Oslo universitetssykehus
Avdeling for medisinsk genetikk

Genpanel for Osteogenesis imperfekta

Genpanel, versjon v01

* Enkelte genomiske regioner har lav eller ingen sekvensdekning ved eksomsekvensering. Dette skyldes at de har stor likhet med andre områder i genomet, slik at spesifikk gjenkjennelse av disse områdene og påvisning av varianter i disse områdene, blir vanskelig og upålitelig. Disse genetiske regionene har vi identifisert ved å benytte USCS segmental duplication hvor områder større enn 1 kb og ≥90% likhet med andre regioner i genomet, gjenkjennes (https://genome.ucsc.edu).

Vi gjør oppmerksom på at ved identifiseringav ekson oppstrøms for startkodon kan eksonnummereringen endres uten at transkript ID endres.

Avdelingens websider har en full oversikt over områder som er affisert av segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ALPL	438	NM_000478.4		2-12	Hypophosphatasia, adult OMIM Hypophosphatasia, childhood OMIM Hypophosphatasia, infantile OMIM Odontohypophosphatasia OMIM
B3GALT6	17978	NM_080605.3		1	Al-Gazali syndrome OMIM Ehlers-Danlos syndrome, spondylodysplastic type, 2 OMIM Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures OMIM
B4GALT7	930	NM_007255.2		1-6	Ehlers-Danlos syndrome, spondylodysplastic type, 1 OMIM
BMP1	1067	NM_006129.4		1-20	Osteogenesis imperfecta, type XIII OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CASR	1514	NM_000388.3		2-7	Hyperparathyroidism, neonatal OMIM Hypocalcemia, autosomal dominant OMIM Hypocalcemia, autosomal dominant, with Bartter syndrome OMIM Hypocalciuric hypercalcemia, type I OMIM {Epilepsy idiopathic generalized, susceptibility to, 8} OMIM
COL1A1	2197	NM_000088.3		1-51	Caffey disease OMIM Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 1 OMIM Ehlers-Danlos syndrome, arthrochalasia type, 1 OMIM Osteogenesis imperfecta, type I OMIM Osteogenesis imperfecta, type II OMIM Osteogenesis imperfecta, type III OMIM Osteogenesis imperfecta, type IV OMIM {Bone mineral density variation QTL, osteoporosis} OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
COL1A2	2198	NM_000089.3		1-52	Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2 OMIM Ehlers-Danlos syndrome, arthrochalasia type, 2 OMIM Ehlers-Danlos syndrome, cardiac valvular type OMIM Osteogenesis imperfecta, type II OMIM Osteogenesis imperfecta, type III OMIM Osteogenesis imperfecta, type IV OMIM {Osteoporosis, postmenopausal} OMIM
COPB2	2232	NM_004766.3		1-22	?Microcephaly 19, primary, autosomal recessive OMIM
CREB3L1	18856	NM_052854.3		1-12	Osteogenesis imperfecta, type XVI OMIM
CRTAP	2379	NM_006371.4		1-7	Osteogenesis imperfecta, type VII OMIM
DSPP	3054	NM_014208.3		2-5	Deafness, autosomal dominant 39, with dentinogenesis OMIM Dentin dysplasia, type II OMIM Dentinogenesis imperfecta, Shields type II OMIM Dentinogenesis imperfecta, Shields type III OMIM
FAM46A	18345	NM_017633.2		2-3	Osteogenesis imperfecta, type XVIII OMIM
FKBP10	18169	NM_021939.4		1-10	Bruck syndrome 1 OMIM Osteogenesis imperfecta, type XI OMIM
GORAB	25676	NM_152281.2		1-5	Geroderma osteodysplasticum OMIM
IFITM5	16644	NM_001025295.2		1-2	Osteogenesis imperfecta, type V OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
KDELR2	6305	NM_006854.3		1-5	Osteogenesis imperfecta 21 OMIM
LEPRE1	19316	NM_022356.3		1-15	Osteogenesis imperfecta, type VIII OMIM
LRP5	6697	NM_002335.3	1, 3-9	1-23	Exudative vitreoretinopathy 4 OMIM Hyperostosis, endosteal OMIM Osteopetrosis, autosomal dominant 1 OMIM Osteoporosis-pseudoglioma syndrome OMIM Osteosclerosis OMIM Polycystic liver disease 4 with or without kidney cysts OMIM van Buchem disease, type 2 OMIM [Bone mineral density variability 1] OMIM {Osteoporosis} OMIM
MESDC2	13520	NM_015154.2		1-3	Osteogenesis imperfecta, type XX OMIM
NBAS	15625	NM_015909.4		1-52	Infantile liver failure syndrome 2 OMIM Short stature, optic nerve atrophy, and Pelger-Huet anomaly OMIM
NOTCH2	7882	NM_024408.4	1-4	1-34	Alagille syndrome 2 OMIM Hajdu-Cheney syndrome OMIM
NUDT6	8053	NM_007083.4		1-5	
P4HB	8548	NM_000918.3		1-11	Cole-Carpenter syndrome 1 OMIM
PLOD2	9082	NM_182943.3		1-20	Bruck syndrome 2 OMIM
PLS3	9091	NM_005032.7		2-16	Bone mineral density QTL18, osteoporosis OMIM
PPIB	9255	NM_000942.4		1-5	Osteogenesis imperfecta, type IX OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
SEC24D	10706	NM_014822.2		2-23	Cole-Carpenter syndrome 2 OMIM
SERPINF1	8824	NM_002615.5		2-8	Osteogenesis imperfecta, type VI OMIM
SERPINH1	1546	NM_001235.3		2-5	Osteogenesis imperfecta, type X OMIM {Preterm premature rupture of the membranes, susceptibility to} OMIM
SP7	17321	NM_001173467.2		2-3	Osteogenesis imperfecta, type XII OMIM
SPARC	11219	NM_003118.3		2-10	Osteogenesis imperfecta, type XVII OMIM
TAPT1	26887	NM_153365.2		1-14	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type OMIM
TMEM38B	25535	NM_018112.2		1-6	Osteogenesis imperfecta, type XIV OMIM
TRPV6	14006	NM_018646.5		1-15	Hyperparathyroidism, transient neonatal OMIM
WNT1	12774	NM_005430.3		1-4	Osteogenesis imperfecta, type XV OMIM {Osteoporosis, early-onset, susceptibility to, autosomal dominant} OMIM