

Table S1. SLC27A4 mutations identified in individuals with ichthyosis prematurity syndrome

Type of mutation	Position	Reference
Nonsense/frameshift		
c.103G>T, p.Gly35*	Exon 2	Sobol et al., 2011 (13)
c.504C>A, p.Cys168*	Exon 3	Klar et al., 2009 (2)
c.1065delT, p.Val355Valfs*4	Exon 8	Bueno et al., 2017 (14)
Missense		
c.1A>G, p.Met1?	Exon 2	Inhoff et al., 2011 (8)
c.274G>A, p.Ala92Thr	Exon 3	Klar et al., 2009 (2)
c.625G>A, p.Gly209Ser	Exon 4	Gertow et al., 2004 (15)
c.739T>C, p.Ser247Pro	Exon 5	Klar et al., 2009 (2)
c.899A>G, p.Gln300Arg	Exon 7	Klar et al., 2009 (2)
c.931C>T, p.Arg311Trp	Exon 7	This study
c.986C>T, p.Thr329Met	Exon 7	Khnykin et al., 2012 (1)
c.1012G>A, p.Cys338Tyr	Exon 8	Bueno et al., 2017 (14)
c.1120C>T, p.Arg374Cys	Exon 8	Morice-Picard et al., 2010 (4)
c.1430T>A, p.Val477Asp	Exon 10	Sobol et al., 2011 (13)
c.1510C>T, p.Arg504Cys	Exon 11	Bueno et al., 2017 (14)
c.1511G>A, p.Arg504Hys	Exon 11	Sobol et al., 2011 (13)
c.1748G>A, p.Arg583His	Exon 12	Klar et al., 2009 (2)
Splicing		
c.716-1G>A	Intron 4	Klar et al., 2009 (2)
c.988-2A>G	Intron 7	Klar et al., 2009 (2)