312S		Irish	M	Morrone	2014
c.935C>G	p.T312S	c.935C>G	p.T312S		Irish	F	Morrone	2014
c.935C>G	p.T312S	c.1106T>C	p.L369P		Irish	F	Morrone	2014
c.935C>G	p.T312S	c.1106T>C	p.L369P		Irish	M	Morrone	2014
c.935C>G	p.T312S	unknown	unknown	Intermediate	British/Irish	F	Yamada	1998
c.935C>G	p.T312S	unknown	unknown	Intermediate	Irish	F	Yamada	1998
c.938C>T	p.T313M	c.938C>T	p.T313M	Severe	Canadian Caucasian, Irish/English	M	Dung	2013
not given	p.E315K	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.947G>T	p.G316V	unknown	unknown		Chinese	M	Wang	2010
c.949G>A	p.G317R	n/a, reported as allele	n/a, reported as allele				Pollard	2013
c.951dupG	c.951dupG	n/a, reported as allele	n/a, reported as allele				Pollard	2013
c.953T>G	p.M318R	c.106_111del	p.L36_L37del	Severe	Chinese	M	Wang	2010
c.953T>G	p.M318R	c.953T>G	p.M318R		Other	M	Morrone	2014
c.953T>G	p.M318R	c.953T>G	p.M318R		Taiwanese		Morrone	2014
c.953T>G	p.M318R	c.953T>G	p.M318R		South-East Asian		Morrone	2014
c.953T>G	p.M318R	c.953T>G	p.M318R		Chinese		He	2013
c.953T>G	p.M318R	c.953T>G	p.M318R		Chinese		He	2013
c.953T>G	p.M318R	c.1567T>G	p.Ter523EextTer93	Severe	Chinese	F	Dung	2013
not given	p.M318R	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.953T>G	p.M318R	unknown	unknown	Severe	Japanese	F	Ogawa	1995
c.953T>G	p.M318R	unknown	unknown		Chinese		He	2013
c.953T>G	p.M318R	unknown	unknown		Chinese		He	2013
not given	p.A324E	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.971C>A	p.A324E	unknown	unknown	Mild	Chinese	M	Wang	2010
c.974G>A	p.W325Ter	c.974G>A	p.W325Ter	Severe	Chinese	F	Wang	2010
c.975G>T	p.W325C	c.1156C>T	p.R386C	Attenuated	Canadian Caucasian	M	Dung	2013
c.975G>T	p.W325C	n/a, reported as allele	n/a, reported as allele	Undefined	Canadian		Tomatsu	2005
c.976_978delTGG	p.W326del	unknown	unknown	Intermediate	Dutch	M	Bunge	1997
c.1000C>T	p.Q334Ter	c.853_855delTTC	p.F285del	Severe	Korean		Lee	2012
c.1000C>T	p.Q334Ter	c.1156C>T	p.R386C	Severe	Korean		Lee	2012
c.1000C>T	p.Q334Ter	c.1156C>T	p.R386C		Korean	F	Park	2013
c.1003-42C>T	c.1003-42C>T	c.1003-42C>T	c.1003-42C>T	Severe	Italian		Tomatsu	1997
c.1003-2A>C	c.1003-2A>C	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.1012C>T	p.Q338Ter	unknown	unknown	Severe	Norwegian	F	Bunge	1997
c.1019G>A	p.G340D	c.1019G>A	p.G340D	Severe	Chinese	M	Wang	2010
c.1019G>A	p.G340D	c.1019G>A	p.G340D	Severe	Chinese	F	Wang	2010
c.1019G>A	p.G340D	c.1019G>A	p.G340D	Severe	Chinese	M	Wang	2010
c.1023C>G	p.S341R	c.1023C>G	p.S341R	Severe	Brazilian	M	Dung	2013
c.1023C>G	p.S341R	c.1156C>T	p.R386C	Severe	Brazilian	M	Dung	2013
c.1023C>G	p.S341R	n/a, reported as allele	n/a, reported as allele	Severe	British		Tomatsu	2005
c.1023C>G	p.S341R	n/a, reported as allele	n/a, reported as allele	Severe	British		Tomatsu	2005

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.1023C>G	p.S341R	unknown	unknown		Sri Lankan	M	Dung	2013
c.1023C>G	p.S341R	unknown	unknown		Sri Lankan	F	Dung	2013
c.1028T>G	p.M343R	c.1038C>A	p.F346L	Severe	Japanese	F	Ogawa	1995
c.1032C>G	p.D344E	c.286G>T	p.G96C	Severe	Vietnam		Bunge	1997
c.1046delG	c.1046delG	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.1052C>T	p.A351V	n/a, reported as allele	n/a, reported as allele	Severe	Italian		Tomatsu	2005
c.1055T>C	p.L352P	c.860C>T	p.S287L	Attenuated			Tomatsu	2005
c.1070delC	p.P357RfsTer21	c.1070delC	p.P357RfsTer21	Severe	Middle Eastern	M	Morrone	2014
c.[1070C>T; c.1156C>T]	p.[P357L; p.R386C]	c.[1070C>T; c.1156C>T]	p.[P357L; p.R386C]	Severe	Italian	F	Tomatsu	2004b
c.1070C>T	p.P357L	c.1156C>T	p.R386C	Moderate	Spanish	M	Morrone	2014
c.1082G>T	p.R361G	c.1082G>T	p.R361G		German	F	Bunge	1997
c.1097T>C	p.L366P	c.1097T>C	p.L366P	Severe	Chinese		He	2013
c.1097T>C	p.L366P	c.1097T>C	p.L366P	Severe	Chinese		He	2013
not given	p.L366P	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.1106T>C	p.L369P	n/a, reported as allele	n/a, reported as allele	Severe	Brazilian		Tomatsu	2005
c.1106T>C	p.L369P	n/a, reported as allele	n/a, reported as allele	Severe	Dutch/Irish/Scottish /French		Tomatsu	2005
c.1114delC	p.L372SfsTer6	c.1114delC	p.L372SfsTer6	Rapidly progressing	Portuguese	M	Morrone	2014
c.1127G>A	p.R376Q	c.337A>T	p.I113F	Severe	Irish	M	Tomatsu	2004c
c.1127G>A	p.R376Q	c.1127G>A	p.R376Q	Severe	British/Irish	M	Yamada	1998
c.1138A>G	p.R380G	c.1138A>G	p.R380G	Rapidly progressing	Cape Verdean	M	Morrone	2014
c.1139G>C	p.R380T	c.1139G>C	p.R380T		Asian-multiethnic	F	Morrone	2014
c.1139G>C	p.R380T	n/a, reported as allele	n/a, reported as allele	Severe	Brazilian-Asian		Tomatsu	2005

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.1140G>T	p.R380S	c.423G>T	p.W141C	Mild	Argentine	M	Tomatsu	2004a
c.1140G>T	p.R380S	c.423G>T	p.W141C	Attenuated			Tomatsu	2005
c.1140G>T	p.R380S	c.1355_1356insT	p.F452fsTer42	Mild	Argentine	M	Tomatsu	2004a
c.1140G>T	p.R380S	c.1355_1356insT	p.F452fsTer42	Attenuated			Tomatsu	2005
c.1142delC	p.P381LfsTer9	c.1142delC	p.P381LfsTer9	Severe	Portuguese	F	Morrone	2014
c.1156C>T	p.R386C	c.423_566del	Exon 5 skipping	Moderate	Spanish	M	Morrone	2014
c.1156C>T	p.R386C	c.502G>A	p.G168R	Severe	Polish	M	Bunge	1997
c.1156C>T	p.R386C	c.530A>G	p.N177S		Chinese		He	2013
c.1156C>T	p.R386C	c.1156C>T	p.R386C	Severe	Turkish	M	Bunge	1997
c.1156C>T	p.R386C	c.1156C>T	p.R386C		Polish	F	Bunge	1997
c.1156C>T	p.R386C	c.1156C>T	p.R386C	Severe	British		Tomatsu	1997
c.1156C>T	p.R386C	c.1156C>T	p.R386C	Severe	Italian	F	Tomatsu	2004b
c.1156C>T	p.R386C	c.1156C>T	p.R386C	Severe	Italian	F	Tomatsu	2004b
c.1156C>T	p.R386C	c.1156C>T	p.R386C	Severe	Argentine	F	Tomatsu	2004a
c.1156C>T	p.R386C	c.1156C>T	p.R386C	Severe	Argentine	M	Tomatsu	2004a
c.1156C>T	p.R386C	c.1156C>T	p.R386C	Severe	Chilean	M	Tomatsu	2004a
c.1156C>T	p.R386C	c.1156C>T	p.R386C	Severe	Colombian	F	Tomatsu	2004a
c.1156C>T	p.R386C	c.1156C>T	p.R386C	Rapidly progressing	Spanish	F	Pajares	2012
c.1156C>T	p.R386C	c.1156C>T	p.R386C	Severe	American Caucasian, Hispanic/Greek	F	Dung	2013
c.1156C>T	p.R386C	c.1156C>T	p.R386C	Moderate	Spanish	M	Morrone	2014
c.1156C>T	p.R386C	c.1156C>T	p.R386C		Chinese		He	2013

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.1156C>T	p.R386C	c.1157G>A	p.R386H	Severe	Chilean	M	Tomatsu	2004a
c.1156C>T	p.R386C	c.1209_1210delGAinsTT	p.W403_T404delinsCS		Chinese		He	2013
c.1156C>T	p.R386C	c.1219A>C	p.N407H	Severe	French	M	Dung	2013
c.1156C>T	p.R386C	c.1219A>C	p.N407H	Slowly progressing	Portuguese	M	Morrone	2014
c.1156C>T	p.R386C	c.1364+1G>A	c.1364+1G>A	Severe	German	F	Bunge	1997
c.1156C>T	p.R386C	c.1483A>T	p.N495Y	Slowly progressing	Spanish	M	Pajares	2012
c.1156C>T	p.R386C	large deletion	large deletion	Severe	Japanese	M	Ogawa	1995
c.1156C>T	p.R386C	large deletion	large deletion	Severe	Japanese	M	Fukuda	1996
c.1156C>T	p.R386C	unknown	unknown	Mild	Argentine	M	Tomatsu	2004a
c.1156C>T	p.R386C	unknown	unknown	Mild	Argentine	M	Tomatsu	2004a
c.1156C>T	p.R386C	unknown	unknown	Severe	Brazilian	F	Tomatsu	2004a
c.1156C>T	p.R386C	unknown	unknown	Severe	Colombian	M	Tomatsu	2004a
c.1156C>T	p.R386C	unknown	unknown	Severe	Mexican	M	Tomatsu	2004c
c.1156C>T	p.R386C	unknown	unknown	Intermediate	Chinese	M	Wang	2010
c.1157G>A	p.R386H	unknown	unknown		British	M	Morrone	2014
c.1162G>A	p.D388N	c.319G>A	p.A107T	Attenuated			Tomatsu	2005
c.1168delC	p.L390Ter	c.1168delC	p.L390Ter	Severe	Turkish	M	Bunge	1997
not given	p.L390P	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.1168delC	p.L390Ter	unknown	unknown		Turkish		Morrone	2014
c.1171A>G	p.M391V	c.331C>T	p.Q111Ter	Attenuated			Tomatsu	2005
c.1171A>G	p.M391V	c.405_422+1del	c.405_422+1del		French Canadian		Morrone	2014
c.1171A>G	p.M391V	c.704C>A	p.T235K		French Canadian		Morrone	2014

