Table1: All reported patients with OI type VI and *SERPINF1* mutations

	Cho et al [2013]	Caparrós-Martin et al [2013]				Tucker et al [2012]	Venturi et al [2012]		
		Family 8 (Patient 9)	Family 8 (Patient 10)	Family 9 (Patient 11)	Family 10 (Patient 12)		Patient V-1	Patient V-3	Patient V-4
OI Type	VI	VI	VI	VI	VI	VI	VI	VI	VI
Birth weight/Birth length	Normal/Normal	_	_	_	_	_	Normal/Normal	Normal/Normal	Normal/Normal
Confirmed prenatal fractures	No	_	_	_	_	_	No	No	No
Age of first fracture (months)	18	4	4	4	6	11	11	6	12
Color of sclerae	White	Faint Blue	Faint Blue	White	Very Faint Blue	_	Grayish	Grayish	Grayish
Dentinogenesis imperfecta	No	Yes	No	No	No	_	No	No	No
Retarded gross motor functions	No	Yes	Yes	Yes	Yes	Yes	No	Mild	No
Normal intelligence	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Hearing impairment	No	No	No	No	No	No	-	-	-
Hipermobility of joints	No	Yes	Yes	No	Yes	_	Yes	Yes	Yes
Age at start of treatment with pamidronate (years)	3	_	_	_	_	7	_	_	_
Ability to walk (with crutches)	No	No	No	No	No	Yes	No	No	Autonomously
Weight (kg)/BMI of last visit)	_	_	_	_	_	_	_	_	_
Lenght (cm)/SD of last visit)	100/>-2	_	_	_	_	159/<3rd centile	_	_	_
Wormian bones	_	Yes	No	Yes	Yes	_	_	_	_
Vertebral fractures	No	Yes	_	Yes	_	No	Yes	Yes	Yes
Old fractures of extremities	Yes	_	Yes	_	_	Yes	Yes	Yes	_
Tapering of ribs	No	Yes	_	Yes	_	_	_	_	_
Popcorn calcification	Yes	Yes	_	Yes	No	_	_	_	_
Rhizomelic shortening of limbs	No		_	Yes	Yes	_	_	_	_
Bowing of upper extremities	_	Severe	Severe	Severe	Yes	_	Yes	Severe	No
Bowing and tapering of femur, tibiae and fibulae	Severe bowing	Bowing of femurs	Bowing	Severe tapering of tibiae and fibulae	Bowing of femur	_	Bowing of femurs	Moderate bowing of femurs	No
Exon	2 ; 4	6				3	4		
DNA change	c.77dupC; c.421dupC	c.752_753insKC847088.1:g.51_393		c.651G>A		c.271_279dup	c.423delG		
Protein	p.Glu27Glyfs*38; p.Arg141Profs*5	_		p.Trp217*		p.Ala91_Ser93dup	p.Ile142 Serfs*9		
Origin	Korean	Egyptian				-	Italian/Sweish/French		

	Shaheen et al [2012]			Rauch et al [2012]/Glorieux et al[2002]					
	OI_F5	**OI_F6**	**OI_F12**	**Patient 1**	**Patient 2**	**Patient 3**	**Patient 4**	**Patient 5**	**Patient 6**
OI Type	VI	VI	VI	VI	VI	VI	VI	VI	VI
Birth weight/Birth length	-	-	-	Normal	Normal	Normal	Normal	Normal	Normal
Confirmed prenatal fractures	-	-	-	No	No	No	No	No	No
Age of first fracture (months)	-	-	-	4-18	4-18	4-18	4-18	4-18	4-18
Color of sclerae	Blue	Blue	White	White	White	White	White	White	White
Dentinogenesis imperfecta	No	No	No	No	No	No	No	No	No
Retarded gross motor functions	-	-	-	-	-	-	-	-	-
Normal intelligence	-	-	-	-	-	-	-	-	-
Hearing impairment	No	No	No	-	-	-	-	-	-
Hipermobility of joints	-	-	-	2 patients					
Age at start of treatment with pamidronate (years)	-	-	-	-	-	-	-	-	-
Ability to walk (with crutches)	No	No	No	4 patients wheelchair bound; 2 patientes ambulatory; 2 patients walked with assistance					
Weight (kg)/BMI of last visit)	-	-	-	-	-	-	-	-	-
Lenght (cm)/SD of last visit)	-	110//<-2SD	-	-	-	-	-	-	-
Wormian bones	-	-	-	No	No	No	No	No	No
Vertebral fractures	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Old fractures of extremities	-	-	-	Yes	Yes	Yes	Yes	-	Yes
Tapering of ribs	-	-	-	-	-	-	-	-	-
Popcorn calcification	-	Yes	-	-	-	-	-	-	-
Rhizomelic shortening of limbs	-	-	-	Yes (femurs)	-	-	-	-	-
Bowing of upper extremities	-	Severe	-	Yes	Yes	-	-	Yes	Bowing of left humerus
Bowing and tapering of femur, tibiae and fibulae	-	Severe bowing of femurs	-	Yes (tibiae)	Bowing of tibiae	-	Bowing of left femur	-	-
Exon	8	1	6	4		3 ; 4	3	4	No Mutation found
DNA change	c.1118_1119del	c.1-4796dupT	c.653del	c.295C>T		c.271_279dup; c.295C>T	c.271_279dup	c.295C>T	
Protein	p.Pro373Glnfs*18	-	p.Val218Glufs*22	p.Arg99*		p.Ala91_Ser93dup; p.Arg99*	p.Ala91_Ser93dup	p.Arg99*	
Origin	Arabian			French-Canadian		French/Canadian/ Irish	Nicaraguan	French/ Canadian/German	North Quebec

