Table SII. Reported WNT10A mutations and genotypes in patients diagnosed with Schöpf-Schulz-Passarge syndrome. The most commonly identified mutation is c.321C>A;p.Cys107*. So far, three heterozygous and eight homozygous WNT10A genotypes resulting from 11 different mutations have been described.

								First allele		Second allele	
Autors	**Year of publi-cation**	**Number of patients**	**Homo-zygous**	**Compound hetero-zygous**	**Mis-sense**	**Non-sense**	**Exon**	**Nucleotide substitution**	**Amino-acid substitution**	**Nucleotide substitution**	**Amino-acid substitution**
Bohring et al.	2009	1	X			X	2	c.321C>A	p.Cys107*	c.321C>A	p.Cys107*
Nagy et al.	2010	1	X			X	2	c.321C>A	p.Cys107*	c.321C>A	p.Cys107*
Castori et al.	2011	1	X		X		4	c.796G>T	p.Gly266Cys	c.796G>T	p.Gly266Cys
		3	X		X		3	c.391G>A	p.Ala131Thr	c.391G>A	p.Ala131Thr
Petrof et al.	2011	1	X			X	2	c.321C>A	p.Cys107*	c.321C>A	p.Cys107*
Wedgeworth et al.	2011	1	X			X	2	c.321C>A	p.Cys107*	c.321C>A	p.Cys107*
Tziotzios et al.	2014	1	X			X	4	c.1168G>T	p.Glu390*	c.1168G>T	p.Glu390*
		1	X		X		4	c.1084T>C	p.Cys362Arg	c.1084T>C	p.Cys362Arg
		1		X	X	X	2,4	c.321C>A	p.Cys107*	c.810C>A	p.Ser270Arg
		1		X	X	X	2,3	c.321C>A	p.Cys107*	c.391G>A	p.Ala131Thr
		3	X			X	2	c.321C>A	p.Cys107*	c.321C>A	p.Cys107*
Vilas-Sueiro et al.	2015	1	X		X		3	c.682T>A	p.Phe228Ile	c.682T>A	p.Phe228Ile
					X		4	c.831G>T	p.Trp277Cys	c.831G>T	p.Trp277Cys
Painsi et al.	2017	1	X		X		4	c.1135C>T	p.Arg379Cys	c.1135C>T	p.Arg379Cys
Pauly et al.	2018	1		X	X		2,4	c.318C>G	p.Asn106Lys	c.1036T>C	p.Cys246Arg
present study	2018	2		X		X	2,3	c.321C>A	p.Cys107*	c.742C>T	p.Arg248*
		1				X	3			c.742C>T	p.Arg248*