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.1171A>G	p.M391V	c.841_867del	p.T281_N289del		French Canadian		Morrone	2014
c.1171A>G	p.M391V	c.1012C>T	p.Q338Ter	Mild	Irish/German/Swedish	F	Tomatsu	2004c
c.1171A>G	p.M391V	c.1171A>G	p.M391V	Mild	French	F	Tomatsu	2004c
c.1171A>G	p.M391V	c.1171A>G	p.M391V	Mild	French	M	Tomatsu	2004c
c.1171A>G	p.M391V	c.1171A>G	p.M391V	Attenuated			Tomatsu	2005
c.1171A>G	p.M391V	c.1171A>G	p.M391V	Severe	American Caucasian, German	F	Dung	2013
c.1171A>G	p.M391V	c.1171A>G	p.M391V		French Canadian		Morrone	2014
c.1171A>G	p.M391V	c.1354T>A	p.F452I	Attenuated	Canadian Caucasian	M	Dung	2013
c.1171A>G	p.M391V	c.1354T>A	p.F452I	Attenuated	Canadian Caucasian	F	Dung	2013
c.1171A>G	p.M391V	unknown	unknown		Canadian Caucasian	M	Dung	2013
c.1175C>T	p.A392V	c.1175C>T	p.A392V		Asian-multiethnic	F	Morrone	2014
c.1175C>T	p.A392V	c.1175C>T	p.A392V		Asian-multiethnic	F	Morrone	2014
c.1175C>T	p.A392V	c.1175C>T	p.A392V		Asian-multiethnic	M	Morrone	2014
c.1175C>T	p.A392V	c.1175C>T	p.A392V	Severe	Chinese		He	2013
c.1176_1178del3	p.A392del	c.1176_1178del3	p.A392del	Severe	Middle Eastern	M	Morrone	2014
c.1176_1178del3	p.A392del	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.1177_1178insT	p.A393CfsTer23	c.1177_1178insT	p.A393CfsTer23		Turkish		Terzioglu	2002
c.1184T>C	p.L395P	n/a, reported as allele	n/a, reported as allele	Undefined	Caucasian		Tomatsu	2005
c.1188delG	c.1188delG	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.1192C>G	p.H398D	c.1192C>G	p.H398D	Mild	Pakistani	M	Tomatsu	2004c
c.1192C>G	p.H398D	c.1192C>G	p.H398D	Mild	Pakistani	F	Tomatsu	2004c

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.1192C>G	p.H398D	c.1192C>G	p.H398D	Attenuated			Tomatsu	2005
c.1201C>T	p.H401Y	n/a, reported as allele	n/a, reported as allele	Undefined	Italian		Tomatsu	2005
c.1209_1210delGAinsTT	p.W403_T404delinsCS	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.1214G>A	p.W405Ter	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.1219A>C	p.N407H	c.1219A>C	p.N407H	Intermediate	German	F	Bunge	1997
c.1219A>C	p.N407H	c.1559G>A	p.W520Ter		British	M	Morrone	2014
c.1219A>C	p.N407H	n/a, reported as allele	n/a, reported as allele	Severe			Tomatsu	2005
c.1219A>C	p.N407H	n/a, reported as allele	n/a, reported as allele	Severe			Tomatsu	2005
c.1219A>C	p.N407H	unknown	unknown	Intermediate	Dutch	F	Bunge	1997
c.1226G>C	p.W409S	c.1226G>C	p.W409S	Attenuated			Tomatsu	2005
c.1243-1G>A	c.1243-1G>A	c.451C>A	p.P151T	Severe	Korean		Lee	2012
c.1243-1G>A	c.1243-1G>A	unknown	unknown	Severe	French	F	Bunge	1997
c.1243-1G>C	c.1243-1G>C	unknown	unknown		Chinese		He	2013
c.1244G>T	p.G415V	c.1244G>T	p.G415V		Portuguese	M	Morrone	2014
c.1247T>C	p.I416T	c.1247T>C	p.I416T		Asian-multiethnic	M	Morrone	2014
c.1259C>G	p.P420R	c.1259C>G	p.P420R		Asian-multiethnic	F	Morrone	2014
not given	p.P420S	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.1262G>A	p.G421E	n/a, reported as allele	n/a, reported as allele				Lissens	1999
c.1264C>A	p.Q422K	c.425A>G	p.H142R	Severe	Chinese	M	Wang	2010
c.1264C>T	p.Q422Ter	c.1264C>T	p.Q422Ter	Severe	Chinese	M	Wang	2010
c.1290_1291delCA	p.H430QfsTer71	c.1030G>A	p.D344N	Severe	Japanese	M	Ogawa	1995
c.1290_1291delCA	p.H430QfsTer71	c.1290_1291delCA	p.H430QfsTer71		Japanese	M	Fukuda	1992

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.1290_1291delCA	p.H430QfsTer71	c.1290_1291delCA	p.H430QfsTer71	Severe	Japanese	F	Ogawa	1995
c.1349A>T	p.E450V	n/a, reported as allele	n/a, reported as allele	Severe	Japanese		Tomatsu	2005
c.1349A>T	p.E450V	unknown	unknown	Severe	Japanese	M	Ogawa	1995
c.1354T>C	p.F452L	c.866A>G	p.G290S	Severe	Chinese	M	Wang	2010
c.1354T>C	p.F452L	c.1019G>A	p.G340D	Severe	Chinese	M	Wang	2010
c.1354T>A	p.F452I	c.1485C>G	p.N495K	Attenuated	Canadian Caucasian	F	Dung	2013
c.1354T>A	p.F452I	n/a, reported as allele	n/a, reported as allele	Severe	American		Tomatsu	2005
not given	p.F452L	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
not given	p.L454P	n/a, reported as allele	n/a, reported as allele		Chinese		Ye	2013
c.1408T>C	p.S470P	n/a, reported as allele	n/a, reported as allele	Undefined	American		Tomatsu	2005
c.1417C>T	p.Q473Ter	c.1417C>T	p.Q473Ter		Turkish		Terzioglu	2002
c.1417C>T	p.Q473Ter	c.1460A>G	p.N487S				Tomatsu	1995
c.1429_1455del	p.E477_Q485del	c.1429_1455del	p.E477_Q485del	Severe	Middle Eastern	F	Morrone	2014
c.1450C>T	p.P484S	c.1A>G	p.M1V	Attenuated			Tomatsu	2005
c.1450C>T	p.P484S	n/a, reported as allele	n/a, reported as allele	Attenuated	Japanese		Tomatsu	2005
c.1474G>A	p.A492T	c.1474G>A	p.A492T	Severe	Middle Eastern	M	Morrone	2014
c.1480A>G	p.M494V	c.1480A>G	p.M494V	Severe	Turkish	M	Bunge	1997
c.1480A>G	p.M494V	c.1480A>G	p.M494V		Turkish		Morrone	2014
c.1485C>G	p.N495K	c.1485C>G	p.N495K	Severe	Middle Eastern	M	Morrone	2014
c.1498G>A	p.G500S	c.1498G>A	p.G500S		Moroccan	F	Morrone	2014
c.1560G>A	p.W520Ter	c.868G>C	p.G290R		Chinese		He	2013
c.1567T>G	p.Ter523EextTer93	c.374C>T	p.P125L		Chinese	F	Zhao	2011

1st allele		2nd allele		Assessed growth phenotype	Reported country/ethnicity	Sex	First author	Year
Base change	Predicted effect on protein	Base change	Predicted effect on protein					
c.1568A>G	p.Ter523Wext	c.1568A>G	p.Ter523Wext	Severe	Korean		Lee	2012
not given	p.Ter523EextTer93	unknown	unknown		Chinese		He	2013
c.422+2_+3insT	p.W141fsTer41	c.1195delA	p.K399fsTer41	Severe	Iraqi	M	Dung	2013
c.422+2_+3insT	p.W141fsTer41	c.1195delA	p.K399fsTer41	Severe	Iraqi	F	Dung	2013
double deletion	double deletion	c.413T>C	p.V138A	Intermediate	Japanese	M	Ogawa	1995
double deletion	double deletion	c.442C>T	p.Q148Ter	Severe	Japanese	M	Ogawa	1995
double deletion	double deletion	c.451C>T	p.P151S	Severe	Japanese	M	Ogawa	1995
double deletion	double deletion	double deletion	double deletion	Severe	Japanese	M	Ogawa	1995
unknown	unknown	c.917T>C	p.F306S		Chinese		He	2013
unknown	unknown	c.953T>G	p.M318R		Chinese		He	2013
unknown	unknown	not given	p.L400RfsTer54		Chinese		He	2013

Supp. Table S3. Frequency of *GALNS* alleles reported from patients with Morquio A