	Rauch et al [2012]/Glorieux et al[2002]					
	Patient 7	**Patient 8**	**Patient 9**	**Patient 10**	**Patient 11**	**Patient 12**
OI Type	VI	VI	VI	VI	VI	VI
Birth weight/Birth length	Normal	Normal	-	-	-	-
Confirmed prenatal fractures	No	No	-	-	-	No
Age of first fracture (months)	4-18	4-18	-	-	-	>6
Color of sclerae	White	White	-	-	-	White
Dentinogenesis imperfecta	No	No	-	-	-	No
Retarded gross motor functions	-	-	-	-	-	-
Normal intelligence	-	-	-	-	-	-
Hearing impairment	-	-	-	-	-	-
Hipermobility of joints	2 patients		-	-	-	-
Age at start of treatment with pamidronate (years)	-	-	-	-	-	-
Ability to walk (with crutches)	4 patients wheelchair bound; 2 patientes ambulatory; 2 patients walked with assistance		-	-	-	-
Weight (kg)/BMI of last visit)	-	-	-	-	-	-
Lenght (cm)/SD of last visit)	-	-	-	-	-	-
Wormian bones	No	No	-	-	-	-
Vertebral fractures	Yes	Yes	-	-	-	-
Old fractures of extremities	Yes	Yes	-	-	-	-
Tapering of ribs	-	-	-	-	-	-
Popcorn calcification	-	-	-	-	-	-
Rhizomelic shortening of limbs	-	-	-	-	-	-
Bowing of upper extremities	-	Yes	-	-	-	-
Bowing and tapering of femur, tibiae and fibulae	-	Bowing of lower extremities	-	-	-	-
Exon	3	4	4	4		7
DNA change	c.271_279dup	c.440_643del	c.440_643del	c.295C>T		c.829_831del
Protein	p.Ala91_Ser93dup	p.Lys147_Gly215 delinsArg	p.Lys147_Gly215 delinsArg	p.Arg99*		p.Phe277del
Origin	German/Polish	Equadorian	Equadorian	French- Canadian		Nepalese

	Becker et al [2011]				Homan et al [2011]		
	Patient 1	**Patient 2**	**Patient 3**	**Patient 4**	**Patient V-1**	**Patient IV-3**	**Patient 3**
OI Type	III	III	III	III	VI	VI	VI
Birth weight/Birth length	Normal/Normal	Normal/Normal	Normal/Normal	Normal/Normal	Normal/_	Normal/_	Normal/_
Confirmed prenatal fractures	No	No	No	No	No	No	-
Age of first fracture (months)	4	Unknown	6	6	9	-	10
Color of sclerae	Grayish Grayish	Grayish	Grayish	Grayish	-	White	Slightly blue
Dentinogenesis imperfecta	No	No	No	No	No	-	-
Retarded gross motor functions	Yes	Yes	Yes	Yes	Yes	Yes	-
Normal intelligence	Yes	Yes	Yes	Yes	-	-	-
Hearing impairment	No	No	No	No	-	-	-
Hipermobility of joints	No	No	No	No	No	No	No
Age at start of treatment with pamidronate (years)	1	11	7	1	0	13	2
Ability to walk (with crutches)	No	No	No	No	No	No	No
Weight (kg)/BMI of last visit)	9,9/17,1	45/44,1	17/37,8	14/22,8	No	No	No
Lenght (cm)/SD of last visit)	76/-5,6	101/-10,2	67/0,2	78,3/-0,7	-	110//<-2SD	-
Wormian bones	-	-	-	-	-	-	v
Vertebral fractures	Multiple	Multiple	Multiple	Multiple	-	-	-
Old fractures of extremities	Yes	Yes	Yes	Yes	Yes	Yes	Yes
Tapering of ribs	-	-	-	-	-	-	-
Popcorn calcification	-	-	-	-	-	Yes	-
Rhizomelic shortening of limbs	Moderate	Severe	Mild upper and moderate lower	Mild	-	-	-
Bowing of upper extremities	Moderate	Severe	Mild	Moderate	-	Severe	-
Bowing and tapering of femur, tibiae and fibulae	Severe bowing	Sebere bowing	Moderate bowing	Moderate bowing	_	Severe bowing of femurs	_
Exon	6		4	8	4	4	8
DNA change	c.696C>G		c.324_325dup	c.1132C>T	g.4130C>T	g.4130C>T	g.10440_10443dupATCA
Protein	p.Tyr232*		p.Tyr109Serfs*5	p.Gln378*	p.R99X	p.R99X	p.H389fsX392
Origin	Arabian		Turkish	Turkish	-	French/ Canadian/Irish	Italian