Stickler Syndrome

Precision Panel

Overview

Stickler Syndrome (SS), also known as hereditary arthroophthalmopathy belongs to the group of connective tissue disorders together with Marshall syndrome, and so have overlapping characteristics. It is caused by mutations of genes in charge of the assembly of collagen. Since collagen is a major component of cartilage, vitreous and nucleus pulposus the clinical manifestations will affect these structures. Affected individuals are at significantly increased risk for retinal detachment and blindness, and early detection and diagnosis are critical in improving visual outcomes of these patients. The mode of inheritance varies from autosomal dominant, recessive and X-linked.

The Igenomix Stickler Syndrome Precision Panel can be used to make a directed and accurate 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 Stickler Syndrome Precision Panel is indicated for those patients with a clinical diagnosis or suspicion with or without the following manifestations:

- Orofacial abnormalities: midfacial underdevelopment and cleft palate
- Ophthalmologic abnormalities: myopia, cataract, retinal detachment
- Hearing loss
- Precocious arthritis

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 surgical repair of orofacial abnormalities, retinal detachment, hearing and visual aids and symptomatic medical treatment for arthropathy. Early and continuous ophthalmologic examination follow-up to prevent further complications.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

- Improvement of delineation of genotype-phenotype correlation.

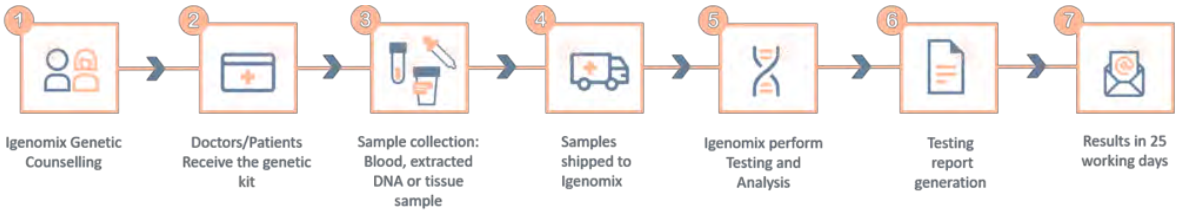
Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ACTA2	Aortic Aneurysm, Moyamoya Disease, Multisystemic Smooth Muscle Dysfunction Syndrome	AD	100	88 of 88
ADAMTS2	Ehlers-Danlos Syndrome	AR	95.99	9 of 10
AEBP1	Ehlers-Danlos Syndrome	AR	99.35	9 of 9
ALDH18A1	Cutis Laxa, Corneal Clouding, Mental Retardation, Spastic Paraplegia, De Barsy Syndrome	AD,AR	100	39 of 40
ATP6V0A2	Cutis Laxa, Wrinkly Skin Syndrome	AR	99.99	55 of 55
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, Joint Laxity	AR	65.09	24 of 39
B3GAT3	Multiple Joint Dislocations, Short Stature, Craniofacial Dysmorphism	AR	99.86	15 of 15
B4GALT7	Ehlers-Danlos Syndrome	AR	99.92	11 of 11
BGN	Meester-Loeys Syndrome, Spondyloepimetaphyseal Dysplasia	X,XR,G	99.87	-
CBS	Homocystinuria	AR	99.98	192 of 194
CHST14	Ehlers-Danlos Syndrome	AR	97.7	21 of 22
COL11A1	Deafness, Fibrochondrogenesis, Marshall Syndrome, Stickler Syndrome, Myopia, Midfacial Retrusion, Sensorineural Hearing Loss, Rhizomelic Dysplasia	AD,AR	100	104 of 106
COL11A2	Deafness, Fibrochondrogenesis, Otospondylomegaepiphyseal Dysplasia, Stickler Syndrome	AD,AR	99.98	58 of 58
COL12A1	Bethlem Myopathy, Ullrich Congenital Muscular Dystrophy, Ehlers-Danlos Syndrome	AD	99.97	18 of 19
COL1A1	Caffey Disease, Ehlers-Danlos Syndrome, Osteogenesis Imperfecta, Osteoporosis, Dermatofibrosarcoma Protuberans	AD	99.98	1156 of 1159
COL1A2	Ehlers-Danlos Syndrome, Osteogenesis Imperfecta, Osteoporosis	AD,AR	100	576 of 581
COL2A1	Achondrogenesis, Avascular Necrosis Of Femoral Head, Czech Dysplasia, Epiphyseal Dysplasia, Kniest Dysplasia, Legg-Calve-Perthes Disease, Osteoarthritis, Stickler Syndrome, Dysspondyloenchondromatosis, Platyspondylic Dysplasia, Spondyloepimetaphyseal Dysplasia	AD,MU	100	583 of 583
COL3A1	Ehlers-Danlos Syndrome, Polymicrogyria, Acrogeria, Cerebral Saccular Aneurysm	AD,AR	100	676 of 676
COL4A1	Angiopathy, Nephropathy, Aneurysms, Microangiopathy, Leukoencephalopathy, Porencephaly, Hanac Syndrome, Walker-Warburg Syndrome	AD	99.99	173 of 173
COL5A1	Ehlers-Danlos Syndrome	AD	99.08	191 of 195
COL5A2	Ehlers-Danlos Syndrome	AD	100	45 of 45
COL9A1	Epiphyseal Dysplasia, Stickler Syndrome	AD,AR	99.98	8 of 8
COL9A2	Epiphyseal Dysplasia, Stickler Syndrome	AD,AR	100	16 of 16
COL9A3	Epiphyseal Dysplasia, Stickler Syndrome	AD	99.98	20 of 20
DSE	Ehlers-Danlos Syndrome	AR	99.94	3 of 3
EFEMP2	Cutis Laxa	AR	99.99	17 of 17
ELN	Cutis Laxa, Supravalvular Aortic Stenosis, Williams-Beuren Syndrome, Thoracic Aortic Aneurysm	AD	99.99	95 of 96
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 Syndrome, Thoracic Aortic Aneurysm, Shprintzen-Goldberg Syndrome	AD	100	2836 of 2845