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.1A>G	p.M1V	1	10	Italian, Japanese, unknown	(Montaño et al., 2007; Tomatsu et al., 2004b; Tomatsu et al., 2005)
c.3G>A	p.M1I	1	2	Chinese, Finnish	(Dung et al., 2013; Ye et al., 2013)
c.29G>A	p.W10Ter	1	7	Austrian, Italian, unknown	(Dung et al., 2013; Tomatsu et al., 2004b; Tomatsu et al., 2005; Bunge et al., 1997)
c.43C>A	p.L15M	1	2	Scottish, unknown	(Tomatsu et al., 2005)
c.47T>A	p.V16E	1	2	Brazilian	(Morrone et al., 2014)
c.67G>C	p.G23R	1	2	Italian, unknown	(Tomatsu et al., 2005; Tomatsu et al., 2004b)
not given	p.N32D	1	1	Chinese	(Ye et al., 2013)
c.106_111del	p.L36_L37del	1	4	Chinese, Taiwanese	(Guo et al., 2012; Wang et al., 2010; Yang et al., 2001)
c.107T>C	p.L36P	1	2	Mexican, unknown	(Tomatsu et al., 2005)
c.107T>G	p.L36R	1	6	Asian-multiethnic	(Morrone et al., 2014)
c.118G>A	p.D40N	1	2	Asian-multiethnic	(Morrone et al., 2014)
c.120+1G>A	c.120+1G>A	Intron 1	23	Tunisian, unknown	(Carraresi et al., 2008; Khedhiri et al., 2011; Laradi et al., 2006)
c.120+1G>C	c.120+1G>C	Intron 1	2	Middle Eastern	(Morrone et al., 2014)
c.120_121ins158	p.D40fsTer20	Intron 1	2	Japanese, Mexican	(Tomatsu et al., 2005)
c.121-1G>A	c.121-1G>A	Intron 1	4	Middle Eastern	(Morrone et al., 2014)
c.121-2A>G	c.121-2A>G	Intron 1	4	Japanese	(Ogawa et al., 1995)
c.121-2779_567-248, c.1140-730_1365-1530	double gene deletion	Intron 1	8	Japanese	(Tomatsu et al., 1996; Tomatsu et al., 2005)
c.121-?_244+?del	p.M41_P81>RfsTer47	Intron 1	2	Japanese	(Tomatsu et al., 2005)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.121A>T	p.M41L	2	8	Acadian, American Caucasian, unknown	(Dung et al., 2013; Tomatsu et al., 2004c; Tomatsu et al., 2005)
c.121-1G>A	p.M41RfsTer46	2	3	American Caucasian	(Dung et al., 2013)
c.121-1G>C	c.121-1G>C	2	2	Ukrainian	(Dung et al., 2013; Tomatsu et al., 2005)
c.122T>A	p.M41K	2	2	American Caucasian	(Dung et al., 2013)
c.125G>A	p.G42E	2	2	Italian	(Dung et al., 2013; Tomatsu et al., 2005)
c.139G>A	p.G47R	2	7	Honduran, Polish, Spanish	(Bunge et al., 1997; Dung et al., 2013; Pajares et al., 2012; Tomatsu et al., 2004c)
c.143T>G	p.V48G	2	1	British	(Morrone et al., 2014)
c.151G>A	p.E51K	2	2	Brazilian	(Morrone et al., 2014)
c.154_165del12	p.P52_E55del	2	1	Irish	(Tomatsu et al., 2004c)
c.158C>T	p.S53F	2	1	Japanese	(Tomatsu et al., 2005)
not given	p.D60E	2	1	Chinese	(Ye et al., 2013)
c.178G>A	p.D60N	2	9	Finnish, unknown	(Bunge et al., 1997; Montaño et al., 2003; Tomatsu et al., 2005)
c.181C>T	p.R61W	2	4	American, American Caucasian, unknown	(Dung et al., 2013; Tomatsu et al., 2005)
c.196G>A	p.G66R	2	5	Tunisian	(Khedhiri et al., 2011; Khedhiri et al., 2012; Laradi et al., 2006)
c.[199C>A, c.1226G>C]	p.[L67M, p.W409S]	[2; 11]	2	Dutch	(Bunge et al., 1997)
c.205T>G	p.F69V	2	2	Colombian	(Kato et al., 1997; Tomatsu et al., 2004c)
c.209_220del	p.N71_S74delinsT	2	2	Irish, unknown	(Tomatsu et al., 2005)
c.218A>G	p.Y73C	2	2	Korean	(Lee et al., 2012; Park et al., 2013)
c.221C>T	p.S74F	2	1	Spanish	(Pajares et al., 2012)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.224C>G	p.A75G	2	4	Colombian	(Tomatsu et al., 2004a)
c.230C>G	p.P77R	2	10	Asian-multiethnic, Caucasian, Middle Eastern, Saudi Arabian	(Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 2004c)
c.235_236delTG	p.C79LfsTer78	2	1	Dutch	(Bunge et al., 1997)
c.236G>A	p.C79Y	2	3	Argentine, Italian	(Catarzi et al., 2012; Tomatsu et al., 2004a)
c.239C>T	p.S80L	2	2	British	(Morrone et al., 2014; Tomatsu et al., 2005)
c.242C>T	p.P81L	2	3	Turkish, unknown	(Morrone et al., 2014)
c.244T>C	p.S82P	2	2	Canadian	(Dung et al., 2013)
c.245C>T	p.S82L	3	3	American Caucasian, Chinese	(Dung et al., 2013; Wang et al., 2010)
c.251C>A	p.A84E	3	2	Portuguese	(Morrone et al., 2014)
c.253G>A	p.A85T	3	2	Tunisian	(Khedhiri et al., 2011)
c.257T>C	p.L86P	3	1	Finnish	(Dung et al., 2013)
c.263C>T	p.T88I	3	1	Chinese	(Wang et al., 2010)
c.268C>T	p.R90W	3	8	Asian-multiethnic, Australian, British, Pakistani, Turkish	(Morrone et al., 2014; Tomatsu et al., 1995; Tomatsu et al., 2004c)
c.272T>C	p.L91P	3	2	Portuguese	(Morrone et al., 2014)
c.278T>A	p.I93N	3	1	American Caucasian: Albanian	(Dung et al., 2013)
c.280C>T	p.R94C	3	11	Brazilian, Canadian, Caucasian, Middle Eastern, unknown	(Cole et al., 1996; Dung et al., 2013; Morrone et al., 2014; Ogawa et al., 1995; Tomatsu et al., 2005)
c.280C>G	p.R94G	3	8	Japanese, Polish, unknown	(Bunge et al., 1997; Ogawa et al., 1995; Tomatsu et al., 2004c; Tomatsu et al., 2005; Tylki-Szymanska et al., 1998)
c.281G>T	p.R94L	3	3	Portuguese, Turkish	(Morrone et al., 2014; Tomatsu et al., 2005)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.286G>T	p.G96C	3	1	Vietnam	(Bunge et al., 1997)
c.287G>T	p.G96V	3	3	Irish	(Tomatsu et al., 1995; Yamada et al., 1998)
c.289T>G	p.F97V	3	2	Canadian, unknown	(Cole et al., 1996; Tomatsu et al., 2005)
c.319G>A	p.A107T	3	7	British, Chinese, unknown	(Morrone et al., 2014; Tomatsu et al., 2004b; Tomatsu et al., 2005; Wang et al., 2010)
c.320–1G>T	p.A107GfsTer54	Intron 3	1	Chilean	(Dung et al., 2013)
c.331C>T	p.Q111Ter	4	2	British, unknown	(Morrone et al., 2014; Tomatsu et al., 2005)
c.332A>G	p.Q111R	4	4	Dutch, unknown	(Bunge et al., 1997; Tomatsu et al., 2005)
c.334delG	p.E112RfsTer15	4	8	Turkish	(Terzioglu et al., 2002)
c.337A>T	p.I113F	4	52	American Caucasian, Australian, British, British/German, British/Irish, Irish, unknown	(Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 1995; Tomatsu et al., 2004c; Tomatsu et al., 2005; Yamada et al., 1998)
c.346G>A	p.G116S	4	12	American Caucasian, American Caucasian: Italian, Asian-multiethnic, Brazilian, Portuguese, Turkish	(Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 2004a)
c.347G>T	p.G116V	4	14	Asian-multiethnic, Norwegian	(Morrone et al., 2014)
c.363G>C	p.E121D	4	2	African-Varied	(Pajares et al., 2012)
c.374C>T	p.P125L	4	9	Chinese, Italian, Japanese, unknown	(Dung et al., 2013; He et al., 2013; Tomatsu et al., 1997; Tomatsu et al., 2004b; Zhao et al., 1990)
c.376G>T	p.E126Ter	4	1	Australian	(Morrone et al., 2014)
c.385A>T	p.K129Ter	4	1	unknown	(Carraresi et al., 2008)
c.405_422+1del	c.405_422+1del	4	2	Canadian Caucasian, French Canadian	(Dung et al., 2013; Morrone et al., 2014)
c.405C>G	p.S135R	4	1	unknown	(Yamada et al., 1998)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.409_420del12	c.409_c.420del12	4	1	Chinese	(Ye et al., 2013)
c.413T>C	p.V138A	4	3	Japanese	(Ogawa et al., 1995; Tomatsu et al., 1996; Tomatsu et al., 2005)
c.415G>A	p.G139S	4	16	American black, American Caucasian, Argentine, Brazilian, Chinese, Irish/German, Korean, Spanish, unknown	(Dung et al., 2013; Lee et al., 2012; Morrone et al., 2014; Pajares et al., 2012; Tomatsu et al., 2004a; Tomatsu et al., 2004c; Tomatsu et al., 2005; Wang et al., 2010)
c.421T>A	p.W141R	4	4	Turkish	(Bunge et al., 1997; Morrone et al., 2014)
c.422G>A	p.W141Ter	4	3	British	(Morrone et al., 2014)
c.422+2_+8del	p.W141CfsTer29	Intron 4	1	Canadian	(Tomatsu et al., 2005)
c.422+2_+3insT	p.W141fsTer41	Intron 4	2	Iraqi	(Dung et al., 2013)
c.423-1G>A	c.423-1G>A	Intron 4	4	Afghan	(Fukuda et al., 1996; Yamada et al., 1998)
c.423_566del	Exon 5 skipping		6	Chinese, Spanish	(Morrone et al., 2014; Pajares et al., 2012; Wang et al., 2010)
c.423G>T	p.W141C	5	2	Argentine, unknown	(Tomatsu et al., 2004a; Tomatsu et al., 2005)
c.425A>G	p.H142R	5	1	Chinese	(Wang et al., 2010)
c.433C>T	p.H145Y	5	2	Canadian	(Morrone et al., 2014)
c.442C>T	p.Q148Ter	5	3	Japanese	(Ogawa et al., 1995; Tomatsu et al., 1996; Tomatsu et al., 2004a)
c.448C>T	p.H150Y	5	2	Italian, unknown	(Tomatsu et al., 2004b; Tomatsu et al., 2005)
c.451C>T	p.P151S	5	3	Japanese	(Ogawa et al., 1995; Tomatsu et al., 1996; Cole et al., 1996)
c.451C>A	p.P151T	5	10	Korean	(Lee et al., 2012; Park et al., 2013)
c.452C>T	p.P151L	5	15	Asian-multiethnic, Japanese, Pakistani	(Cole et al., 1996; Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 1995; Tomatsu et al., 1996)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.463G>A	p.G155R	5	4	Bulgarian, Chinese, Spanish	(Bunge et al., 1997; Pajares et al., 2012; Wang et al., 2010)
