

**Table SI. Phenotypes of cases 1–3 compared with typical Mal de Meleda (MDM) and Gamborg-Nielsen (GN) palmoplantar keratoderma (PPK)**

	Case 1	Case 2	Case 3	MDM	GN
SLURP1 mutation	c.178G>A p.(Glu60Lys)	c.178G>A p.(Glu60Lys)	c.82del p.(Cys28Alafs*5)		
SLURP1 mutation	c.218_220del p.(Cys73del)	c.178G>A p.(Glu60Lys)	c.286C>T p.(Arg96*)		
PPK					
Diffuse	+	+	+	+	+
Erythematous border	+	+	+	+	+
Transgrediens	+	+	-	+	+
Progrediens	+	-	-	+	-/+
Inflammatory erythema	-	-	-	+	-
Aquagenic whitening	+	+	-	NR	NR
Hyperkeratosis					
Elbows	+	+	-	+	-
Knees	+	-	-	+	-
Knuckle pads	+	-	-	+	+
Nail abnormalities	+	+	-	+	-
Hyperhidrosis	-	+	-	+	+
Conical tapering of fingers	-	-	-	+	+
Brachydactyly	-	-	-	+	-
Pseudoainhum	-	-	-	+	-
Perioral erythema	-	-	-	+	-

NR: not reported.