Panel de tejido conectivo y trastornos relacionados

Nuestro panel de tejido conectivo y trastornos relacionados proporciona una evaluación profunda en un solo paso de varios genes para detectar diferentes trastornos con fenotipos similares, como el síndrome de Marfan, Loeys-Dietz, cutis laxa, Ehlers-Danlos, síndrome de Stickler y aneurisma aórtico torácico familiar y disección.

Nº de genes:	76
Entrega:	25 días
Cobertura:	≥99,5% ≥20x Cobertura media con profundidad ≥ 150 x
Detalles:	Análisis CNV incluido

Síndromes y trastornos comunes cubiertos

Cutis laxa

Síndrome de Ehlers-Danlos

Aneurisma y disección de la aorta torácica familiar

Síndrome de Loeys-Dietz

Síndrome de Marfan

Síndrome de Stickler

ByGENS

Resumen de genes y enfermedades asociadas (OMIM) incluidos en este panel:

Genes	OMIM (Gen)	Enfermedades asociadas (OMIM)	Herencia
ABCC6	603234	Pseudoxanthoma elasticum, forme fruste;Arterial calcification, generalized, of infancy, 2;Pseudoxanthoma elasticum	AD, AR
ACTA2	102620	Aortic aneurysm, familial thoracic 6;Moyamoya disease 5;Multisystemic smooth muscle dysfunction syndrome	AD
ADAMTS2	604539	Ehlers-Danlos syndrome, dermatosparaxis type	AR
ADAMTSL2	612277	Geleophysic dysplasia 1	AR
AEBP1	602981	Ehlers-Danlos syndrome, classic-like, 2	AR
ALDH18A1	138250	Cutis laxa, AD 3;Cutis laxa, AR, type IIIA;Spastic paraplegia 9B, AR;Spastic paraplegia 9A, AD	AD, AR
ATP6V0A2	611716	Cutis laxa, AR, type IIA;Wrinkly skin syndrome	AR
ATP6V1A	607027	Epileptic encephalopathy, infantile or early childhood, 3;Cutis laxa, AR, type IID	AD, AR
ATP6V1E1	108746	Cutis laxa, AR, type IIC	AR

ATP7A	300011	Menkes disease;Occipital horn syndrome;Spinal muscular atrophy, distal, XL 3	XLR
B3GAT3	606374	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects	AR
B4GALT7	604327	Ehlers-Danlos syndrome, spondylodysplastic type, 1	AR
BGN	301870	Meester-Loeys syndrome;Spondyloepimetaphyseal dysplasia, XL	XL, XLR
C1R	613785	Ehlers-Danlos syndrome, periodontal type, 1	AD
C1S	120580	C1s deficiency;Ehlers-Danlos syndrome, periodontal type, 2	AD
CBS	613381	Thrombosis, hyperhomocysteinemic;Homocystinuria, B6-responsive and nonresponsive types	AR
CHST14	608429	Ehlers-Danlos syndrome, musculocontractural type 1	AR
COL11A1	120280	Marshall syndrome;Fibrochondrogenesis 1;Lumbar disc herniation, susceptibility to;?Deafness, AD 37;Stickler syndrome, type II	AD, AR

COL11A2	120290	Deafness, AR 53;Otospondylomegaepiphyseal dysplasia, AR;Fibrochondrogenesis 2;Otospondylomegaepiphyseal dysplasia, AD;Deafness, AD 13	AR, AD, AR, AD
COL12A1	120320	Ullrich congenital muscular dystrophy 2;Bethlem myopathy 2	AR, AD
COL1A1	120150	Ehlers-Danlos syndrome, arthrochalasia type, 1;Bone mineral density variation QTL, osteoporosis;Osteogenesis imperfecta, type III;Osteogenesis imperfecta, type I;Caffey disease;Osteogenesis imperfecta, type IV;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1;Osteogenesis imperfecta, type II	AD

COL1A2	120160	Osteoporosis, postmenopausal;Ehlers-Danlos syndrome, cardiac valvular type;Ehlers-Danlos syndrome, arthrochalasia type, 2;Osteogenesis imperfecta, type III;Osteogenesis imperfecta, type IV;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2;Osteogenesis imperfecta, type II	AD, AR
COL2A1	120140	Legg-Calve-Perthes disease;Stickler syndrome, type I;Osteoarthritis with mild chondrodysplasia;Platyspondylic skeletal dysplasia, Torrance type;Spondyloepiphyseal dysplasia, Stanescu type;Kniest dysplasia;Czech dysplasia;Stickler sydrome, type I, nonsyndromic ocular;?Vitreoretinopathy with phalangeal epiphyseal dysplasia;?Epiphyseal dysplasia, multiple, with myopia and deafness;Avascular necrosis of the femoral head;Spondyloperipheral dysplasia;Achondrogenesis, type II or hypochondrogenesis;SMED Strudwick type;SED congenita	AD

COL3A1	120180	Ehlers-Danlos syndrome, vascular type;Polymicrogyria with or without vascular-type EDS	AD, AR
COL5A1	120215	Fibromuscular dysplasia, multifocal;Ehlers-Danlos syndrome, classic type, 1	AD
COL5A2	120190	Ehlers-Danlos syndrome, classic type, 2	AD
COL9A1	120210	Epiphyseal dysplasia, multiple, 6;Stickler syndrome, type IV	AD
COL9A2	120260	Stickler syndrome, type V;Epiphyseal dysplasia, multiple, 2	AR, AD
COL9A3	120270	Epiphyseal dysplasia, multiple, 3, with or without myopathy;Intervertebral disc disease, susceptibility to	AD
CREB3L1	616215	Osteogenesis imperfecta, type XVI	AR
DSE	605942	Ehlers-Danlos syndrome, musculocontractural type 2	AR
EFEMP2	604633	Cutis laxa, AR, type IB	AR
ELN	130160	Cutis laxa, AD;Supravalvar aortic stenosis	AD
FBLN5	604580	Cutis laxa, AR, type IA;Neuropathy, hereditary, with or without age-related macular degeneration;Macular degeneration, age-related, 3;?Cutis laxa, AD 2	AR, AD