c.464G>A	p.G155E	5	1	Dutch/Irish/Scottish	(Tomatsu et al., 2004c)
c.466T>C	p.F156L	5	2	Middle Eastern	(Morrone et al., 2014)
c.467T>C	p.F156S	5	2	French, unknown	(Bunge et al., 1997; Tomatsu et al., 2005)
c.467T>G	p.F156C	5	2	Australian, British/Irish	(Morrone et al., 2014; Yamada et al., 1998)
c.477G>T	p.W159C	5	10	Middle Eastern, Saudi Arabian	(Morrone et al., 2014; Qubbaj et al., 2008)
c.477G>A	p.W159Ter	5	2	American Caucasian	(Dung et al., 2013)
not given	p.G161E	5	1	Chinese	(Ye et al., 2013)
c.485C>T	p.S162F	5	7	Colombian	(Dung et al., 2013; Kato et al., 1997)
c.488C>A	p.P163H	5	2	Chinese	(Wang et al., 2010; Ye et al., 2013)
c.489delC	p.N164TfsTer34	5	2	Norwegian, unknown	(Bunge et al., 1997; Morrone et al., 2014)
c.491A>C	p.N164T	5	3	Brazilian	(Tomatsu et al., 2004a)
c.494G>A	p.C165Y	5	2	Brazilian	(Kubaski et al., 2013)
c.497A>G	p.H166R	5	2	British	(Morrone et al., 2014)
c.498delC	p.H166fsTer32	5	5	American, American Caucasian, unknown	(Dung et al., 2013; Tomatsu et al., 2005)
c.498C>G	p.H166Q	5	4	Australian, Irish/Danish, unknown	(Morrone et al., 2014; Tomatsu et al., 2004c; Tomatsu et al., 2005)
c.499T>G	p.F167V	5	5	Italian, unknown	(Montaño et al., 2007; Tomatsu et al., 2005)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.501insT	p.G168WfsTer3	5	1	German	(Bunge et al., 1997)
c.502_503delGGinsTT	p.G168L	5	2	Chinese	(Wang et al., 2010; Ye et al., 2013)
c.502G>A	p.G168R	5	1	Polish	(Bunge et al., 1997)
c.512A>C	p.D171A	5	4	unknown	(Sukegawa et al., 2000; Tomatsu et al., 2005)
c.530A>G	p.N177S	5	1	Chinese	(He et al., 2013)
c.532_533delAT	p.I178PfsTer13	5	1	Lebanese	(Bunge et al., 1997)
c.535C>T	p.P179S	5	2	Turkish	(Morrone et al., 2014; Terzioglu et al., 2002)
c.536C>A	p.P179H	5	2	unknown	(Bunge et al., 1997)
c.536C>T	p.P179L	5	2	unknown	(Yamada et al., 1998)
c.554A>G	p.E185G	5	1	British	(Tomatsu et al., 2005)
c.[566+?_1003-?dup; 1139+?_1140-?del]	complex rearrangement: duplication from intron 5 to intron 9, deletion in intron 10	Intron 5	1	Middle Eastern	(Morrone et al., 2014)
c.567-131ins42	c.567-131ins42	Intron 5	3	Mexican, unknown	(Tomatsu et al., 2005)
c.567-1G>T	c.567-1G>T	Intron 5	1	Chinese	(Wang et al., 2010)
c.600delG	p.E202KfsTer115	6	2	British, New Zealander	(Morrone et al., 2014; Tomatsu et al., 2004c)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.602G>A	p.G201E	6	6	Middle Eastern	(Morrone et al., 2014)
c.608C>T	p.A203V	6	3	Brazilian	(Dung et al., 2013; Tomatsu et al., 2005)
c.612C>G	p.N204K	6	13	American Caucasian, Japanese, unknown	(Dung et al., 2013; Fukuda et al., 1996; Ogawa et al., 1995; Tomatsu et al., 2004c; Tomatsu et al., 2005)
c.627C>G	p.Y209Ter	6	2	unknown	(Morrone et al., 2014)
c.631C>T	p.Q211Ter	6	1	Italian	(Tomatsu et al., 2004b)
c.634-1G>A	c.634-1G>A	Intron 6	1	Chinese	(Wang et al., 2010)
c.634-1G>T	p.E212VfsTer9	Intron 6	1	Greek	(Dung et al., 2013)
c.641T>C	p.L214P	7	2	Turkish	(Morrone et al., 2014)
c.647T>C	p.F216S	7	2	Middle Eastern	(Morrone et al., 2014)
c.688T>G	p.W230G	7	4	Asian-multiethnic, Pakistani	(Morrone et al., 2014)
c.689G>A	p.W230Ter	7	3	Finnish, unknown	(Montaño et al., 2003; Tomatsu et al., 2005)
c.697G>A	p.D233N	7	6	German/Polish, Middle Eastern, Polish	(Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 2004c)
c.704C>A	p.T235K	7	1	French Canadian	(Morrone et al., 2014)
c.706C>G	p.H236D	7	2	Chinese	(Wang et al., 2010)
c.708delC	p.H236RfsTer25	8	4	Turkish	(Terzioglu et al., 2002)
c.715G>T	p.V239F	7	4	Brazilian	(Dung et al., 2013; Tomatsu et al., 2005)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.719A>G	p.Y240C	7	2	Chinese, unknown	(Morrone et al., 2014; Ye et al., 2013)
c.725C>G	p.S242C	7	2	Korean	(Lee et al., 2012; Park et al., 2013)
c.740G>A	p.G247D	7	9	American Caucasian, American Caucasian: German/Swedish, Australian, British, German, Irish/German, Norwegian, unknown	(Bunge et al., 1997; Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 2005)
c.751C>T	p.R251Ter	7	1	British	(Morrone et al., 2014)
c.752G>A	p.R251Q	7	6	British, Irish, Korean, Chinese, unknown	(Lee et al., 2012; Morrone et al., 2014; Ye et al., 2013)
c.757C>T	p.R253W	7	18	Pakistani, unknown	(Morrone et al., 2014; Tomatsu et al., 2005)
c.758+1G>C	c.758+1G>C	Intron 7	1	Japanese	(Tomatsu et al., 2004c)
c.758+4A>T	c.758+4A>T	Intron 7	1	Greek	(Morrone et al., 2014)
c.758G>A	p.R253Q	8	1	American Caucasian	(Dung et al., 2013)
c.761A>G	p.Y254C	8	6	American Caucasian, Spanish	(Dung et al., 2013; Pajares et al., 2012)
c.769G>A	p.A257T	8	1	Polish	(Yamada et al., 1998)
c.776G>A	p.R259Q	8	11	Austrian, Hispanic, Polish, unknown	(Bunge et al., 1997; Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 2005; Tylki-Szymanska et al., 1998)
c.778G>A	p.E260K	8	1	Spanish	(Pajares et al., 2012)
c.780G>C	p.E260D	8	1	Danish	(Tomatsu et al., 2005)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.791G>C	p.S264T	8	2	British	(Morrone et al., 2014)
c.833_859del27	p.A277_N288del	8	1	unknown	(Tomatsu et al., 2005)
c.841_867del	p.T281_N289del	8	7	British, Dutch, French Canadian, Irish	(Bunge et al., 1997; Morrone et al., 2014; Tomatsu et al., 1995)
c.850_852delTTC	p.F284del	8	1	Italian	(Tomatsu et al., 2004b)
c.850T>G	p.F284V	8	5	British, Turkish	(Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 2005; Yamada et al., 1998)
c.853_855delTTC	p.F285del	8	2	American Caucasian, British/German/Portuguese, Korean	(Dung et al., 2013; Lee et al., 2012)
c.860C>T	p.S287L	8	20	Austrian, Greek, Irish/Italian/Polish, Macedonian, Middle Eastern, New Zealander, Polish, Spanish, Turkish, unknown	(Bunge et al., 1997; Dung et al., 2013; Morrone et al., 2014; Pajares et al., 2012; Tomatsu et al., 2004c; Tomatsu et al., 2005)
c.865A>G	p.N289D	8	1	Australian	(Morrone et al., 2014)
c.866A>G	p.N289S	8	2	Chinese	(Wang et al., 2010)
c.868G>C	p.G290R	8	1	Chinese	(He et al., 2013)
c.868G>A	p.G290S	8	4	British, Japanese, Chinese	(He et al., 2013; Tomatsu et al., 2005; Wang et al., 2010)
c.871G>A	p.A291T	8	20	Asian-multiethnic, British, Chinese, Finnish, Japanese, Pakistani, unknown	(Bunge et al., 1997; Montaño et al., 2003; Morrone et al., 2014; Tomatsu et al., 2004c; Tomatsu et al., 2005; Wang et al., 2010)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.872C>A	p.A291D	8	2	French, unknown	(Bunge et al., 1997; Tomatsu et al., 2005)
c.874delG	p.A292PfsTer25	8	2	Turkish	(Terzioglu et al., 2002)
c.884C>T	p.S295F	8	4	Austrian, Dutch, unknown	(Bunge et al., 1997; Tomatsu et al., 2004c; Tomatsu et al., 2005)
c.898+1G>A	c.898+1G>A	Intron 8	2	Lebanese	(Bunge et al., 1997)
c.898+1G>C	c.898+1G>C	Intron 8	4	Czech/German/Irish/Polish, unknown	(Tomatsu et al., 2004c; Tomatsu et al., 2005)
c.898+1G>A	p.G300DfsTer34	Intron 8	1	Ukrainian	(Dung et al., 2013)
c.899-1G>C	c.899-1G>C	Intron 8	2	British, unknown	(Carraresi et al., 2008; Morrone et al., 2014)
c.899-2A>G	c.899-2A>G	Intron 8	1	Mexican/Lebanese	(Tomatsu et al., 2004c)
c.901G>T	p.G301C	9	45	African-Varied, American Caucasian, Australian, Brazilian, Chinese, Colombian, French, French Canadian, Moroccan, Portuguese, Spanish	(Bunge et al., 1997; Dung et al., 2013; Kato et al., 1997; Morrone et al., 2014; Pajares et al., 2012; Tomatsu et al., 2004c; Ye et al., 2013)
c.917T>C	p.F306S	9	1	Chinese	(Ye et al., 2013)
c.920T>C	p.L307P	9	2	Brazilian	(Tomatsu et al., 2004a)
c.922T>C	p.C308R	9	1	American Caucasian	(Dung et al., 2013)
c.925G>A	p.G309R	9	2	British	(Morrone et al., 2014; Tomatsu et al., 2005)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.930G>C	p.K310N	9	2	American, unknown	(Tomatsu et al., 2005)
c.934A>G	p.T312A	9	1	Chinese	(Wang et al., 2010)
c.935C>G	p.T312S	9	22	British/Irish, Irish, unknown	(Morrone et al., 2014; Tomatsu et al., 2004c; Tomatsu et al., 2005; Yamada et al., 1998)
c.[937A>G; 977G>C]	p[T313A; W326S]	9	1	Australian	(Morrone et al., 2014)
c.938C>T	p.T313M	9	2	Canadian Caucasian, Irish/English	(Dung et al., 2013)
