Table SI. *Families, age, gender, clinical characteristics, family history, modes of inheritance, and results of molecular genetic investigations. Mutations named according to the international recommendations for nomenclature for sequence variants (http: //www.hgvs.org/mutnomen/).*

Family No. Sex/age	Affected family members	Diagnosis/mode of inheritance/ OMIM No.	Gene affected, and mutation	Effect of mutation on protein	Clinical presentation					
					Skin	Hair	Nails	Teeth	Sweat glands	Other
1: F/23	Yes	Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy OMIM: 240300	Not tested		Eczema	Alopecia totalis	Dystrophy	Hypoplasia	Normal	Keratoconjunctivitis Hypoparathyroidism Adrenal insufficiency Recurrent candidiasis Peripheral facial palsy
2: F/3, F/61, F†, F/91, F/92, M/30	Yes	Clouston syndrome OMIM: 129500	*GJB6* c.31G>A[a]	p.G11R	Hyperkeratosis	Hypotrichosis	Hyperkeratosis	Normal Sharp incisors	Hypohidrosis	Recurrent paronychia
3: M/67	No	Clouston syndrome	*GJB6* c.31G>A[a]	p.G11R	Hyperkeratosis Squamous cell carcinoma	Hypotrichosis	Hyperkeratosis Onycholysis	Normal	Normal Hypohidrosis	
4: F/27, M/25	Yes	Clouston syndrome	*GJB6* c.31G>A[a]		Normal	Hypotrichosis	Hyperkeratosis	Normal	Normal	
5: M/34	No	Clouston syndrome	Not tested		Normal	Hypotrichosis	Dystophy Hyperkeratosis Hnycholysis	Enamel defect	Hypohidrosis	
6: F/14	No	Ectodermal dysplasia syndactyly syndrome OMIM: 613573	*PVRL4* exon 2 deletion[a]	In-frame deletion	Normal	Hypotrichosis	Hyperkeratosis	Enamel defect	Normal	Syndactyly
7: F/7	No	Focal dermal hypoplasia OMIM: 305600	*PORCN* c.947-1G>C	Splicing defect	Dermal focal hypoplasia Xerosis cutis	Hypotrichosis	Normal	Hypodontia	Normal	Syndactyly
8: F/5	No	Focal dermal hypoplasia	*PORCN* c.813-1G>T[a]	Splicing defect	Dermal focal hypoplasia Xerosis cutis	Hypotrichosis	Normal	Hypodontia	Normal	Syndactyly Anisomelia
9: M/16	No	Focal dermal hypoplasia	Not found		Dermal focal hypoplasia Xerosis cutis	Hypotrichosis	Normal	Hypodontia	Normal	Syndactyly
10: F/20	No	Focal dermal hypoplasia	Not tested		Dermal focal hypoplasia Xerosis cutis	Normal	Normal	Hypodontia	Normal	Syndactyly
11: F/23, M/15	Yes	Microcephaly, growth retardation, cataract, hearing loss, and unusual appearance OMIM: 612947	Not found		Dry skin Eczema	Normal	Normal	Enamel defect	Normal	Hearing loss Almond-shaped eyes Bulbous nose Delayed bone age
12: F/17	No	Ankyloblepharon-ectodermal dysplasia-clefting syndrome OMIM: 106260	*TP63* c.1649T>C	p.I550T	Eczema	Hypotrichosis	Dystrohy	Oligodontia	Normal	Lacrimal duct obstruction Cleft-lip-palate Ankyloblepharon
13: F/19, M/15	Yes	XLHED OMIM: 305100	*EDA* c.1045G>A	p.A349T	Eczema	Hypotrichosis	Normal	Oligodontia	Hypohidrosis	Nasal discharge Rhinitis Saddle nose

Table SI contd.

