Table SI. Characteristics of the *PNPLA1* mutations identified in our cohort of autosomal recessive congenital ichthyosis patients

Family	Nucleotide change	Aminoacidic change	Location	Domain	Resultant change	Mutation type	In Silico Prediction			MAF	Reference
							Mutation taster	SIFT	Align GVGD		
18, 62	c.100G>A	p.(Ala34Thr)	Exon 1	Patatin	Moderately conserved residue	Missense	Disease causing	Tolerated	Least likely to interfere with function	A= 0	14
46, 115	c.282dup	p.(Lys95*)	Exon 2	Patatin	Truncated mRNA	Nonsense					This study
Various	c.417_418delinsTC	p.(Ser140Pro)	Exon 2	Patatin	Highly conserved residue	Missense	Disease causing	Deleterious	Most likely to interfere with function	TC= 1.8E-06	4, 5
69	c.729C>G	p.(Tyr243*)	Exon 5	Extended patatin	Truncated mRNA	Nonsense					This study
137	c.820del	p.(Arg274Glyfs*8)	Exon 6	Extended patatin	Truncated mRNA	Frameshift					6
98	c.892C>T	p.(Arg298*)	Exon 6		Truncated mRNA	Nonsense				T= 8.9E-07	This study
62	c.1143del	p.(Pro382Alafs*74)	Exon 6		Truncated mRNA	Frameshift					4

Mutation nomenclature: the Human Genome Sequence Variation guideline was followed. Reference sequences PNPLA1 (NM_001145717, NP_001139189) were used for naming the nucleotide and protein variations respectively. Available Minor Allele Frequencies (MAF) of European Non-Finnish population were revised in the gnomAD database (http://gnomad.broadinstitute.org/). References can be consulted in the manuscript.