not given	p.E315K	9	1	Chinese	(Ye et al., 2013)
c.947G>T	p.G316V	9	1	Chinese	(Wang et al., 2010)
c.949G>A	p.G317R	9	1	(blank)	(Pollard et al., 2013)
c.951dupG	c.951dupG	9	1	(blank)	(Pollard et al., 2013)
c.953T>G	p.M318R	9	19	Chinese, Japanese, South-East Asian, Taiwanese, unknown	(Dung et al., 2013; He et al., 2013; Morrone et al., 2014; Ogawa et al., 1995; Wang et al., 2010; Yang et al., 2001)
c.971C>A	p.A324E	9	2	Chinese, unknown	(Wang et al., 2010; Ye et al., 2013)
c.974G>A	p.W325Ter	9	2	Chinese	(Wang et al., 2010)
p.W325_W326del	p.W325_W326del	9	1	Dutch	(Bunge et al., 1997)
c.975G>T	p.W325C	9	2	Canadian, Canadian Caucasian	(Dung et al., 2013; Tomatsu et al., 2005)
c.[977G>C; 937A>G]	p[W326S; T313A]	9	1	Australian	(Morrone et al., 2014)
c.1000C>T	p.Q334Ter	9	7	Korean	(Lee et al., 2012; Park et al., 2013)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.1003-2A>C	c.1003-2A>C	Intron 9	1	Chinese	(Ye et al., 2013)
c.1003-42C>T	c.1003-42C>T	Intron 9	2	Italian	(Tomatsu et al., 1997)
c.1012C>T	p.Q338Ter	10	2	Irish/German/Swedish, Norwegian	(Bunge et al., 1997; Tomatsu et al., 2004c)
c.1019G>A	p.G340D	10	10	Austrian, Chinese	(He et al., 2013; Tomatsu et al., 2004c; Wang et al., 2010)
c.1023C>G	p.S341R	10	9	Brazilian, British, Sri Lankan	(Dung et al., 2013; Tomatsu et al., 2004a; Tomatsu et al., 2005)
c.1028T>G	p.M343R	10	1	Japanese	(Ogawa et al., 1995)
c.1030G>A	p.D344N	10	1	Japanese	(Ogawa et al., 1995)
c.1032C>G	p.D344E	10	1	Vietnam	(Bunge et al., 1997)
c.1034T>C	p.L345P	10	1	German/Polish	(Tomatsu et al., 2004c)
c.1038C>A	p.F346L	10	1	Japanese	(Ogawa et al., 1995)
c.1046delG	c.1046delG	10	1	Chinese	(Ye et al., 2013)
c.1052C>T	p.A351V	10	1	Italian	(Tomatsu et al., 2005)
c.1055T>C	p.L352P	10	2	Irish/Italian/Polish, unknown	(Tomatsu et al., 2004c; Tomatsu et al., 2005)
c.1070C>T	p.P357L	10	1	Spanish	(Morrone et al., 2014)
c.[1070C>T; c.1156C>T]	p.[P357L; p.R386C]	10	2	Italian	(Tomatsu et al., 2004b)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.1070delC	p.P357RfsTer21	10	2	Middle Eastern	(Morrone et al., 2014)
c.1082G>T	p.R361G	10	3	British/German, German	(Tomatsu et al., 2004c; Bunge et al., 1997)
c.1097T>C	p.L366P	10	7	Chinese	(Guo et al., 2012; He et al., 2013; Wang et al., 2010; Ye et al., 2013)
c.1106T>C	p.L369P	10	4	Brazilian, Dutch/Irish/Scottish/French, Irish	(Morrone et al., 2014; Tomatsu et al., 2005)
c.1114delC	p.L372SfsTer6	10	2	Portuguese	(Morrone et al., 2014)
c.1120C>T	p.Q374Ter	10	1	Italian	(Tomatsu et al., 2004b)
c.1127G>A	p.R376Q	10	3	British/Irish, Irish	(Tomatsu et al., 2004c; Yamada et al., 1998)
c.1138A>G	p.R380G	10	2	Cape Verdean	(Morrone et al., 2014)
c.1139G>C	p.R380T	10	3	Asian-multiethnic, Brazilian-Asian	(Morrone et al., 2014; Tomatsu et al., 2005)
c.1140G>T	p.R380S	11	7	Argentine, Italian, unknown	(Montaño et al., 2007; Tomatsu et al., 2004a; Tomatsu et al., 2005)
c.1142delC	p.P381LfsTer9	11	3	Portuguese, Spanish	(Morrone et al., 2014; Pajares et al., 2012)
c.1155C>A	p.Y385Ter	11	2	British	(Morrone et al., 2014)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.1156C>T	p.R386C	11	55	American Caucasian, American Caucasian: Hispanic/Greek, American Caucasian: Italian, Argentine, Brazilian, British, Canadian Caucasian, Chilean, Chinese, Colombian, French, German, Italian, Japanese, Korean, Mexican, Polish, Portuguese, Spanish, Turkish, unknown	(Bunge et al., 1997; Dung et al., 2013; Fukuda et al., 1996; He et al., 2013; Lee et al., 2012; Morrone et al., 2014; Ogawa et al., 1995; Pajares et al., 2012; Park et al., 2013; Tomatsu et al., 2004a; Tomatsu et al., 1997; Tomatsu et al., 2004c; Tomatsu et al., 2004b; Wang et al., 2010)
c.1157G>A	p.R386H	11	3	British, Chilean, unknown	(Morrone et al., 2014; Tomatsu et al., 2004a)
c.1162G>A	p.D388N	11	2	Italian, unknown	(Tomatsu et al., 2004b; Tomatsu et al., 2005)
not given	p.L390P	11	1	Chinese	(Ye et al., 2013)
c.1168delC	p.L390Ter	11	4	Turkish	(Terzioglu et al., 2002)
c.1171A>G	p.M391V	11	22	American Caucasian: German, British, Canadian Caucasian, French, French Canadian, Irish/German/Swedish, unknown	(Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 1995; Tomatsu et al., 2004c; Tomatsu et al., 2005)
c.1175C>T	p.A392V	11	10	Asian-multiethnic, Chinese, Italian, unknown	(He et al., 2013; Morrone et al., 2014; Tomatsu et al., 2004b; Tomatsu et al., 2005)
c.1176_1178del3	p.A392del	11	3	Chinese, Middle Eastern	(Morrone et al., 2014; Ye et al., 2013)
c.1177_1178insT	p.A393CfsTer23	11	2	Turkish	(Terzioglu et al., 2002)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.1180A>C	p.T394P	11	1	Spanish	(Pajares et al., 2012)
c.1183C>G	p.L395V	11	1	Irish	(Yamada et al., 1998)
c.1184T>C	p.L395P	11	1	Caucasian	(Tomatsu et al., 2005)
c.1188delG	c.1188delG	11	1	Chinese	(Ye et al., 2013)
c.1192C>G	p.H398D	11	6	Pakistani, unknown	(Tomatsu et al., 2004c; Tomatsu et al., 2005)
c.1195delA	p.K399fsTer41	11	2	Iraqi	(Dung et al., 2013)
c.1201C>T	p.H401Y	11	1	Italian	(Tomatsu et al., 2005)
c.1209_1210delGAinsTT	p.W403_T404delinsCS	11	2	Chinese	(He et al., 2013; Ye et al., 2013)
c.1214G>A	p.W405Ter	11	1	Chinese	(Ye et al., 2013)
c.1215G>A	p.W405Ter	11	1	unknown	(Dung et al., 2013)
c.1219A>C	p.N407H	11	9	American black, American Caucasian, British, Dutch, French, German, Portuguese, unknown	(Bunge et al., 1997; Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 2005)
c.1226G>C	p.W409S	11	2	unknown	(Tomatsu et al., 2005)
c.1240C>T	p.Q414Ter	11	1	New Zealander	(Morrone et al., 2014)
c.1243-1G>A	c.1243-1G>A	Intron 11	3	French, Korean	(Bunge et al., 1997; Lee et al., 2012; Park et al., 2013)
c.1243-1G>C	c.1243-1G>C	Intron 11	1	Chinese	(He et al., 2013)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.1244G>T	p.G415V	12	2	Portuguese	(Morrone et al., 2014)
c.1247T>C	p.I416T	12	2	Asian-multiethnic	(Morrone et al., 2014)
c.1259C>G	p.P420R	12	2	Asian-multiethnic, Spanish, Portuguese	(Morrone et al., 2014)
not given	p.P420S	12	1	Chinese	(Ye et al., 2013)
c.1262G>A	p.G421E	12	1	unknown	(Lissens and Liebaers, 1999)
c.1264C>A	p.Q422K	12	1	Chinese	(Wang et al., 2010)
c.1264C>T	p.Q422Ter	12	2	Chinese	(Wang et al., 2010)
c.1275delA	p.V427SfsTer13	12	1	British	(Morrone et al., 2014)
c.1290_1291delCA	p.H430QfsTer71	12	5	Japanese	(Fukuda et al., 1996; Ogawa et al., 1995)
not given	p.L400RfsTer54	12	1	Chinese	(He et al., 2013)
c.1319delT	p.L440RfsTer54	12	4	British, Finnish, Irish, unknown	(Montaño et al., 2003; Morrone et al., 2014; Tomatsu et al., 1995; Tomatsu et al., 2005)
c.1349A>T	p.E450V	12	2	Japanese	(Ogawa et al., 1995; Tomatsu et al., 2005)
c.1354T>A	p.F452I	12	4	American, Canadian Caucasian	(Dung et al., 2013; Tomatsu et al., 2005)
c.1354T>C	p.F452L	12	3	Chinese	(Wang et al., 2010; Ye et al., 2013)
c.1355_1356insT	p.F452fsTer42	12	1	Argentine	(Tomatsu et al., 2004a)
not given	p.L454P	12	1	Chinese	(Ye et al., 2013)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.1364+1G>A	c.1364+1G>A	Intron 12	1	German	(Bunge et al., 1997)
c.1408T>C	p.S470P	13	1	American	(Tomatsu et al., 2005)
c.1417C>T	p.Q473Ter	13	3	Turkish, unknown	(Terzioglu et al., 2002; Tomatsu et al., 1995)
c.1429_1455del	p.E477_Q485del	13	2	Middle Eastern	(Morrone et al., 2014)
c.1450C>T	p.P484S	13	3	Japanese, unknown	(Montaño et al., 2007; Tomatsu et al., 2005)
c.1460A>G	p.N487S	13	1	unknown	(Tomatsu et al., 1995)
c.1474G>A	p.A492T	13	2	Middle Eastern	(Morrone et al., 2014)
c.1480A>G	p.M494V	13	5	Canadian Caucasian, Turkish	(Bunge et al., 1997; Dung et al., 2013; Morrone et al., 2014)
c.1483A>T	p.N495Y	14	1	Spanish	(Pajares et al., 2012)
c.1485C>G	p.N495K	14	4	American Caucasian, Canadian Caucasian, Middle Eastern	(Dung et al., 2013; Morrone et al., 2014)
c.1498G>A	p.G500S	14	2	Moroccan	(Morrone et al., 2014)
c.1520G>T	p.C507F	14	1	Hispanic	(Morrone et al., 2014)
c.1559G>A	p.W520Ter	14	1	British	(Morrone et al., 2014)
c.1560G>A	p.W520Ter	14	1	Chinese	(He et al., 2013)
c.1567T>G	p.Ter523EextTer93	14	3	Chinese	(Dung et al., 2013; He et al., 2013; Zhao et al., 2011)