14: M/56	Yes	XLHED	*EDA* c.466C>T	p.R156C	Eczema	Hypotrichosis	Normal	Oligodontia	Hypohidrosis	Frontal bossing Nasal discharge Saddle nose Cataract Retinal detachment
15:F/8, F/11, M/37	Yes	XLHED	Not found		Eczema	Alopecia Hypotrichosis	Normal	Oligodontia Enamel defect	Anhidrosis Hypohidrosis	Athelia Goiter Poland syndrome
16:F/11, F/43, F/43, M/3, M/11, M/15	Yes	HED OMIM: 129490	*EDAR* c.1082delG[a]	Frameshift	Eczema	Hypotrichosis	Dystrophy	Oligodontia Enamel defect	Hypohidrosis	Asthma Nasal discharge Athelia Sicca symptoms Mandibular hypoplasia
17:M/13	Yes	HED	*EDA* c.708C>T	p.R156C	Eczema	Hypotrichosis	Normal	Oligodontia	Hypohidrosis	Frontal bossing Asthma Saddle nose
18:M/43	No	HED OMIM: 224900	*EDAR* c.126delG[a]	p.G42fsX61	Eczema	Hypotrichosis	Normal	Oligodontia	Anhidrosis	Nasal discharge
19:F/2	No	HED	*EDAR* c.84delC[a] (*de novo*)	Frameshift	Eczema	Hypotrichosis	Normal	Oligodontia	Hypohidrosis	Nasal discharge Frontal bossing
20:M/6	No	HED	Not found		Eczema	Hypotrichosis	Normal	Oligodontia	Hypohidrosis	Frontal bossing Nasal discharge Rhinitis
21:M/37	No	HED	Not tested		Eczema	Hypotrichosis	Normal	Oligodontia	Hypohidrosis	Nasal discharge Saddle nose
22:M/33	No	HED	Not tested		Eczema	Hypotrichosis	Normal	Oligodontia	Anhidrosis	Frontal bossing Asthma
23:M/15	No	HED	Not tested		Eczema	Normal	Normal	Oligodontia	Anhidrosis	Asthma
24:M/46	No	HED	Not tested		Normal	Normal	Normal	Oligodontia	Hypohidrosis	
25:F/39	Yes	Incontinentia pigmenti OMIN: 308300	*IKBKG* exon 4-10 deletion	Truncated protein	Hypopigmentation Dermal scarring	Normal	Normal	Normal	Normal	
26:F†	Yes	Incontinentia pigmenti	*IKBKG* exon 4-10 deletion	Truncated protein	Inflamation Verrucous plaques	Normal	Normal	Normal	Normal	
27:F/10	Yes	Incontinentia pigmenti	*IKBKG* exon 4-10 deletion	Truncated protein	Hypopigmentation Dermal scarring	Normal	Normal	Normal	Normal	
28:F/11	Yes	Incontinentia pigmenti	*IKBKG* exon 4-10 deletion	Truncated protein	Hypopigmentation Dermal scarring	Normal	Normal	Normal	Normal	
29:F/31	No	Incontinentia pigmenti	*IKBKG* exon 4-10 deletion	Truncated protein	Hypopigmentation Dermal scarring	Alopecia	Dystrophy	Hypodontia	Normal	
30:F/33	No	Incontinentia pigmenti	*IKBKG* exon 4-10 deletion	Truncated protein	Hypopigmentation Dermal scarring	Normal	Normal	Hypodontia	Normal	
31:F†	Yes	Incontinentia pigmenti	Not tested		Inflammation Vesicles	Normal	Dystrophy	Hypodontia	Normal	
32:M/11	No	Cardiofaciocutaneous syndrome OMIM: 115150	Not found		Normal	Brittle Curly	Normal	Normal	Normal	Growth and psychomotoric retardation Valvular pulmonal stenosis Sensorineuronal hearing loss Prominent forehead Ocular hypertelorism

Table SI contd.

33M/25	No	Keratitis–ichthyosis–deafness syndrome OMIM: 148210	*GBJ2* c.148G>A	p.D50N	Ichthyosis Recurrent candidiasis Keratoderma	Hypotrichosis	Paronychia	Normal	Hypohidrosis	Frontal bossing Keratitis Sensorineural hearing loss
34:F/14, F/40	Yes	Monilethrix OMIM: 158000	*KRT86* p.1237G>A[a]	p.E413K	Keratosis pilaris	Dry Brittle	Normal	Normal	Normal	
35:F/40	No	Monilethrix	Not tested		Keratosis pilaris	Dry Brittle	Normal	Normal	Normal	
36:F/27	No	Pachyonychia congenita 1 OMIM: 167200	*KRT 6A* c.513C>A	p.N171K	Keratoderma	Normal	Pachyonychia	Normal	Hyperhidrosis	Oral leucoplakia
37:F/41, M/14	Yes	Pachyonychia congenita 1	Not tested		Keratoderma	Normal	Pachyonychia	Normal	Hypohidrosis	Oral leucoplakia
38:F/10, M/41	Yes	Pachyonychia congenita 2 OMIM: 167210	*KRT17* c.236T>G	p.M88R	Normal	Normal	Pachyonychia	Neonatal teeth	Normal	Steatocystoma multiplex
39:F/12, F/49, M/15	Yes	Pachyonychia congenita 2	Not tested		Normal	Normal	Pachyonychia	Neonatal teeth	Normal	Steatocystoma multiplex
40:F/19	No	Trichorhinophalangeal syndrome 1 OMIM: 190350	TRPS1 c.3424delA[a] (*de novo*)	p.S1142fsX63	Normal	Hypotrichosis	Dystrophy	Normal	Normal	Prominent nose Brachydactyly Clinodactyly Short stature
41:M/31	No	Xeroderma pigmentosum-trichothiodystrophy overlap OMIM: 601675	*ERCC2* c.2173G>C, and c.50T>C[a]	p.A725P, and p. Y18H	Basal cell carcinoma Ichthyosis UV hyper-sensitivity	Brittle	Dystrophy	Normal	Normal	Cataract Chorioretinal dysplasia Diabetes Ataxia Perceptive hearing loss Hypogonadism
42:F/25, M/21, M/23	Yes	Trichothiodystrophy OMIM: 601675	Not tested		Ichthyosis	Brittle	Dystrophy	Normal	Normal	Ataxia Growth Mental retardation
43:M/32	No	Trichothiodystrophy	Not tested		Ichthyosis	Brittle	Normal	Enamel defect	Normal	Psychomotoric retardation Ataxia
44:M/10	No	Trichothiodystrophy	Not tested		Ichthyosis	Brittle	Koilonychia	Normal	Normal	Growth hormone deficiency Psychomotoric retardation Cataract
45:F/7	No	Poikiloderma congenitale OMIM: 268400	Not tested		Poikiloderma	Normal	Normal	Normal	Normal	

[a]Mutation not previously reported.