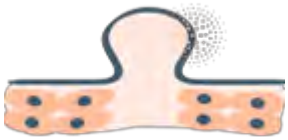
Cutis Laxa

Precision Panel



Overview

Cutis Laxa (CL), also known as elastolysis, is an inherited or acquired group of connective tissue disorders characterized by inelastic skin that hangs loosely in folds. Since the connective tissue is the tissue that helps body growth as well as serving as a scaffold for cells and organs, the clinical presentation and mode of inheritance is heterogeneous. Clinically, multiple organ systems are involved, leading to a severe, lethal multisystem disorder. Both acquired and inherited forms exist, the latter being inherited in an autosomal dominant, recessive and X-linked recessive patterns. Inborn errors of metabolism have been associated.

The Igenomix Cutis Laxa Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of connective tissue disorders due to their overlapping phenotypic features 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 Cutis Laxa Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Early appearing loose, wrinkled skin, hanging in folds most prominent around the eyes, face, neck, shoulders and thighs
- Osteoporosis
- Gastrointestinal manifestations: diverticula of small and large bowel
- Pulmonary findings: bronchiectasis, emphysema
- Cardiovascular findings: cardiomegaly, congestive heart failure, murmurs, aortic aneurysms

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 treatment with a multidisciplinary team in the form of medical treatment and surveillance to prevent complications and/or surgical care or redundant skin folds and other organic manifestations.

- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation due to overlapping features of connective tissue disorders.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCC6	Arterial Calcification, Pseudoxanthoma Elasticum	AD,AR	99	346 of 349
ADAMTS2	Ehlers-Danlos Syndrome	AR	95.99	9 of 10
AEBP1	Ehlers-Danlos Syndrome	AR	99.35	9 of 9
ALDH18A1	Cutis Laxa, Spastic Paraplegia, De Barsy Syndrome	AD,AR	100	39 of 40
ANTXR1	Gapo Syndrome, Hemangioma	AD,AR	100	19 of 19
ATP6AP2	Congenital Disorder Of Glycosylation, Parkinsonism-Spasticity Syndrome	X,XR,G	100	-
ATP6V0A2	Cutis Laxa, Wrinkly Skin Syndrome	AR	99.99	55 of 55
ATP6V1A	Cutis Laxa, Epileptic Encephalopathy	AD,AR	99.98	9 of 9
ATP6V1E1	Cutis Laxa	AR	100	2 of 2
ATP7A	Cutis Laxa, Menkes Disease, Spinal Muscular Atrophy, Occipital Horn Syndrome	X,XR,G	99.83	-
B3GALT6	Ehlers-Danlos Syndrome, Spondyloepimetaphyseal Dysplasia With Joint Laxity	AR	65.09	24 of 39
B3GAT3	Multiple Joint Dislocations, Craniofacial Dysmorphism With Or Without Congenital Heart Defects	AR	99.86	15 of 15
B4GALT7	Ehlers-Danlos Syndrome	AR	99.92	11 of 11
BAZ1B	Williams Syndrome	-	99.05	5 of 5
BCL11B	Immunodeficiency, Intellectual Developmental Disorder With Speech Delay, Dysmorphic Facies, And T-Cell Abnormalities	AD	96.06	12 of 12
BRAF	Cardiofaciocutaneous Syndrome, Leopard Syndrome, Noonan Syndrome, Craniopharyngioma	AD	100	80 of 80
C1R	Ehlers-Danlos Syndrome	AD	98.89	16 of 16
CD96	C Syndrome	AD	100	4 of 4
CEP55	Multinucleated Neurons, Anhydramnios, Renal Dysplasia, Cerebellar Hypoplasia, Hydranencephaly, Meckel Syndrome	AR	99.22	3 of 3
CHST14	Ehlers-Danlos Syndrome	AR	97.7	21 of 22
CHST3	Multiple Joint Dislocations, Craniofacial Dysmorphism With Or Without Congenital Heart Defects, Spondyloepiphyseal Dysplasia, Skeletal Dysplasia	AR	99.97	38 of 38
CLIP2	Williams Syndrome	-	99.99	1 of 1
COL3A1	Ehlers-Danlos Syndrome, Polymicrogyria, Acrogeria, Cerebral Saccular Aneurysm	AD,AR	100	676 of 676
CSPP1	Joubert Syndrome, Meckel Syndrome	AR	98.32	29 of 30
DSE	Ehlers-Danlos Syndrome	AR	99.94	3 of 3
EED	Cohen-Gibson Syndrome, Weaver Syndrome	AD	99.92	10 of 10
EFEMP2	Cutis Laxa	AR	99.99	17 of 17
ELN	Cutis Laxa, Supravalvular Aortic Stenosis, Williams-Beuren Syndrome, Familial Thoracic Aortic Aneurysm And Aortic Dissection	AD	99.99	95 of 96
EXT1	Chondrosarcoma, Exostoses, Multiple Osteochondromas, Trichorhinophalangeal Syndrome	AD,AR	99.97	518 of 525
EZH2	Weaver Syndrome	AD	99.82	40 of 41
FBLN5	Cutis Laxa, Neuropathy, Macular Degeneration	AD,AR	97.43	23 of 23
FBN1	Acromicric Dysplasia, Ectopia Lentis, Geleophysic Dysplasia, Marfan Syndrome, Mass Syndrome, Stiff Skin Syndrome, Weill-Marchesani	AD	100	2836 of 2845

