

Table S1. Characteristics of the 9 families (12 patients) reported in the literature and our patient

Reference	3	6	7	8, 9	10	11	12	13	14	Our patient
Familial cases	-	2 dizygotic twins, 1 sibling	-	-	2 dizygotic twins	-	-	-	-	-
Origin	ND	African	Caucasian	Caucasian	African	ND	ND	Asian	ND	African
Suspected mode of inheritance	<i>De novo</i>	Germline, mosaic	<i>De novo</i>	<i>De novo</i>	Somatic, mosaic	<i>De novo</i>	<i>De novo</i>	<i>De novo</i>	<i>De novo</i>	<i>De novo</i>
Age at first signs	Birth	Birth	2 months	Birth	Birth	Birth	Birth	Birth	Birth	1 Month
Dystrophic nails	+	+	-	+	+	-	-	+	+	+
Keratitis	-	-	+	+	-	-	-	+	+	+
Hearing deficit	+	ND	+	+	ND	ND	+	+	+	ND
Other abnormalities	+	+	+	-	+	+	+	-	-	-
Age at death	3 months	1 month, 1 month, 5 months	12 months	6 months	30 days, 5 months	2 months	3 months	51 days	77 days	15 months
Cause of death	Septicaemia	Septicaemia	Septicaemia	Septicaemia	Septicaemia	Septicaemia	Septicaemia	Septicaemia	Septicaemia	Septicaemia
<i>GJB2</i> mutation	p.Ala88Val	p.Gly45Glu	p.Gly45Glu	p.Gly45Glu	p.Gly45Glu	ND	ND	ND	ND	p.Ser17Phe

ND: not determined.