Nucleotide alteration	Predicted effect on protein	Exon	Alleles reported	Reported country/ethnicity	Reference
c.1568A>G	p.Ter523Wext	14	2	Korean	(Lee et al., 2012)
double deletion	double deletion		5	Japanese	(Ogawa et al., 1995)
large deletion	large deletion		2	Japanese	(Fukuda et al., 1996; Ogawa et al., 1995)

Supp. Table S4. Patients with Morquio A assessed as homozygous for a *GALNS* mutation with an assessed growth phenotype

Both base changes	Both predicted effects on protein	Both allele types	Growth phenotype	Author	Year
c.121A>T	p.M41L	Missense	Less severe	Tomatsu	2005
c.121A>T	p.M41L	Missense	Less severe	Dung	2013
c.121A>T	p.M41L	Missense	Less severe	Tomatsu	2004c
c.244T>C	p.S82P	Missense	Less severe	Dung	2013
c.332A>G	p.Q111R	Missense	Less severe	Tomatsu	2005
c.512A>C	p.D171A	Missense	Less severe	Tomatsu	2005
not given	p.D171A	Missense	Less severe	Sukegawa	2000
c.612C>G	p.N204K	Missense	Less severe	Tomatsu	2005
c.612C>G	p.N204K	Missense	Less severe	Ogawa	1995
c.612C>G	p.N204K	Missense	Less severe	Ogawa	1995
c.706C>G	p.H236D	Missense	Less severe	Wang	2010
c.757C>T	p.R253W	Missense	Less severe	Tomatsu	2005
c.776G>A	p.R259Q	Missense	Less severe	Tomatsu	2005
c.776G>A	p.R259Q	Missense	Less severe	Bunge	1997
c.850T>G	p.F284V	Missense	Less severe	Dung	2013
c.866A>G	p.N289S	Missense	Less severe	Wang	2010
c.898+1G>C	c.898+1G>C	Intronic	Less severe	Tomatsu	2005
c.935C>G	p.T312S	Missense	Less severe	Tomatsu	2005
c.935C>G	p.T312S	Missense	Less severe	Yamada	1998
c.1171A>G	p.M391V	Missense	Less severe	Tomatsu	2005
c.1171A>G	p.M391V	Missense	Less severe	Tomatsu	2004c
c.1171A>G	p.M391V	Missense	Less severe	Tomatsu	2004c
c.1192C>G	p.H398D	Missense	Less severe	Tomatsu	2005
c.1192C>G	p.H398D	Missense	Less severe	Tomatsu	2004c
c.1192C>G	p.H398D	Missense	Less severe	Tomatsu	2004c
c.1226G>C	p.W409S	Missense	Less severe	Tomatsu	2005
c.[199C>A, c.1226G>C]	p.[L67M, p.W409S]	Missense	Intermediate	Bunge	1997
c.332A>G	p.Q111R	Missense	Intermediate	Bunge	1997
c.363G>C	p.E121D	Missense	Intermediate	Pajares	2012
c.901G>T	p.G301C	Missense	Intermediate	Pajares	2012
c.1156C>T	p.R386C	Missense	Intermediate	Morrone	2014
c.1219A>C	p.N407H	Missense	Intermediate	Bunge	1997
c.1A>G	p.M1V	Missense	Most severe	Tomatsu	2004b
c.1A>G	p.M1V	Missense	Most severe	Tomatsu	2004b
c.1A>G	p.M1V	Missense	Most severe	Tomatsu	2004b
c.1A>G	p.M1V	Missense	Most severe	Tomatsu	2004b
c.29G>A	p.W10Ter	Nonsense	Most severe	Tomatsu	2004b
c.121-1G>A	c.121-1G>A	Intronic	Most severe	Morrone	2014

Both base changes	Both predicted effects on protein	Both allele types	Growth phenotype	Author	Year
c.121-1G>A	c.121-1G>A	Intronic	Most severe	Morrone	2014
c.121-2A>G	c.121-2A>G	Intronic	Most severe	Ogawa	1995
c.121-2A>G	c.121-2A>G	Intronic	Most severe	Ogawa	1995
c.121A>T	p.M41L	Missense	Most severe	Tomatsu	2004c
c.122T>A	p.M41K	Missense	Most severe	Dung	2013
c.139G>A	p.G47R	Missense	Most severe	Bunge	1997
c.139G>A	p.G47R	Missense	Most severe	Tomatsu	2004c
c.139G>A	p.G47R	Missense	Most severe	Dung	2013
c.224C>G	p.A75G	Missense	Most severe	Tomatsu	2004a
c.224C>G	p.A75G	Missense	Most severe	Tomatsu	2004a
c.230C>G	p.P77R	Missense	Most severe	Tomatsu	1995
c.230C>G	p.P77R	Missense	Most severe	Tomatsu	2004b
c.230C>G	p.P77R	Missense	Most severe	Dung	2013
c.251C>A	p.A84E	Missense	Most severe	Morrone	2014
c.268C>T	p.R90W	Missense	Most severe	Tomatsu	1995
c.280C>T	p.R94C	Missense	Most severe	Morrone	2014
c.280C>T	p.R94C	Missense	Most severe	Morrone	2014
c.281G>T	p.R94L	Missense	Most severe	Morrone	2014
c.287G>T	p.G96V	Missense	Most severe	Tomatsu	1995
c.319G>A	p.A107T	Missense	Most severe	Tomatsu	2004b
c.346G>A	p.G116S	Missense	Most severe	Morrone	2014
c.346G>A	p.G116S	Missense	Most severe	Tomatsu	2004a
c.346G>A	p.G116S	Missense	Most severe	Tomatsu	2004a
c.374C>T	p.P125L	Missense	Most severe	Tomatsu	1997
c.374C>T	p.P125L	Missense	Most severe	Tomatsu	2004b
c.421T>A	p.W141R	Missense	Most severe	Bunge	1997
c.423-1G>A	c.423-1G>A	Intronic	Most severe	Yamada	1998
c.452C>T	p.P151L	Missense	Most severe	Tomatsu	1995
c.452C>T	p.P151L	Missense	Most severe	Tomatsu	1995
c.452C>T	p.P151L	Missense	Most severe	Dung	2013
c.463G>A	p.G155R	Missense	Most severe	Bunge	1997
c.466T>C	p.F156L	Missense	Most severe	Morrone	2014
c.477G>A	p.W159Ter	Nonsense	Most severe	Dung	2013
c.485C>T	p.S162F	Missense	Most severe	Kato	1997
c.485C>T	p.S162F	Missense	Most severe	Kato	1997
c.491A>C	p.N164T	Missense	Most severe	Tomatsu	2004a
c.536C>A	p.P179H	Missense	Most severe	Bunge	1997
c.536C>T	p.P179L	Missense	Most severe	Yamada	1998
c.688T>G	p.W230G	Missense	Most severe	Tomatsu	1995
c.715G>T	p.V239F	Missense	Most severe	Dung	2013
c.761A>G	p.Y254C	Missense	Most severe	Pajares	2012
c.761A>G	p.Y254C	Missense	Most severe	Pajares	2012
c.860C>T	p.S287L	Missense	Most severe	Bunge	1997

Both base changes	Both predicted effects on protein	Both allele types	Growth phenotype	Author	Year
c.860C>T	p.S287L	Missense	Most severe	Dung	2013
c.860C>T	p.S287L	Missense	Most severe	Pajares	2012
c.871G>A	p.A291T	Missense	Most severe	Tomatsu	1995
c.898+1G>C	c.898+1G>C	Intronic	Most severe	Tomatsu	2004c
c.901G>T	p.G301C	Missense	Most severe	Morrone	2014
c.901G>T	p.G301C	Missense	Most severe	Morrone	2014
c.901G>T	p.G301C	Missense	Most severe	Morrone	2014
c.901G>T	p.G301C	Missense	Most severe	Bunge	1997
c.901G>T	p.G301C	Missense	Most severe	Bunge	1997
c.901G>T	p.G301C	Missense	Most severe	Kato	1997
c.901G>T	p.G301C	Missense	Most severe	Kato	1997
c.901G>T	p.G301C	Missense	Most severe	Kato	1997
c.901G>T	p.G301C	Missense	Most severe	Kato	1997
c.901G>T	p.G301C	Missense	Most severe	Kato	1997
c.901G>T	p.G301C	Missense	Most severe	Kato	1997
c.901G>T	p.G301C	Missense	Most severe	Kato	1997
c.901G>T	p.G301C	Missense	Most severe	Tomatsu	2004c
c.901G>T	p.G301C	Missense	Most severe	Dung	2013
c.901G>T	p.G301C	Missense	Most severe	Dung	2013
c.938C>T	p.T313M	Missense	Most severe	Dung	2013
c.974G>A	p.W325Ter	Nonsense	Most severe	Wang	2010
c.1003-42C>T	c.1003-42C>T	Intronic	Most severe	Tomatsu	1997
c.1019G>A	p.G340D	Missense	Most severe	Wang	2010
c.1019G>A	p.G340D	Missense	Most severe	Wang	2010
c.1019G>A	p.G340D	Missense	Most severe	Wang	2010
c.1023C>G	p.S341R	Missense	Most severe	Dung	2013
c.[1070C>T; c.1156C>T]	p.[P357L; p.R386C]	Missense	Most severe	Tomatsu	2004b
c.1070delC	p.P357RfsTer21	Insertion and/or deletion	Most severe	Morrone	2014
c.1097T>C	p.L366P	Missense	Most severe	He	2013
c.1097T>C	p.L366P	Missense	Most severe	He	2013
c.1114delC	p.L372SfsTer6	Insertion and/or deletion	Most severe	Morrone	2014
c.1127G>A	p.R376Q	Missense	Most severe	Yamada	1998
c.1138A>G	p.R380G	Missense	Most severe	Morrone	2014
c.1142delC	p.P381LfsTer9	Insertion and/or deletion	Most severe	Morrone	2014
c.1156C>T	p.R386C	Missense	Most severe	Pajares	2012
c.1156C>T	p.R386C	Missense	Most severe	Bunge	1997
c.1156C>T	p.R386C	Missense	Most severe	Tomatsu	1997
c.1156C>T	p.R386C	Missense	Most severe	Tomatsu	2004b
c.1156C>T	p.R386C	Missense	Most severe	Tomatsu	2004b
c.1156C>T	p.R386C	Missense	Most severe	Tomatsu	2004a
c.1156C>T	p.R386C	Missense	Most severe	Tomatsu	2004a

Both base changes	Both predicted effects on protein	Both allele types	Growth phenotype	Author	Year
c.1156C>T	p.R386C	Missense	Most severe	Tomatsu	2004a
c.1156C>T	p.R386C	Missense	Most severe	Tomatsu	2004a
c.1156C>T	p.R386C	Missense	Most severe	Dung	2013
c.1168delC	p.L390Ter	Insertion and/or deletion	Most severe	Bunge	1997
c.1171A>G	p.M391V	Missense	Most severe	Dung	2013
c.1175C>T	p.A392V	Missense	Most severe	He	2013
c.1176_1178del3	p.A392del	Insertion and/or deletion	Most severe	Morrone	2014
c.1264C>T	p.Q422Ter	Nonsense	Most severe	Wang	2010
c.1290_1291delCA	p.H430QfsTer71	Insertion and/or deletion	Most severe	Ogawa	1995
c.1429_1455del	p.E477_Q485del	Insertion and/or deletion	Most severe	Morrone	2014
c.1474G>A	p.A492T	Missense	Most severe	Morrone	2014
c.1480A>G	p.M494V	Missense	Most severe	Bunge	1997
c.1485C>G	p.N495K	Missense	Most severe	Morrone	2014
c.1568A>G	p.Ter523Wext	Missense	Most severe	Lee	2012
double deletion	double deletion	Insertion and/or deletion	Most severe	Ogawa	1995

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Supp. Table S5. Missense alleles reported from patients with Morquio A with either a MutPred or PolyPhen score below 0.75

