

Table S1. Summary of reported autosomal recessive congenital ichthyosis (ARCI) families with ALOXE3 mutations

Family	Mutations	Phenotype	Collodion membrane	Erythema	PPK	Depression/fungal infection/ other features	Ref.
1	p.Val1500Phe	CIE	+	+	ND	ND	1
2	p.Arg234X/p.Arg234X	CIE	+	+	ND	ND	1
3	p.Arg396Ser/p.Arg396Ser	CIE	+	+	ND	ND	1
4	p.Arg234X/p.Gly281Val	ND	ND	ND	ND	ND	3
5	p.Arg234X/p.Arg234X	CIE	-	+	-	+	3
6	p.Leu237Met/p.Leu237Met	CIE	ND	+	ND	ND	3
7	p.Gln544X/p.Gln544X	ND	ND	ND	ND	ND	3
8	p.630Leu/p.630Leu	CIE/LI	+	+, mild	-	Anhidrosis	3
9	p.630Leu/p.630Leu	CIE	+	+, only facial	-	ND	3
10	p.Arg234X/p.630Leu	LI	+	-	-	Reduced sweating	4
11	p.Gln344_Ala347delinsPro/	LI	+	-	-	Reduced sweating	4
12	p.Gln344_Ala347delinsPro	CIE/LI	+	+, mild, on the scalp and face	-	Reduced sweating	4
13	p.Arg234X/p.630Leu	CIE	+	+, mild, on the face, trunk and abdomen	-	Reduced sweating, hyperlinearity on hands/feet	4
14	p.Arg234X/p.630Leu	CIE	-	+, mild	-	Reduced sweating, hyperlinearity on hands/feet	4
15	p.630Leu/p.630Leu	CIE	+	+, mild, on the trunk and abdomen	-	Hyperlinearity on hands/feet	4
16	p.630Leu/p.630Leu	CIE	-	-	-	Reduced sweating, hyperlinearity on hands/feet	4
17	p.Arg234X/p.Arg234X	CIE	+	+, mild	-	Hyperlinearity on hands/feet	4
18	p.Lys240ArgfsX40/p.Pro630Leu	CIE	ND	+, mild to moderate	+	ND	4
19	p.Arg234X/p.Arg234X	CIE	+	+, partial	+	Hyperlinearity on hands/feet	4
20	p.630Leu/p.630Leu	CIE	-	+, mild	+	Hyperlinearity on hands/feet	4
21	p.Arg119GlyfsX12/p.Arg119GlyfsX12	ND	ND	ND	ND	ND	4
22	p.Cys109X/p.630Leu	SICI	+	ND	+	Allergic rhinitis and prematurity	5
23	p.Leu427Pro/=	SICI	+	ND	-	Pruritus	5
24	p.Arg234X/p.630Leu	SICI	+	ND	-	Syndactyly of 2 fingers	5
25	p.Leu237Met/=	LI	-	+	+	Prematurity and alopecia	6
26	p.Arg119GlyfsX12/p.Arg234X	CIE	-	-	-	ND	6
27	p.Gln344_Ala347delinsPro/	CIE	ND	ND	ND	ND	7
28	p.Gln344_Ala347delinsPro	CIE	ND	ND	ND	ND	7
29	p.Gln344_Ala347delinsPro	LI	-	-	+	-	8
30	p.Leu331Pro/p.Leu331Pro	CIE	-	+	+	Fungal infection	9
31	p.Arg272X/p.Arg272X	CIE	-	+	ND	Minor sweating, severe heat intolerance, and bleeding from scalling skin	10
32	p.Arg140X/p.Arg140X	CIE	-	+	ND	Minor sweating, severe heat intolerance, and bleeding from scalling skin	10
33	gross deletion (delEx1_6InsC)/gross deletion (delEx1_6InsC)	CIFS	-	ND	+	Anhidrosis	11
34	c.353-1G>C/c.353-1G>C	LI	-	ND	-	Anhidrosis	11
35	p.Pro630Leu/p.Pro630Leu	CIFS	-	ND	+	Anhidrosis	11
36	p.Cys109X/p.Pro630Leu	CIE	-	+	-	Depression and fungal infection	Present case

ARCI: autosomal recessive congenital ichthyosis; CIE: congenital ichthyosiform erythroderma; ND: not described; LI: lamellar ichthyosis; SICI: self-improving collodion; CIFS: congenital ichthyosis with fine/focal scaling; PPK: palmoplantar keratoderma.