	Syndrome, Thoracic Aortic Aneurysm And Aortic Dissection, Shprintzen-Goldberg Syndrome			
FGF20	Renal Hypodysplasia, Renal Agenesis	AR	99.76	2 of 2
FGFR2	Antley-Bixler Syndrome, Apert Syndrome, Bent Bone Dysplasia Syndrome, Crouzon Syndrome, Cutis Gyrata Syndrome, Scaphocephaly Syndrome, Jackson-Weiss Syndrome, Lacrimoauriculodentodigital Syndrome, Pfeiffer Syndrome, Saethre-Chotzen Syndrome	AD	98	140 of 143
FGFR3	Achondroplasia, Crouzon Syndrome With Acanthosis Nigricans, Epidermal Nevus, Hypochondroplasia, Lacrimoauriculodentodigital Syndrome, Muenke Syndrome, Thanatophoric Dysplasia, Camptodactyly-Tall Stature-Scoliosis-Hearing Loss Syndrome, Brachycephaly, Plagiocephaly, Saethre-Chotzen Syndrome	AD,AR	99.89	77 of 78
FIG4	Amyotrophic Lateral Sclerosis, Charcot-Marie-Tooth Disease, Cleidocranial Dysplasia With Micrognathia, Polymicrogyria, Bilateral Parasagittal Parieto-Occipital Polymicrogyria, Yunis-Varon Syndrome	AD,AR	99.92	72 of 72
FLNA	Cardiac Valvular Dysplasia, Fg Syndrome, Frontometaphyseal Dysplasia, Heterotopia, Intestinal Pseudoobstruction, Melnick-Needles Syndrome, Otopalatodigital Syndrome, Terminal Osseous Dysplasia, Short Bowel Syndrome, Ehlers-Danlos Syndrome	X,XR,XD,G	100	-
FOXC1	Axenfeld-Rieger Syndrome, Iridogoniodysgenesis, Peters Anomaly	AD	88.98	94 of 100
GGCX	Pseudoxanthoma Elasticum-Like Disorder With Retinitis Pigmentosa, Vitamin K-Dependent Clotting Factors, Body Skin Hyperlaxity	AR	100	62 of 62
GORAB	Geroderma Osteodysplastica	AR	96	17 of 18
GPX4	Spondylometaphyseal Dysplasia	AR	79.72	3 of 3
GSN	Amyloidosis	AD	96.69	16 of 17
GTF2I	Williams Syndrome	-	63.79	-
GTF2IRD1	Williams Syndrome	-	99.98	1 of 1
HPGD	Clubbing Of Digits, Hypertrophic Osteoarthropathy, Cranio-Osteoarthropathy, Pachydermoperiostosis	AR	100	17 of 17
HRAS	Costello Syndrome, Epidermal Nevus, Giant Pigmented Hairy Nevus, Schimmelpenning-Feuerstein-Mims Syndrome, Linear Nevus Sebaceus Syndrome	AD	100	34 of 34
IFT43	Cranioectodermal Dysplasia, Retinitis Pigmentosa, Short-Rib Thoracic Dysplasia With Polydactyly	AR	100	6 of 6
KIAA0586	Joubert Syndrome, Short-Rib Thoracic Dysplasia	AR	99.84	31 of 32
KRAS	Aplasia Cutis Congenita With Epibulbar Dermoids, Arteriovenous Malformation Of The Brain, Cardiofaciocutaneous Syndrome, Leukemia, Noonan Syndrome, Autoimmune Lymphoproliferative Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Encephalocraniocutaneous Lipomatosis, Linear Nevus Sebaceus Syndrome, Lynch Syndrome, Toriello-Lacassie-Droste Syndrome	AD	100	38 of 38
LIMK1	Williams Syndrome		100	2 of 2
LTBP4	Cutis Laxa, Duchenne Muscular Dystrophy	AR	97.45	27 of 27
MAN1B1	Non-Syndromic Intellectual Disability	AR	99.97	29 of 30
MAP2K1	Cardiofaciocutaneous Syndrome, Melorheostosis, Noonan Syndrome	AD	100	31 of 31
MAP2K2	Cardiofaciocutaneous Syndrome, Noonan Syndrome	AD	100	37 of 37
MEGF8	Carpenter Syndrome	AR	98.97	22 of 22
MLXIPL	Williams-Beuren Syndrome	AD	99.42	-
MRPS16	Oxidative Phosphorylation Deficiency	AR	100	1 of 1
MRPS22	Combined Oxidative Phosphorylation Deficiency, Ovarian Dysgenesis, XX Gonadal Dysgenesis	AR	100	10 of 10
NAA10	Microphthalmia, Ogden Syndrome	X,XR,XD,G	99.86	-
NBAS	Infantile Liver Failure Syndrome, Short Stature, Optic Nerve Atrophy, Pelger-Huet Anomaly	AR	99.98	60 of 61
NDUFB11	Linear Skin Defects, Microphthalmia, Complex I Deficiency	X,XD,G	97.48	-
NEPRO	Anauxetic Dysplasia	AR	-	-

