

Table SI. Detailed information on the 4 cases

	1	2	3	4
Histological findings	Not available	Flat epidermis, pronounced hypoplasia of appendages, reduction of elastic material, arrangement of collagen-bundles parallel to the skin surface (hypoplastic lungs)	Flat epidermis without rete ridges, pronounced hypoplasia of appendages, reduction of elastic material, arrangement of collagen-bundles parallel to the skin surface No clavicular, small thorax	Slightly broadened orthohyperkeratotic epithelium, flat dermis, partly hypoplasia of appendages, arrangement of collagen-bundles parallel to the skin surface No pathological findings Thin, tense, vulnerable and translucent skin with superficial erosions
X-ray	Hypoplastic lungs and clavicular	Erythrodermia, tense, scaling and translucent skin, superficial erosions	Thin, tense, vulnerable and translucent skin	Thin, tense, vulnerable and translucent skin with superficial erosions
Facial dysmorphic signs	Small open-mouth in the o-position, small pinched nose, antimongoloid palpebral fissure, low-set ears, hypertelorism, micro- and retrognathia, high forehead	Small open-mouth in the o-position, low-set ears, hypertelorism, micrognathia	Small open-mouth in the o-position, small pinched nose, small palpebral fissure, low-set ears, micrognathia	Small open-mouth in the o-position, small pinched nose, low-set ears, hypertelorism, micrognathia
Musculoskeletal system	Arthrogryposis of all joints	Arthrogryposis of all joints	Arthrogryposis of all joints	Arthrogryposis of all joints
Genetic mutation	Not available.	Not available.	Not available.	Homozygous for c.1085-1086insT, ZMPSTE24-Gene
Family				Prior miscarriage: Similar clinical and histological findings. Besides hypertrophy of the pancreatic islets of Langerhans, probable cause of death: hypoglycaemia
Birth	33+4 weeks reduced movements APGAR: 1-6-6	32+4 weeks, reduced movements (premature rupture of the membranes) APGAR: 8-8-8	29+6 weeks reduced movements (premature rupture of the membranes) APGAR: 4-5-6	29+5 weeks reduced movements (premature rupture of the membranes) APGAR: 8-9-9
Survival/cause of death	6 days cardio-respiratory insufficiency	32 days respiratory insufficiency	9 days Not available	14 days respiratory insufficiency