TABLE I. Genes and Chromosomal Regions for Selected Ectodermal Dysplasias

Chromosome	Gene	Protein or gene product	Inheritance	Ectodermal dysplasia (ED)	Protein Function	OMIM Number
Xp11.23	*PORCN*	Five isoforms (PORCA-PORCE)	XLD	Focal dermal hypoplasia (Goltz syndrome)	Membrane targeting and secretion of Wnt proteins necessary for embryonic tissue development	305600
Xq12-q13.1	*EDA1*	Ectodysplasin-A	XLR	XLHED	Triggering ligand molecule	305100
Xq28	*IKBKG (NEMO)*	IKK- γ (NF-kappa-B essential modulator)	XLD	Incontinentia pigmenti 2	NFκB cytoplasmic inhibitor	308300
			XLR	OL-HED-ID syndrome		300301
			XLR	Hypohidrotic ED with immune deficiency		300291
1q32	*PKP1*	Plakophilin 1	AR	ED/skin fragility syndrome	Desmosomal plaque accessory protein	604536
1q42.2-q43	*EDARADD*	Ectodysplasin-A receptor adapter	AD or AR	ADHED and ARHED	Intracellular molecule adaptor of EDAR death domain	129490, 224900
2q11-q13	*EDAR*	Ectodysplasin-A receptor	AD or AR	ADHED and ARHED	Transmembrane receptor of EDA	129490, 224900
2q35	*WNT10A*	Wingless-type MMTV integration site family, member 10A	AR	Odontoonychodermal dysplasia [OODD)	β catenin-mediated specific intracellular signaling	257980
3q27	*TP63*	p63	AD	ADULT syndrome	Transcription factor	103285
			AD	Ectrodactyly, ED, cleft lip/palate syndrome 3 (EEC3)		604292
			AD	Limb- mammary syndrome		603543
			AD	Ankyloblepharon-ectodermal defects-clefting (AEC)		106260
			AD	SHFM4 syndrome		605289
			AD	Rapp-Hodgkin syndrome (RHS)		129400
4p16.1	*MSX1*	Msx1	AD	Witkop syndrome	Transcription factor	189500
6q21-q23.2	*GJA1*	Connexin 43	AD	Oculodentodigital dysplasia (ODDD)	Connexin protein, intercellular junction	164200
7q11.2-q21.3	*EEC1*	Unknown	AD	Ectrodactyly, ED, cleft lip/palate syndrome 1 (EEC1)	Unknown	129900
11q23-q24	*PVRL1*	Nectin 1	AR	Cleft lip/palate-ED syndrome (CLPED1)	Tight junction cellular membrane stability	225060
			AR	Rosselli-Gulienetti syndrome		225000
12q13	*KRT6A*	Keratins 6A and 6B	AD	Pachyonychia congenita 1 and 2	Structural component of hair and nails	167200
	KRT6B				Structural component of hair and nails	167210

12q13	*KRTHBS*	Keratin 85	AD	ED, 'pure' hair-nail type	Structural component of hair and nails	602032
13qll-q12	*GJB2*	Connexin 26	AD	Palmoplantar keratoderma, with deafness	Connexin protein, intercellular junction	148350
			AD	Keratitis-ichthyosis·deafness syndrome, AD (KID, AD)		148210
			AD	Ichthyosis, hystrix-like, with deafness (HID syndrome)		602540
13q12	*GJB6*	Connexin 30	AD	Clouston syndrome	Connexin protein, intercellular junction	129500
14q13	*IKBA*	IκBα	AD	Hypohidrotic ED with immune deficiency	NFκB cytoplasmic inhibitor	164008
16q22.1	*CDH3*	Cadherin·3	AR	ED, ectrodactyly, and macular dystrophy (EEM)	Adhesion molecule cell-cell binding function	225280
17q12-q21	*KRT14*	Keratin 14	AD	Naegeli-Franceschetti-Jadassohn syndrome	Structural component of hair and nails	161000
17q12-q21	*KRT16*	Keratins 16 and 17	AD	Pachyonychia congenita 1 and 2	Structural component of hair and nails	167200,
	KRT17				Structural component of hair and nails	167210
17q21.3-q22	*DLX3*	Homeobox protein DLX-3	AD	Trichodentoosseous syndrome	Transcription factor	190320
19	*EEC2*	Unknown	AD	Ectrodactyly, ED, cleft lip/palate syndrome 1 (EEC2)	Unknown	