Nucleotide alteration	Predicted effect on protein	MutPred score	PolyPhen-2 score	Alleles reported	Reported country/ethnicity	Estimated likelihood of familial relationship between patients	Reference
c.1A>G	p.M1V	0.89	**0.38**	10	Italian, Japanese, unknown	Low	(Montaño et al., 2007; Tomatsu et al., 2004b; Tomatsu et al., 2005)
c.3G>A	p.M1I	0.859	**0.378**	2	Chinese, Finnish	Low	(Dung et al., 2013; Ye et al., 2013)
c.43C>A	p.L15M	0.752	**0.193**	2	Scottish, unknown	Unknown	(Tomatsu et al., 2005)
c.67G>C	p.G23R	**0.719**	**0.089**	2	Italian, unknown	Unknown	(Tomatsu et al., 2005; Tomatsu et al., 2004b)
c.405C>G	p.S135R	0.94	**0.529**	1	unknown	N/A	(Yamada et al., 1998)
c.448C>T	p.H150Y	**0.727**	0.915	2	Italian, unknown	Unknown	(Tomatsu et al., 2004b; Tomatsu et al., 2005)
c.488C>A	p.P163H	**0.601**	1	2	Chinese	Possible	(Wang et al., 2010; Ye et al., 2013)
c.494G>A	p.C165Y	**0.6**	1	2	Brazilian	N/A, 1 homozygous patient	(Kubaski et al., 2013)
c.499T>G	p.R167V	0.775	**0.529**	5	Italian, unknown	Possible	(Montaño et al., 2007; Tomatsu et al., 2005)
c.554A>G	p.E185G	0.791	**0.529**	1	British	N/A	(Tomatsu et al., 2005)
c.608C>T	p.A203V	0.763	**0.243**	3	Brazilian	Possible	(Dung et al., 2013; Tomatsu et al., 2005)
c.612C>G	p.N204K	0.846	**0.285**	13	American Caucasian, Japanese, unknown	Low	(Dung et al., 2013; Fukuda et al., 1996; Ogawa et al., 1995; Tomatsu et al., 2004c; Tomatsu et al., 2005)
c.719A>G	p.Y240C	**0.738**	0.999	2	Chinese, unknown	Unknown	(Morrone et al., 2014; Ye et al., 2013)
c.725C>G	p.S242C	**0.64**	1	2	Korean	Possible	(Lee et al., 2012; Park et al., 2013)
c.740G>A	p.G247D	0.8	**0.367**	9	American Caucasian, American Caucasian: German/Swedish, Australian, British, German, Irish/German, Norwegian, unknown	Low	(Bunge et al., 1997; Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 2005)

Nucleotide alteration	Predicted effect on protein	MutPred score	PolyPhen-2 score	Alleles reported	Reported country/ethnicity	Estimated likelihood of familial relationship between patients	Reference
c.752G>A	p.R251Q	**0.69**	0.99	6	British, Irish, Korean, Chinese, unknown	Low	(Lee et al., 2012; Morrone et al., 2014; Ye et al., 2013)
c.776G>A	p.R259Q	0.765	**0.73**	11	Austrian, Hispanic, Polish, unknown	Low	(Bunge et al., 1997; Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 2005; Tylki-Szymanska et al., 1998)
c.850T>G	p.R284V	0.772	**0.164**	5	British, Turkish	Low	(Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 2005; Yamada et al., 1998)
c.934A>G	p.T312A	**0.745**	0.969	1	Chinese	N/A	(Wang et al., 2010)
c.935C>G	p.T312S	0.83	**0.444**	22	British/Irish, Irish, unknown	Low	(Morrone et al., 2014; Tomatsu et al., 2004c; Tomatsu et al., 2005; Yamada et al., 1998)
c.1023C>G	p.S341R	0.929	**0.724**	9	Brazilian, British, Sri Lankan	Low	(Dung et al., 2013; Tomatsu et al., 2004a; Tomatsu et al., 2005)
c.1052C>T	p.A351V	0.854	**0.027**	1	Italian	N/A	(Tomatsu et al., 2005)
c.1070C>T	p.P357L	0.768	**0.027**	1	Spanish	N/A	(Morrone et al., 2014)
c.1097T>C	p.L366P	**0.706**	0.95	7	Chinese	No familial relationship	(Guo et al., 2012; He et al., 2013; Wang et al., 2010; Ye et al., 2013)
c.1127G>A	p.R376Q	**0.725**	**0.003**	3	British/Irish, Irish	Possible	(Tomatsu et al., 2004c; Yamada et al., 1998)
c.1162G>A	p.D388N	0.881	**0.004**	2	Italian, unknown	Unknown	(Tomatsu et al., 2004b; Tomatsu et al., 2005)
c.1171A>G	p.M391V	0.857	**0.17**	22	American Caucasian: German, British, Canadian Caucasian, French, French Canadian, Irish/German/Swedish, unknown	Low	(Dung et al., 2013; Morrone et al., 2014; Tomatsu et al., 1995; Tomatsu et al., 2004c; Tomatsu et al., 2005)
c.1183C>G	p.L395V	0.815	**0.01**	1	Irish	N/A	(Yamada et al., 1998)
c.1184T>C	p.L395P	0.873	**0.076**	1	Caucasian	N/A	(Tomatsu et al., 2005)
c.1192C>G	p.H398D	0.905	**0.006**	6	Pakistani, unknown	Possible	(Tomatsu et al., 2004c; Tomatsu et al., 2005)
c.1226G>C	p.W409S	0.886	**0.259**	2	unknown	-	(Tomatsu et al., 2005)
c.1258C>T	p.P420S	**0.515**	1	1	Chinese	N/A	(Ye et al., 2013)
c.1264C>A	p.Q422K	**0.56**	0.955	1	Chinese	N/A	(Wang et al., 2010)

Nucleotide alteration	Predicted effect on protein	MutPred score	PolyPhen-2 score	Alleles reported	Reported country/ethnicity	Estimated likelihood of familial relationship between patients	Reference
c.1354T>C	p.R452L	**0.573**	**0.02**	3	Chinese	No familial relationship	(Wang et al., 2010; Ye et al., 2013)
c.1354T>A	p.R452I	0.754	**0.345**	4	American, Canadian Caucasian	Low	(Dung et al., 2013; Tomatsu et al., 2005)
c.1450C>T	p.P484S	0.742	**0.728**	3	Japanese, unknown	-	(Montaño et al., 2007; Tomatsu et al., 2005)
c.1460A>G	p.N487S	**0.648**	**0.345**	1	unknown	N/A	(Tomatsu et al., 1995)
c.1480A>G	p.M494V	**0.737**	**0.118**	5	Canadian Caucasian, Turkish	Low	(Bunge et al., 1997; Dung et al., 2013; Morrone et al., 2014)
c.1483A>T	p.N495Y	**0.66**	0.99	1	Spanish	N/A	(Pajares et al., 2012)

MutPred (Li et al., 2009) or PolyPhen-2 (Adzhubei et al., 2010) score(s) below 0.75 are indicated in bold. The likelihood of a familial relationship was estimated based on the reporting manuscripts and reported ethnicities but in most cases cannot conclusively be determined from the available information.

Supp. Table S6. Variants reported in dbSNP and/or Exome Variant Server for which phenotypic information is unavailable

cDNA mutation	Predicted effect on protein	First Referenced/Reported
c.79G>A	A27T	dbSNP
c.143T>C	V48A	dbSNP
c.170C>T	P57L	dbSNP, Exome Variant Server
c.229C>G	P77A	dbSNP
c.238T>A	S80T	Exome Variant Server
c.304G>A	A102T	Exome Variant Server
c.325A>G	T109A	dbSNP
c.359C>T	S120L	dbSNP, Exome Variant Server
c.364C>T	Q122Ter	Exome Variant Server
c.373C>T	P125S	Exome Variant Server
c.412G>A	V138I	dbSNP, Exome Variant Server
c.487C>A	P163T	dbSNP
c.515A>G	N172S	dbSNP, Exome Variant Server
c.516C>G	N172L	Exome Variant Server
c.550T>C	W184R	dbSNP
c.559G>C	V187L	dbSNP
c.664C>T	R222W	dbSNP, Exome Variant Server
c.665G>A	R222Q	dbSNP, Exome Variant Server
c.685T>C	Y229H	dbSNP, Exome Variant Server
c.700G>A	A234T	Exome Variant Server
c.715G>A	V239I	dbSNP, Exome Variant Server
c.734T>C	F245S	dbSNP, Exome Variant Server
c.794T>C	I265T	dbSNP, Exome Variant Server
c.832G>A	A278T	Exome Variant Server
c.833C>T	A278V	dbSNP, Exome Variant Server
c.857C>T	T286M	dbSNP, Exome Variant Server
c.887C>T	A296V	dbSNP
c.956G>A	R319K	Exome Variant Server
c.994G>A	A332T	Exome Variant Server
c.1001A>G	Q334R	dbSNP, Exome Variant Server
c.1078G>A	D360N	dbSNP, Exome Variant Server
c.1088T>C	I363T	dbSNP
c.1100A>G	N367S	dbSNP, Exome Variant Server

cDNA mutation	Predicted effect on protein	First Referenced/Reported
c.1105C>T	L369F	Exome Variant Server
c.1134G>A	M378I	dbSNP, Exome Variant Server
c.1135G>A	D379N	dbSNP, Exome Variant Server
c.1144A>G	I382V	dbSNP, Exome Variant Server
c.1186G>A	G396R	Exome Variant Server
c.1198G>A	A400T	dbSNP
c.1217C>T	T406I	dbSNP, Exome Variant Server
c.1360C>T	L454F	dbSNP
c.1414G>A	V472I	dbSNP, Exome Variant Server
c.1438G>T	V480F	dbSNP, Exome Variant Server
c.1475C>T	A492V	dbSNP, Exome Variant Server
c.1526C>G	T509R	dbSNP
c.1540A>G	I514V	dbSNP

Supp. References

Adzhubei IA, Schmidt S, Peshkin L, Ramensky VE, Gerasimova A, Bork P, Kondrashov AS, Sunyaev SR. 2010. A method and server for predicting damaging missense mutations. Nat Methods 7:248-249.

Bunge S, Kleijer WJ, Tylki-Szymanska A, Steglich C, Beck M, Tomatsu S, Fukuda S, Poorthuis BJ, Czartoryska B, Orii T, Gal A. 1997. Identification of 31 novel mutations in the N-acetylgalactosamine-6-sulfatase gene reveals excessive allelic heterogeneity among patients with Morquio A syndrome. Hum Mutat 10:223-232.

Carraresi L, Parini R, Filoni C, Caciotti A, Sersale G, Tomatsu S, Orlando C, Zammarchi E, Guerrini R, Donati MA, Morrone A. 2008. GALNS gene expression profiling in Morquio A patients' fibroblasts. Clin Chim Acta 397:72-76.

Catarzi S, Giunti L, Papadia F, Gabrielli O, Guerrini R, Donati MA, Genuardi M, Morrone A. 2012. Morquio A syndrome due to maternal uniparental isodisomy of the telomeric end of chromosome 16. Mol Genet Metab 105:438-442.

Cole DE, Fukuda S, Gordon BA, Rip JW, LeCouteur AN, Rupar CA, Tomatsu S, Ogawa T, Sukegawa K, Orii T. 1996. Heteroallelic missense mutations of the galactosamine-6-sulfate sulfatase (GALNS) gene in a mild form of Morquio disease (MPS IVA). Am J Med Genet 63:558-565.

Dung VC, Tomatsu S, Montaño AM, Gottesman G, Bober MB, Mackenzie W, Maeda M, Mitchell GA, Suzuki Y, Orii T. 2013. Mucopolysaccharidosis IVA: Correlation between genotype, phenotype and keratan sulfate levels. Mol Genet Metab 110:129-138.