NPR2	Acromesomelic Dysplasia, Epiphyseal Chondrodysplasia, Short Stature With Nonspecific Skeletal Abnormalities	AD,AR	100	81 of 81
NSD1	Sotos Syndrome, Weaver Syndrome	AD	99.8	451 of 459
OSMR	Amyloidosis	AD	100	14 of 14
PEX1	Peroxisome Biogenesis Disorder, Zellweger Syndrome, Deafness-Enamel Hypoplasia-Nail Defects Syndrome, Refsum Disease, Arenoleukodystrophy	AR	97.02	126 of 134
PITX2	Iridogoniodysgenesis, Ring Dermoid Of Cornea, Axenfeld-Rieger Syndrome, Peters Anomaly	AD	99.97	104 of 107
PLOD1	Ehlers-Danlos Syndrome	AR	100	36 of 36
PTDSS1	Lenz-Majewski Hyperostotic Dwarfism	AD	100	7 of 7
PYCR1	Cutis Laxa, Geroderma Osteodysplastica	AR	100	44 of 44
RFC2	Williams Syndrome	-	100	3 of 3
RIN2	Macrocephaly, Alopecia, Cutis Laxa, Scoliosis, Rin2 Syndrome	AR	99.6	4 of 4
RIT1	Noonan Syndrome	AD	99.85	27 of 27
RPS6KA3	Coffin-Lowry Syndrome	X,XD,G	99.95	-
SLC25A24	Fontaine Progeroid Syndrome, Gorlin-Chaudhry-Moss Syndrome	AD	99.59	2 of 2
SLC2A10	Arterial Tortuosity Syndrome	AR	100	35 of 35
SLC6A8	Creatine Deficiency Syndrome	X,XR,G	99.87	-
SLC7A7	Lysinuric Protein Intolerance	AR	100	61 of 61
SPINT2	Diarrhea With Or Without Other Congenitalanomalies	AR	100	14 of 14
SRD5A3	Congenital Disorder Of Glycosylation, Kahrizi Syndrome	AR	100	15 of 15
SUZ12	Imagawa-Matsumoto Syndrome, Weaver Syndrome	AD	98.82	3 of 3
TBL2	Williams Syndrome	-	96.14	-
TBX15	Pelviscapular Dysplasia	AR	100	3 of 3
TRPS1	Trichorhinophalangeal Syndrome	AD	99.45	108 of 112
TWIST2	Ablepharon-Macrostomia Syndrome, Barber-Say Syndrome, Focal Facial Dermal Dysplasia	AD,AR	99.82	9 of 9
VAC14	Striatonigral Degeneration, Yunis-Varon Syndrome	AR	100	11 of 11
WDR19	Asphyxiating Thoracic Dystrophy, Cranioectodermal Dysplasia, Nephronophthisis, Senior-Loken Syndrome, Jeune Syndrome	AR	99.96	47 of 49
WDR35	Cranioectodermal Dysplasia, Short-Rib Thoracic Dysplasia With Or Without Polydactyly	AR	100	31 of 33
WDR37	Neurooculocardiogenitourinary Syndrome	AD	100	5 of 5
XYLT1	Desbuquois Syndrome, Pseudoxanthoma Elasticum	AR	92.61	19 of 23
XYLT2	Pseudoxanthoma Elasticum, Spondylo-Ocular Syndrome	AR	99.98	12 of 12
ZNF469	Brittle Cornea Syndrome	AR	99.91	79 of 79

*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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