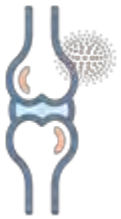
Osteogenesis Imperfecta

Precision Panel



Overview

Osteogenesis Imperfecta (OI) is a disorder of bone fragility caused generally by mutations in the COL1A1 and COL1A2 genes that encode type I collagen. OI is one of the most common skeletal dysplasias. It is a generalized disease that is phenotypically and molecularly heterogeneous manifesting with a broad array of signs and symptoms including connective tissue and systemic manifestations in addition to bone fragility. The more prevalent autosomal dominant forms of osteogenesis imperfecta are caused by primary defects in type 1 collagen, whereas autosomal recessive forms are caused by deficiency of proteins which interact with type 1 procollagen. There are at least 8 different types of the disease based on the inheritance. The differential diagnosis of OI includes child abuse, rickets, osteomalacia and other rare skeletal syndromes.

The Igenomix Osteogenesis Imperfecta Precision Panel can be used to make a directed and accurate differential diagnosis of bone fragility 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 Osteogenesis Imperfecta Precision Panel is indicated for those patients with a suspected clinical diagnosis of osteogenesis imperfecta presenting with the following manifestations:

- Blue sclerae
- Triangular facies
- Macrocephaly
- Hearing loss
- Defective dentition
- Barrel chest
- Scoliosis
- Limb deformities
- Pregnancy ultrasound: Limb-length abnormalities at 15-18 weeks' gestation, decreased mineralization of calvaria, bowing of the long bones, multiple rib fractures
- Fractures
- Joint laxity
- Growth retardation

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, encompassing physical rehabilitation and surgical procedures, management of hearing, dental and pulmonary abnormalities, as well as drugs, such as bisphosphonates and recombinant human growth hormone.
- Prenatal detection of osteogenesis imperfecta for a directed obstetric and perinatal treatment of affected infants.
- Combining phenotypic and genotypic data to improve diagnostic rate of these patients in the target population.
- Risk assessment of asymptomatic family members according to the mode of inheritance.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ALPL	Adult Hypophosphatasia, Childhood Hypophosphatasia	AD,AR	100%	320 of 321
ARCN1	Rhizomelic Short Stature With Microcephaly, Micrognathia, And Developmental Delay	AD	99.91%	4 of 4
B3GAT3	Multiple Joint Dislocations, Short Stature, Craniofacial Dysmorphism With Or Without Congenital Heart Defects	AR	99.86%	15 of 15
B4GALT7	B4GALT7-Related Spondylodysplastic Ehlers-Danlos Syndrome	AR	99.92%	11 of 11
BMP1	Osteogenesis Imperfecta Type XIII	AR	99.98%	27 of 28
CLCN5	Dent Disease, X-linked Recessive Hypophosphatemic Rickets, Nephrolithiasis With Renal Failure, Low Molecular Weight Proteinuria With Hypercalciuria And Nephrocalcinosis	X,XR,G	99.39%	NA of NA
COL1A1	Arthrochalasia, Ehlers-Danlos Syndrome, Caffey Disease, Dermatofibrosarcoma Protuberans, Osteogenesis Imperfecta Type I, IIA, III, IV	AD	99.98%	1156 of 1159
COL1A2	Arthrochalasia Ehlers-Danlos Syndrome, Cardiac-Valvular Ehlers-danlos Syndrome, Osteogenesis Imperfecta Type IIA, III, IV, Osteoporosis	AD,AR	100%	576 of 581
CREB3L1	Osteogenesis Imperfecta Type XVI	AR	99.98%	4 of 4
CRTAP	Osteogenesis Imperfecta Type VII	AR	99.98%	29 of 30
FGF23	Autosomal Dominant Hypophosphatemic Rickets, Familial Hyperphosphatemic Tumoral Calcinosis	AD,AR	100%	21 of 21
FKBP10	Bruck Syndrome, Kuskokwim Syndrome, Osteogenesis Imperfecta Type XI	AR	100%	51 of 51
IFITM5	Osteogenesis Imperfecta, Type V	AD	100%	4 of 4
KDELR2	Congenital Sucrase-Isomaltase Deficiency		75.90%	NA of NA
LRP5	Autosomal Dominant Endosteal Hyperostosis, Exudative Vitreoretinopathy, Hyperostosis Corticalis Generalisata, Isolated Polycystic Liver Disease, Autosomal Dominant Osteopetrosis, Osteoporosis-Pseudoglioma Syndrome, Osteosclerosis-Developmental Delay-Craniosynostosis Syndrome, Retinopathy Of Prematurity, Van Buchem Disease Type 2	AD,AR	98.12%	265 of 269
MBTPS2	Bresek Syndrome, Ichthyosis Follicularis-Alopecia-Photophobia Syndrome, Keratosis Follicularis Spinulosa Decalvans, X-linked Mutilating Palmoplantar Keratoderma With Periorificial Keratotic Plaques, Osteogenesis Imperfecta Type XIX, X-linked Mutilating Palmoplantar Keratoderma With Periorificial Keratoticplaques	X,XR,G	100%	NA of NA
MESD	Osteogenesis Imperfecta Type XX	AR	99.89%	NA of NA
P3H1	Osteogenesis Imperfecta Type VIII	AR	94.60%	NA of NA
PHEX	X-linked Dominant Hypophosphatemic Rickets	X,XD,G	99.42%	NA of NA
PLOD2	Bruck Syndrome	AR	99.97%	29 of 29
PLS3	Bone Mineral Density Quantitative Trait Locus	X,XD,G	95.54%	NA of NA

PPIB	Osteogenesis Imperfecta Type IX	AR	100%	14 of 14
SEC24D	Cole-Carpenter Syndrome	AR	99.97%	14 of 14
SERPINF1	Osteogenesis Imperfecta Type VI	AR	99.73%	42 of 46
SERPINH1	Osteogenesis Imperfecta Type X, Preterm Premature Rupture Of The Membranes	AR,MU,P	100%	9 of 10
SGMS2	Calvarial Doughnut Lesions With Bone Fragility	AD	99.93%	3 of 3
SLC34A3	Hereditary Hypophosphatemic Rickets With Hypercalciuria, Hereditary Hypophosphatemic Rickets With Hypercalciuria	AR	100%	52 of 52
SP7	Osteogenesis Imperfecta Type XII	AR	99.98%	3 of 3
SPARC	Osteogenesis Imperfecta Type XVII	AR	100%	4 of 4
TENT5A	Osteogenesis Imperfecta Type XVIII	AR	99.84%	NA of NA
TMEM38B	Osteogenesis Imperfecta Type XIV	AR	99.99%	5 of 6
WNT1	Idiopathic Juvenile Osteoporosis, Osteogenesis Imperfecta Type XV	AR	99.84%	57 of 59

*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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