FBN2	Contractural Arachnodactyly, Macular Degeneration	AD	100	115 of 115
FKBP14	Ehlers-Danlos Syndrome, Myopathy, Hearing Loss	AR	99.98	7 of 8
FLNA	Cardiac Valvular Dysplasia, Fg Syndrome, Frontometaphyseal Dysplasia, Heterotopia, Intestinal Pseudoobstruction, Melnick-Needles Syndrome, Otopalatodigital Syndrome, Terminal Osseous Dysplasia, Congenital Short Bowel Syndrome, Ehlers-Danlos Syndrome	X,XR,XD,G	100	-
LOX	Aortic Aneurysm	AD	95.47	8 of 8
LOXL3	Stickler Syndrome	-	99.97	7 of 7
LRP2	Donnai-Barrow Syndrome	AR	99.99	58 of 58
LTBP4	Cutis Laxa, Duchenne Muscular Dystrophy	AR	97.45	27 of 27
MAT2A	Thoracic Aortic Aneurysm, Aortic Dissection	-	100	3 of 3
MED12	Lujan-Fryns Syndrome, Ohdo Syndrome, Opitz-Kaveggia Syndrome, Blepharophimosis, Intellectual Disability Syndrome, Fg Syndrome	X,XR,G	100	-
MFAP5	Thoracic Aortic Aneurysm, Aortic Dissection	AD	100	3 of 3
MYH11	Thoracic Aortic Aneurysm, Aortic Dissection, Megacystis, Microcolon, Intestinal Hypoperistalsis	AD	100	67 of 67
MYLK	Thoracic Aortic Aneurysm, Aortic Dissection, Megacystis, Microcolon, Intestinal Hypoperistalsis	AD	99.95	50 of 50
NOTCH1	Adams-Oliver Syndrome, Aortic Valve Disease	AD	99.83	178 of 179
PLOD1	Ehlers-Danlos Syndrome	AR	100	36 of 36
PRDM5	Brittle Cornea Syndrome	AR	99.86	13 of 13
PRKG1	Thoracic Aortic Aneurysm, Aortic Dissection	AD	99.93	6 of 6
PYCR1	Cutis Laxa, Geroderma Osteodysplastica	AR	100	44 of 44
RIN2	Macrocephaly, Alopecia, Cutis Laxa, Scoliosis, Rin2 Syndrome	AR	99.6	4 of 4
SKI	1p36 Deletion Syndrome, Shprintzen-Goldberg Syndrome	AD	99.66	39 of 39
SLC2A10	Arterial Tortuosity Syndrome	AR	100	35 of 35
SLC39A13	Ehlers-Danlos Syndrome	AR	100	9 of 9
SMAD2	Osteopoikilosis	-	100	19 of 19
SMAD3	Loeys-Dietz Syndrome, Aneurysm-Osteoarthritis Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection	AD	100	128 of 128
SMAD4	Polyposis Syndrome, Hemorrhagic Telangiectasia, Myhre Syndrome, Pancreatic Cancer, Thoracic Aortic Aneurysm, Aortic Dissection	AD	99.56	136 of 136
TAB2	Congenital Heart Defects, Polyvalvular Heart Disease Syndrome	AD	99	13 of 13
TGFB2	Loeys-Dietz Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection	AD	99.9	41 of 44
TGFB3	Right Ventricular Dysplasia, Loeys-Dietz Syndrome, Thoracic Aortic Aneurysm, Aortic Dissection	AD	100	34 of 35
TGFBR1	Loeys-Dietz Syndrome, Self-Healing Squamous Epithelioma, Thoracic Aortic Aneurysm, Aortic Dissection	AD	94	96 of 100
TGFBR2	Colorectal Cancer, Esophageal Cancer, Loeys-Dietz Syndrome, Familial Thoracic Aortic Aneurysm, Aortic Dissection, Lynch Syndrome	AD	99.9	165 of 166
TNXB	Vesicoureteral Reflux, Ehlers-Danlos Syndrome	AD,AR	92.75	29 of 33
VCAN	Wagner Syndrome	AD	99.91	11 of 21
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



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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.

References

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