FBN1	134797	Marfan lipodystrophy syndrome;Geleophysic dysplasia 2;Acromicric dysplasia;Marfan syndrome;Weill-Marchesani syndrome 2, dominant;Stiff skin syndrome;MASS syndrome;Ectopia lentis, familial	AD
FBN2	612570	Contractural arachnodactyly, congenital;Macular degeneration, early-onset	AD
FKBP14	614505	Ehlers-Danlos syndrome, kyphoscoliotic type, 2	AR
FLCN	607273	Birt-Hogg-Dube syndrome;Pneumothorax, primary spontaneous;Renal carcinoma, chromophobe, somatic;Colorectal cancer, somatic	AD
FLNA	300017	Frontometaphyseal dysplasia 1;Heterotopia, periventricular, 1;Terminal osseous dysplasia;Congenital short bowel syndrome;Otopalatodigital syndrome, type II;Melnick-Needles syndrome;Cardiac valvular dysplasia, XL;Intestinal pseudoobstruction, neuronal;?FG syndrome 2;Otopalatodigital syndrome, type I	XLR, XLD, XL
GORAB	607983	Geroderma osteodysplasticum	AR

ITGB4	147557	Epidermolysis bullosa, junctional, non-Herlitz type;Epidermolysis bullosa of hands and feet;Epidermolysis bullosa, junctional, with pyloric atresia	AR, AD
LAMA3	600805	Laryngoonychocutaneous syndrome;Epidermolysis bullosa, junctional, Herlitz type;Epidermolysis bullosa, generalized atrophic benign	AR
LAMB3	150310	Epidermolysis bullosa, junctional, non-Herlitz type;Amelogenesis imperfecta, type IA;Epidermolysis bullosa, junctional, Herlitz type	AR, AD
LAMC2	150292	Epidermolysis bullosa, junctional, non-Herlitz type;Epidermolysis bullosa, junctional, Herlitz type	AR
LOX	153455	Aortic aneurysm, familial thoracic 10	AD
LRP2	600073	Donnai-Barrow syndrome	AR
LTBP3	602090	Geleophysic dysplasia 3;Dental anomalies and short stature	AD, AR
MBTPS2	300294	Osteogenesis imperfecta, type XIX;?Olmsted syndrome, XL;Keratosis follicularis spinulosa decalvans, XL;IFAP syndrome with or without BRESHECK syndrome	XLR

MED12	300188	Opitz-Kaveggia syndrome;Lujan-Fryns syndrome;Ohdo syndrome, XL;Hardikar syndrome	XLR, XLD
MFAP5	601103	Aortic aneurysm, familial thoracic 9	AD
MYH11	160745	Aortic aneurysm, familial thoracic 4;Visceral myopathy 2;Megacystis-microcolon-intestinal hypoperistalsis syndrome 2	AD, AR
MYLK	600922	Aortic aneurysm, familial thoracic 7;Megacystis-microcolon-intestinal hypoperistalsis syndrome	AD, AR
NOTCH1	190198	Adams-Oliver syndrome 5;Aortic valve disease 1	AD
PLOD1	153454	Ehlers-Danlos syndrome, kyphoscoliotic type, 1	AR
PRDM5	614161	Brittle cornea syndrome 2	AR
PRKG1	176894	Aortic aneurysm, familial thoracic 8	AD
PYCR1	179035	Cutis laxa, AR, type IIB;Cutis laxa, AR, type IIIB	AR
RIN2	610222	Macrocephaly, alopecia, cutis laxa, and scoliosis	AR
SKI	164780	Shprintzen-Goldberg syndrome	AD
SLC2A10	606145	Arterial tortuosity syndrome	AR
SLC39A13	608735	Ehlers-Danlos syndrome, spondylodysplastic type, 3	AR

SMAD2	601366	Loeys-Dietz syndrome 6;Congenital heart defects, multiple types, 8, with or without heterotaxy	AD
SMAD3	603109	Loeys-Dietz syndrome 3	AD
SMAD4	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome;Pancreatic cancer, somatic;Myhre syndrome;Polyposis, juvenile intestinal	AD
SMAD6	602931	Craniosynostosis 7, susceptibility to;Aortic valve disease 2;Radioulnar synostosis, nonsyndromic	AD
SP7	606633	Osteogenesis imperfecta, type XII	AR
SPARC	182120	Osteogenesis imperfecta, type XVII	AR
TENT5A	611357	Osteogenesis imperfecta, type XVIII	AR
TGFB2	190220	Loeys-Dietz syndrome 4	AD
TGFB3	190230	Loeys-Dietz syndrome 5;Arrhythmogenic right ventricular dysplasia 1	AD
TGFBR1	190181	Loeys-Dietz syndrome 1;Multiple self-healing squamous epithelioma, susceptibility to	AD
TGFBR2	190182	Colorectal cancer, hereditary nonpolyposis, type 6;Loeys-Dietz syndrome 2;Esophageal cancer, somatic	AD
TNXB	600985	Vesicoureteral reflux 8;Ehlers-Danlos syndrome, classic-like, 1	AD, AR

VCAN	118661	Wagner syndrome 1	AD
WNT1	164820	Osteogenesis imperfecta, type XV;Osteoporosis, early-onset, susceptibility to, AD	AR
ZNF469	612078	Brittle cornea syndrome 1	AR