

Table SII. List of SDR9C7 mutations associated with autosomal recessive congenital ichthyosis (ARCI)

Nucleotide change ^a	Amino acid change ^a	Exon	Type of variation	dbSNP number	MAF in <i>GnomAD</i>	References
c.112G>A	p.(Gly38Arg)	Exon 1	Missense	rs764593071	5.66e-5	(3) ; (11)
c.214C>T	p.(Arg72Trp)	Exon 1	Missense	rs530109812	3.98e-6	(5)
c.309G>A	p.(Trp103*)	Exon 2	Nonsense	rs1441151450	abs.	This report (P3)
c.355G>A	p.(Glu119Lys)	Exon 2	Missense	rs538068583	3.98e-6	(11) ; (12)
c.364dupA	p.(Tyr122Asnfs*4)	Exon 2	Frameshift	rs760309815	3.98e-5	(7)
c.551A>G	p.(Asp184Gly)	Exon 2	Missense	rs138435128	4.61e-4	(3) ; (4) ; this report (P2)
c.562C>T	p.(Arg188Cys)	Exon 3	Missense	rs150520393	4.25e-5	(3)
c.563G>A	p.(Arg188His)	Exon 3	Missense	rs141626978	1.35e-4	This report (P2)
c.599T>C	p.(Ile200Thr)	Exon 3	Missense	rs770729222	1.59e-5	(4) ; (5)
c.658C>T	p.(Arg220*)	Exon 3	Nonsense	rs774363396	1.99e-5	(3) ; (11)
c.704G>A	p.(Gly235Glu)	Exon 3	Missense	rs756591404	3.98e-6	This report (P1)
c.774-795del	p.(Arg258Serfs*32)	Exon 4	Frameshift	abs.	abs.	(9)
c.826C>T	p.(Arg276Cys)	Exon 4	Missense	rs755404310	1.06e-5	This report (P3)
c.831_832delCA	p.(Tyr277*)	Exon 4	Frameshift	abs.	abs.	(3)
c.897delT	p.(Phe300Serfs*3)	Exon 4	Frameshift	abs.	abs.	(8)

^aReference sequences SDR9C7: NM_148897.2, NP_683695.1

dbSNP: Single Nucleotide Polymorphism Database; MAF: minimum allele frequency; *GnomAD*: Genome Aggregation Database.