Supplemental table 1. Sequence variants - Point mutations in our population (N=55)

EXON	DNA	PROTEIN*	FREQUENCY	REFERENCE**	PHENOTYPE	PHENOTYPE IN OUR POPULATION
5'UTR Exon 2	c.-55C>T	p.-	10/55	Niesler (2007) Hum Mutat 26(2)-63-8	LWD/ISS	LWD (8 patients) /No sign (1 patient) / Not available (1 patient)
5'UTR Exon 2	c.-9del	p.-	1/55	not described		LWD
Exon 2	c.169C>T	p.Gln57*	1/55	not described		LWD
Exon 2	c.178A>C	p.Thr60Pro	2/55	Huber (2006) J Med Genet 43:735	ISS	LWD (IC) / Not available (RC)
5'UTR Exon 2	c.243G>A	p.-	1/55	Esoterix (2007) SHOX LSDB entry Unpublished		LWD
Exon 2	c.257dup	p.Thr87Hisfs*10	1/55	not described		LWD
Intron 2	c.278-27G>T	p.-	1/55	not described		LWD
Intron 2	c.278-89C>T	p.-	1/55	not described		LWD
Exon 3	c.334C>T	p.Gln112*	1/55	Huber (2001) J Med Genet 38:323	LWD	LWD
Exon 3	c.391G>T	p.Glu131*	2/55	Niesler (2007) Hum Mutat 26(2)-63-8	LWD/ISS	LWD
Exon 3	c.394C>G	p.Leu132Val	3/55	Grigelioniene (2000) Hum Genet 107:145	LWD	LWD
Exon 3	c.397-452dup	p.Ser151Argfs*60	1/55	not described		LWD
Exon 3	c.414G>C	p.Glu138Asp	1/55	Esoterix (2007) SHOX LSDB entry Unpublished	ISS	LWD
Exon 3	c.445G>T	p.Glu149*	1/55	Huber (2001) J Med Genet 38:323	LWD	LWD
Exon 3	c.452G>A	p.Ser151Asn	2/55	not described		LWD
Exon 3	c.479G>T	p.Arg160Leu	1/55	Esoterix (2007) SHOX LSDB entry Unpublished	LWD/ISS	LWD
Intron 3	c.487-2A>G	p.-	2/55	Niesler (2007) Hum Mutat 26(2)-63-8	LWD	LWD
Exon 4	c.491G>C	p.Trp164Ser	2/55	Niesler (2007) Hum Mutat 26(2)-63-8	LMD	LWD (IC) / Not available (RC)
Exon 4	c.508G>C	p.Ala170Pro	4/55	Sabherwal (2004) J Med Genet 41:e83	LWD	LWD
Exon 4	c.517C>T	p.Arg173Cys	2/55	Huber (2001) J Med Genet 38:323	LWD	LWD
Exon 5	c.583C>T	p.Arg195*	2/55	Rao (1997) Nat Genet 16:54	LWD	LWD (IC) / Not available (RC)
Exon 5	c.584dup	p.Val196Serfs*195	4/55	not described		ISS (IC) / LWD (RC) / Not available (2 RC)
Exon 5	c.586G>C	p.Val196Leu	1/55	not described		Not available
Exon 5	c.599T>A	p.Val200Asp	1/55	not described		LWD
Exon 5	c.632A>T	p.Gln211Leu	1/55	not described		LWD

Intron 5	c.634-14G>T	p.-	1/55	Esoterix (2007) SHOX LSDB entry Unpublished		LWD
Exon 6	c.676C>T	p.Pro226Ser	1/55	not described		Not available
Exon 6	c.755C>A	p.Ser252*	1/55	Esoterix (2007) SHOX LSDB entry Unpublished	LWD	LWD
Exon 6a	c.784G>T	p.Val262Leu	2/55	not described		LWD
Exon 6a	c.827T>C	p.Ile276Thr	1/55	Huber (2006) J Med Genet 43:735	LWD/ISS	LWD

*According Genome version Hg19 NM_000451.3

** According The Human Gene Mutation Database (HGMD)

LWD= Leri-Weill dyschondrosteosis; LMD= Langer Mesomelic dysplasia; ISS= Isolated Short Stature; IC= Index Case; RC= Related Case

Not available= missing data to make the difference between ISS and LWD