Fukuda S, Tomatsu S, Masuno M, Ogawa T, Yamagishi A, Rezvi GM, Sukegawa K, Shimozawa N, Suzuki Y, Kondo N, Imaizumi K, Kuroki Y, Okabe T, Orii T. 1996. Mucopolysaccharidosis IVA: submicroscopic deletion of 16q24.3 and a novel R386C mutation of N-acetylgalactosamine-6-sulfate sulfatase gene in a classical Morquio disease. Hum Mutat 7:123-134.

Guo YB, Ai Y, Zhao Y, Tang J, Jiang WY, Du ML, Ma HM, Zhong YF. 2012. Rapid prenatal genetic diagnosis of a fetus with a high risk for Morquio A syndrome. Zhonghua Yi Xue Yi Chuan Xue Za Zhi 29:126-130.

He D, Huang Y, Ou Z, Sheng H, Li S, Zhao X, Li R, Zheng J, Liu L. 2013. Molecular genetic assay of Mucopolysaccharidosis IVA in South China. Gene 532:46-52.

Kato Z, Fukuda S, Tomatsu S, Vega H, Yasunaga T, Yamagishi A, Yamada N, Valencia A, Barrera LA, Sukegawa K, Orii T, Kondo N. 1997. A novel common missense mutation G301C in the N-acetylgalactosamine-6-sulfate sulfatase gene in Mucopolysaccharidosis IVA. Hum Genet 101:97-101.

Khedhiri S, Chkioua L, Bouzidi H, Dandana A, Ferchichi S, Ben Turkia H, Miled A, Laradi S. 2012. Mucopolysaccharidosis IVA within Tunisian patients: Confirmation of the two novel GALNS gene mutations. Pathol Biol (Paris) 60:190-192.

Khedhiri S, Chkioua L, Ferchichi S, Miled A, Laradi S. 2011. Morquio A disease: clinical and molecular study of Tunisian patients. Ann Biol Clin (Paris) 69:425-429.

Kubaski F, Brusius-Facchin AC, Palhares HM, Balarin MA, Viapiana-Camelier M, Guidobono R, Burin MG, Giugliani R, Leistner-Segal S. 2013. Identification of a novel missense mutation in Brazilian patient with a severe form of Mucopolysaccharidosis type IVA. Gene 517:112-115.

Laradi S, Tukel T, Khediri S, Shabbeer J, Erazo M, Chkioua L, Chaabouni M, Ferchichi S, Miled A, Desnick RJ. 2006. Mucopolysaccharidosis type IV: N-acetylgalactosamine-6-sulfatase mutations in Tunisian patients. Mol Genet Metab 87:213-218.

Lee NH, Cho SY, Maeng SH, Jeon TY, Sohn YB, Kim SJ, Park HD, Jin DK. 2012. Clinical, radiologic, and genetic features of Korean patients with Mucopolysaccharidosis IVA. Korean J Pediatr 55:430-437.

Li B, Krishnan VG, Mort ME, Xin F, Kamati KK, Cooper DN, Mooney SD, Radivojac P. 2009. Automated inference of molecular mechanisms of disease from amino acid substitutions. Bioinformatics 25:2744-2750.

Lissens W, Liebaers I. 1999. Human Genetics 105:189.

Montaño AM, Kaitila I, Sukegawa K, Tomatsu S, Kato Z, Nakamura H, Fukuda S, Orii T, Kondo N. 2003. Mucopolysaccharidosis IVA: characterization of a common mutation found in Finnish patients with attenuated phenotype. Hum Genet 113:162-169.

Montaño AM, Sukegawa K, Kato Z, Carrozzo R, Di Natale P, Christensen E, Orii KO, Orii T, Kondo N, Tomatsu S. 2007. Effect of 'attenuated' mutations in mucopolysaccharidosis IVA on molecular phenotypes of N-acetylgalactosamine-6-sulfate sulfatase. J Inherit Metab Dis 30:758-767.

Morrone A, Tylee KL, Al-Sayed M, Brusius-Facchin AC, Caciotti A, Church HJ, Coll MJ, Davidson D, Fietz MJ, Gort L, Hegde M, Kubaski F, Lacerda L, Laranjeira F, Leistner-Segal S, Mooney S, Pajares S, Pollard L, Ribeiro I, Wang RY, Miller N. 2014. Molecular Testing of 163 Patients with Morquio A (Mucopolysaccharidosis IVA) Identifies 39 Novel *GALNS* Mutations. Mol Genet Metab Advance online publication. pii: S1096-7192(14)00092-4. doi: 10.1016/j.ymgme.2014.03.004.:.

Ogawa T, Tomatsu S, Fukuda S, Yamagishi A, Rezvi GM, Sukegawa K, Kondo N, Suzuki Y, Shimozawa N, Oru T. 1995. Mucopolysaccharidosis IVA: screening and identification of mutations of the N-acetylgalactosamine-6-sulfate sulfatase gene. Hum Mol Genet 4:341-349.

Pajares S, Alcalde C, Couce ML, Del Toro M, Gonzalez-Meneses A, Guillen E, Pineda M, Pintos G, Gort L, Coll MJ. 2012. Molecular analysis of Mucopolysaccharidosis IVA (Morquio A) in Spain. Mol Genet Metab 106:196-201.

Park HD, Ko AR, Ki CS, Lee SY, Kim JW, Cho SY, Kim SH, Park SW, Sohn YB, Jin DK. 2013. Five novel mutations of GALNS in Korean patients with mucopolysaccharidosis IVA. Am J Med Genet A 161A:509-517.

Pollard LM, Jones JR, Wood TC. 2013. Molecular characterization of 355 mucopolysaccharidosis patients reveals 104 novel mutations. J Inherit Metab Dis 36:179-187.

Qubbaj W, Al-Aqeel AI, Al-Hassnan Z, Al-Duraihim A, Awartani K, Al-Rejjal R, Coskun S. 2008. Preimplantation genetic diagnosis of Morquio disease. Prenat Diagn 28:900-903.

Sukegawa K, Nakamura H, Kato Z, Tomatsu S, Montaño AM, Fukao T, Toietta G, Tortora P, Orii T, Kondo N. 2000. Biochemical and structural analysis of missense mutations in N-acetylgalactosamine-6-sulfate sulfatase causing mucopolysaccharidosis IVA phenotypes. Hum Mol Genet 9:1283-1290.

Terzioglu M, Tokatli A, Coskun T, Emre S. 2002. Molecular analysis of Turkish mucopolysaccharidosis IVA (Morquio A) patients: identification of novel mutations in the N-acetylgalactosamine-6-sulfate sulfatase (GALNS) gene. Hum Mutat 20:477-478.

Tomatsu S, Dieter T, Schwartz IV, Sarmient P, Giugliani R, Barrera LA, Guelbert N, Kremer R, Repetto GM, Gutierrez MA, Nishioka T, Serrato OP, Montaño AM, Yamaguchi S, Noguchi A. 2004a. Identification of a common mutation in mucopolysaccharidosis IVA: correlation among genotype, phenotype, and keratan sulfate. J Hum Genet 49:490-494.

Tomatsu S, Filocamo M, Orii KO, Sly WS, Gutierrez MA, Nishioka T, Serrato OP, Di Natale P, Montaño AM, Yamaguchi S, Kondo N, Orii T, Noguchi A. 2004b. Mucopolysaccharidosis IVA (Morquio A): identification of novel common mutations in the N-acetylgalactosamine-6-sulfate sulfatase (GALNS) gene in Italian patients. Hum Mutat 24:187-188.

Tomatsu S, Fukuda S, Cooper A, Wraith JE, Ferreira P, Di Natale P, Tortora P, Fujimoto A, Kato Z, Yamada N, Isogai K, Yamagishi A, Sukegawa K, Suzuki Y, Shimozawa N, Kondo N, Sly WS, Orii T. 1997. Fourteen novel mucopolysaccharidosis IVA producing mutations in GALNS gene. Hum Mutat 10:368-375.

Tomatsu S, Fukuda S, Cooper A, Wraith JE, Rezvi GM, Yamagishi A, Yamada N, Kato Z, Isogai K, Sukegawa K. 1995. Mucopolysaccharidosis type IVA: identification of six novel mutations among non-Japanese patients. Hum Mol Genet 4:741-743.

Tomatsu S, Fukuda S, Yamagishi A, Cooper A, Wraith JF, Hori T, Kato Z, Yamada N, Isogai K, Sukegawa K, Kondo N, Suzuki Y, Shimozawa N, Orii T. 1996. Mucopolysaccharidosis IVA: four new exonic mutations in patients with N-acetylgalactosamine-6-sulfate sulfatase deficiency. Am J Hum Genet 58:950-962.

Tomatsu S, Nishioka T, Montaño AM, Gutierrez MA, Pena OS, Orii KO, Sly WS, Yamaguchi S, Orii T, Paschke E, Kircher SG, Noguchi A. 2004c. Mucopolysaccharidosis IVA: identification of mutations and methylation study in GALNS gene. Journal of Medical Genetics 41:e98-e98.

Tomatsu S, Montaño AM, Nishioka T, Gutierrez MA, Peña OM, Tranda firescu GG, Lopez P, Yamaguchi S, Noguchi A, Orii T. 2005. Mutation and polymorphism spectrum of the GALNS gene in mucopolysaccharidosis IVA (Morquio A). Hum Mutat 26:500-512.

Tylki-Szymanska A, Czartoryska B, Bunge S, van Diggelen OP, Kleijer WJ, Poorthuis BJ, Huijmans JG, Gorska D. 1998. Clinical, biochemical and molecular findings in a two-generation Morquio A family. Clin Genet 53:369-374.

Wang Z, Zhang W, Wang Y, Meng Y, Su L, Shi H, Huang S. 2010. Mucopolysaccharidosis IVA mutations in Chinese patients: 16 novel mutations. J Hum Genet 55:534-540.

Yamada N, Fukuda S, Tomatsu S, Muller V, Hopwood JJ, Nelson J, Kato Z, Yamagishi A, Sukegawa K, Kondo N, Orii T. 1998. Molecular heterogeneity in mucopolysaccharidosis IVA in Australia and Northern Ireland: nine novel mutations including T312S, a common allele that confers a mild phenotype. Hum Mutat 11:202-208.

Yang CF, Tsai FJ, Lin SP, Lee CC, Wu JY. 2001. A novel in-frame deletion mutation (c106-111del) identified in a Taiwan Chinese patient with type IVA mucopolysaccharidosis. Hum Mutat 18:254.

Ye J, Lei HL, Zhang HW, Qiu WJ, Han LS, Wang Y, Li XY, Gu XF. 2013. Analysis of GALNS gene mutation in thirty-eight Chinese patients with mucopolysaccharidosis type IVA. Zhonghua Er Ke Za Zhi 51:414-419.

Zhao H, Van Diggelen OP, Thoomes R, Huijmans J, Young E, Mazurczak T, Kleijer WJ. 1990. Prenatal diagnosis of Morquio disease type A using a simple fluorometric enzyme assay. Prenat Diagn 10:85-91.

Zhao Y, Meng YX, Guo YB, Du ML, Ai Y. 2011. Identification of a novel mutation of GALNS gene from a Chinese pedigree with mucopolysaccharidosis type IV A. Zhonghua Yi Xue Yi Chuan Xue Za Zhi 28:241-246.