Ectodermal Dysplasias With Identified Genes and Genetic Testing Available

The following ectodermal dysplasias have identified genes and have genetic tests. This information was taken from the Online Mendelian Inheritance in Man in August of 2015.

Acto-dermato-ungual-lacrimal-tooth (ADULT) syndrome

Ankyloblepharon-ectodermal defects-cleft lip/palate (AEC) syndrome

Autoimmune polyendocrine syndrome

Basan syndrome*

Cardiofaciocutaneous syndrome

Cartilage-hair hypoplasia

Cleft lip/palate-ectodermal dysplasia syndrome

Clouston syndrome

Cranioectodermal dysplasia 1

Dyskeratosis congenita, autosomal dominant, 1

Dyskeratosis congenita, autosomal recessive, 1

Dyskeratosis congenita

Ectrodactyly-ectodermal dysplasia- clefting (EEC)

Ectodermal dysplasia, anhidrotic, with immunodeficiency, osteopetrosis, and lymphedema

Ectodermal dysplasia, ectrodactyly, and macular dystrophy syndrome

X-linked hypohidrotic ectodermal dysplasia (XLHED or ectodermal dysplasia 1, hypohidrotic, x-linked)

Autosomal dominant hypohidrotic ectodermal dysplasia (ADHED or ectodermal dysplasia 10a, hypohidrotic/hair/nail type, autosomal dominant)

Autosomal recessive hypohidrotic ectodermal dysplasia (ARHED or ectodermal dysplasia 10b, hypohidrotic/hair/tooth type, autosomal recessive)

Hypohidrotic ectodermal dysplasia with immune deficiency (HED-ID or ectodermal dysplasia, hypohidrotic, with immune deficiency

Ectodermal dysplasia/skin fragility syndrome

Goltz syndrome (focal dermal hypoplasia)

Focal facial dermal dysplasia 3

Focal facial dermal dysplasia 4

Gapo syndrome

Hypertrichosis, congenital generalized, with or without gingival hyperplasia (135400)*

Hypotrichosis, congenital, with juvenile macular dystrophy

IFAP syndrome with or without bresheck syndrome

Incontinentia pigmenti

Insensitivity to pain, congenital, with anhidrosis

Johanson-blizzard syndrome

Keratitis-ichthyosis-deafness (KID) syndrome, autosomal dominant

Kohlschutter-tonz syndrome

Limb-mammary syndrome

Marshall syndrome

Monilethrix

Naegeli syndrome

Oculodentodigital dysplasia (ODD) syndrome

Odonoonychodermal dysplasia

Orofaciodigital syndrome I

Pachyonychia congenita 1

Pachyonychia congenita 2

Poikiloderma with neutropenia

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Rothmund-thomson syndrome
Saethre-chotzen syndrome
Scalp-ear-nipple syndrome
Schinzel-giedion midface retraction syndrome
Schopf-schulz-passarge syndrome
Trichodentoosseous (TDO) syndrome
Trichorhinophalangeal syndrome, type 1 (TRPS1)
Trichorhinophalangeal syndrome, type 2 (TRPS2)
Trichorhinophalangeal syndrome, type 3
Trichothiodystrophy 1, photosensitive
Trichothiodystrophy 2, photosensitive
Trichothiodystrophy 3, photosensitive
Trichothiodystrophy 4, nonphotosensitive
Trichothiodystrophy 5, nonphotosensitive
Ulnar-mammary syndrome
Weyers acrofacial dysostosis
Witkop syndrome