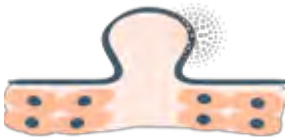
Ectodermal Dysplasia

Precision Panel



Overview

Ectodermal Dysplasias (EDs) are a heterogeneous group of approximately 200 inherited disorders characterized by anomalies in at least two of the structures derived from embryonic ectoderm, with at least one involving skin appendages (hair, nails, sweat glands). Other tissues that can be involved include mammary glands, adrenal medulla, central nervous system, inner ear etc. Eds are congenital, diffuse and nonprogressive. Ectodermal dysplasias caused by genetic mutations include hypohidrotic ED, hypohidrotic ED with immune deficiency and hidrotic ED, the most common being X-linked recessive hypohidrotic ectoderma dysplasia, also known as Christ-Siemens-Touraine syndrome. Morbidity and mortality is related to the absence or dysfunction of eccrine and mucous glands. Beyond early childhood, life expectancy ranges from normal to slightly reduced.

The Igenomix Ectodermal Dysplasias Precision Panel can be used to make an accurate and directed diagnosis ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Ectodermal Dysplasias Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Hyperthermia with fever and seizures
- Decreased tears (xerophthalmia) and conjunctivitis
- Deficient hearing or vision
- Decreased saliva (xerostomia) and frequent dental caries
- Developmental delay
- Dysphagia
- Growth failure
- Frequent upper respiratory tract infections

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.

- Early initiation of multidisciplinary treatment in the form of medical care encouraging protective hydrating routines, dental evaluation and antibiotic treatment to prevent complications. Surgical care may be needed.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
BCS1L	Bjornstad Syndrome, Gracile Syndrome, Leigh Syndrome, Mitochondrial Complex Iii Deficiency	AR,MI	99.96	40 of 42
CDH3	Eem Syndrome, Hypotrichosis, Congenital, Macular Degeneration	AR	95	34 of 36
DLX3	Amelogenesis Imperfecta, Trichodentoosseous Syndrome	AD	100	10 of 10
DSP	Right Ventricular Dysplasia, Cardiomyopathy, Woolly Hair, Palmoplantar Keratoderma, Toothagenesis, Epidermolysis Bullosa, Carvajal Syndrome, Pulmonary Fibrosis	AD,AR	99.91	366 of 369
EDA	Ectodermal Dysplasia, Tooth Agenesis, Oligodontia	X,XR,XD,G	98.99	-
EDAR	Ectodermal Dysplasia,	AD,AR	100	69 of 69
EDARADD	Ectodermal Dysplasia, Oligodontia	AD,AR	100	10 of 10
ERCC2	Cerebrooculofacioskeletal Syndrome, Trichothiodystrophy, Xeroderma Pigmentosum, Cofs Syndrome	AR	100	102 of 102
EVC	Ellis-Van Creveld Syndrome, Weyers Acrofacial Dysostosis, Acrofacial Dysostosis	AD,AR	94.04	68 of 73
EVC2	Ellis-Van Creveld Syndrome, Weyers Acrofacial Dysostosis, Acrofacial Dysostosis	AD,AR	99.98	75 of 75
GJB2	Deafness, Ichthyosis, Palmoplantar Keratoderma, Knuckle Pads, Kid Syndrome	AD,AR,X,XR,MU,D,G	99.89	413 of 419
GJB6	Deafness, Ectodermal Dysplasia, Kid Syndrome	AD,AR,X,XR,MU,D,G	99.89	28 of 28
GRHL2	Corneal Dystrophy, Deafness, Ectodermal Dysplasia	AD,AR	100	8 of 11
HOXC13	Ectodermal Dysplasia	AR	99.69	7 of 7
HR	Alopecia Universalis, Atrichia, Marie Unna Hypotrichosis	AD,AR	99.94	52 of 52
IFT122	Cranioectodermal Dysplasia	AR	99.83	22 of 22
JUP	Right Ventricular Dysplasia, Naxos Disease, Lethal Acantholytic Epidermolysis Bullosa	AD,AR	100	56 of 56
KDF1	Ectodermal Dysplasia	AD	-	-
KREMEN1	Ectodermal Dysplasia	AR	93.48	5 of 5
KRT74	Ectodermal Dysplasia, Hypotrichosis, Woolly Hair	AD,AR	100	4 of 4
KRT85	Ectodermal Dysplasia	AR	100	6 of 6
LRP6	Coronary Artery Disease, Tooth Agenesis, Oligodontia	AD	100	44 of 44
LTBP3	Geleophysic Dysplasia, Platyspondyly, Amelogenesis Imperfecta, Acromicric Dysplasia	AD,AR	97.67	22 of 23
MBTPS2	Ichthyosis Follicularis, Atrichia, Photophobia, Keratosis Follicularis Spinulosa Decalvans, Osteogenesis Imperfecta, Palmoplantar Keratoderma, Bresek Syndrome	X,XR,G	100	-
MPLKIP	Trichothiodystrophy	AR	100	13 of 13
MSX1	Tooth Agenesis, Witkop Syndrome, Cleft Lip/Palate, Hypodontia, Oligodontia	AD	99.54	50 of 51
NECTIN1	Cleft Lip/Palate, Ectodermal Dysplasia	AR	100	-
NECTIN4	Ectodermal Dysplasia, Syndactyly	AR	100	-
NFKBIA	Ectodermal Dysplasia, T-Cell Immunodeficiency	AD	99.98	13 of 13
NLRP1	Autoinflammation, Arthritis, Dyskeratosis, Corneal Intraepithelial Dyskeratosis, Ectodermal Dysplasia, Respiratory Papillomatosis	AD,AR,MU,P	99.37	15 of 15

ORAI1	Immunodeficiency, Myopathy, Stormorken-Sjaastad-Langslet Syndrome	AD,AR	91.93	20 of 22
PAX9	Tooth Agenesis, Oligodontia	AD	100	53 of 55
PKP1	Ectodermal Dysplasia, Skin Fragility Syndrome, Epidermolysis Bullosa	AR	100	18 of 18
PORCN	Focal Dermal Hypoplasia	X,XD,G	100	-
PRKD1	Congenital Heart Defects, Ectodermal Dysplasia	AD	97.39	8 of 9
RMRP	Anauxetic Dysplasia, Cartilage-Hair Hypoplasia, Metaphyseal Dysplasia, Omenn Syndrome	AR	-	-
TP63	Adult Syndrome, Ankyloblepharon, Cleft Lip/Palate, Ectrodactyly, Ectodermal Dysplasia, Limb-Mammary Syndrome, Rapp-Hodgkin Syndrome, Split-Hand And Foot Malformation, Bladder Exstrophy, Eec Syndrome	AD	99.98	144 of 144
TWIST2	Ablepharon-Macrostomia Syndrome, Barber-Say Syndrome, Focal Facial Dermal Dysplasia	AD,AR	99.82	9 of 9
WDR35	Cranioectodermal Dysplasia, Short-Rib Thoracic Dysplasia, Polydactyly, Short Rib-Polydactyly Syndrome	AR	100	31 of 33
WNT10A	Odontoonychodermal Dysplasia, Schopf-Schulz-Passarge Syndrome, Tooth Agenesis, Oligodontia	AD,AR	99.91	90 of 90

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology

Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

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