Oslo universitetssykehus
Avdeling for medisinsk genetikk

Genpanel for skjelettdysplasier

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).

For noen gener ligger alle ekson i områder med segmentale duplikasjoner: **GJA1, RBM8A, SBDS, SHOX**

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
ABCC9	60	NM_020297.3		1-38	Hypertrichotic osteochondrodysplasia OMIM
ABL1	76	NM_005157.6		1-11	Congenital heart defects and skeletal malformations syndrome OMIM
ACAN	319	NM_013227.3		2-18	?Spondyloepiphyseal dysplasia, Kimberley type OMIM Short stature and advanced bone age, with or without early-onset osteoarthritis and/or osteochondritis dissecans OMIM Spondyloepimetaphyseal dysplasia, aggrecan type OMIM
ACP5	124	NM_001111035.1		4-7	Spondyloenchondrodysplasia with immune dysregulation OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ACVR1	171	NM_001111067.2		3-11	Fibrodysplasia ossificans progressiva OMIM
ADAMTS10	13201	NM_030957.4		3-26	Weill-Marchesani syndrome 1, recessive OMIM
ADAMTS17	17109	NM_139057.3		1-22	Weill-Marchesani 4 syndrome, recessive OMIM
ADAMTSL2	14631	NM_001145320.1	10-19	2-19	Geleophysic dysplasia 1 OMIM
AGA	318	NM_001171988.1		1-9	Aspartylglucosaminuria OMIM
AGPS	327	NM_003659.3		1-20	Rhizomelic chondrodysplasia punctata, type 3 OMIM
ALG12	19358	NM_024105.4		2-10	Congenital disorder of glycosylation, type Ig OMIM
ALG3	23056	NM_001006941.2		1-9	Congenital disorder of glycosylation, type Id OMIM
ALG9	15672	NM_001077691.2		5-15	Congenital disorder of glycosylation, type Il OMIM Gillessen-Kaesbach-Nishimura syndrome OMIM
ALPL	438	NM_001177520.1		2-10	Hypophosphatasia, adult OMIM Hypophosphatasia, childhood OMIM Hypophosphatasia, infantile OMIM Odontohypophosphatasia OMIM
ALX1	1494	NM_006982.3		1-4	Frontonasal dysplasia 3 OMIM
ALX3	449	NM_006492.3		1-4	Frontonasal dysplasia 1 OMIM
ALX4	450	NM_021926.4		1-4	Frontonasal dysplasia 2 OMIM
AMER1	26837	NM_152424.3		2	Osteopathia striata with cranial sclerosis OMIM
ANKH	15492	NM_054027.4		1-12	Chondrocalcinosis 2 OMIM Craniometaphyseal dysplasia OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ANKRD11	21316	NM_001256183.2	13	3-13	KBG syndrome OMIM
ANO5	27337	NM_001142649.1		1-22	Gnathodiaphyseal dysplasia OMIM
ANTXR2	21732	NM_058172.6		1-17	Hyaline fibromatosis syndrome OMIM
ARHGAP31	29216	NM_020754.4		1-12	Adams-Oliver syndrome 1 OMIM
ARL6	13210	NM_001278293.1		2-8	Bardet-Biedl syndrome 3 OMIM
ARSB	714	NM_000046.4		1-8	Mucopolysaccharidosis type VI (Maroteaux-Lamy) OMIM
ARSE	719	NM_001282631.1	8-10	1-10	Chondrodysplasia punctata, X-linked recessive OMIM
ASXL1	18318	NM_015338.5		1-12	Bohring-Opitz syndrome OMIM
ASXL2	23805	NM_018263.4		1-12	Shashi-Pena syndrome OMIM
ATP6V0A2	18481	NM_012463.3		1-20	Cutis laxa, autosomal recessive, type IIA OMIM Wrinkly skin syndrome OMIM
ATP7A	869	NM_000052.7		2-23	Menkes disease OMIM Occipital horn syndrome 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
B3GALTL	20207	NM_194318.3		1-15	Peters-plus syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
B3GAT3	923	NM_012200.3	3-5	1-5	Multiple joint dislocations, short stature, craniofacial dysmorphism, with or without congenital heart defects OMIM
B4GALT7	930	NM_007255.2		1-6	Ehlers-Danlos syndrome, spondylodysplastic type, 1 OMIM
B9D1	24123	NM_001243475.1		2-7	?Meckel syndrome 9 OMIM Joubert syndrome 27 OMIM
BBS1	966	NM_024649.4		1-17	Bardet-Biedl syndrome 1 OMIM
BBS10	26291	NM_024685.3		1-2	Bardet-Biedl syndrome 10 OMIM
BBS12	26648	NM_152618.2		2	Bardet-Biedl syndrome 12 OMIM
BBS2	967	NM_031885.3		1-17	Bardet-Biedl syndrome 2 OMIM
BBS4	969	NM_033028.4		1-16	Bardet-Biedl syndrome 4 OMIM
BBS5	970	NM_152384.3		1-12	Bardet-Biedl syndrome 5 OMIM
BBS7	18758	NM_176824.2		1-19	Bardet-Biedl syndrome 7 OMIM
BBS9	30000	NM_198428.2		2-23	Bardet-Biedl syndrome 9 OMIM
BHLHA9	35126	NM_001164405.1		1	?Camptosynpolydactyly, complex OMIM Syndactyly, mesoaxial synostotic, with phalangeal reduction OMIM
BMP1	1067	NM_006129.4		1-20	Osteogenesis imperfecta, type XIII OMIM
BMP2	1069	NM_001200.4		2-3	Brachydactyly, type A2 OMIM Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
BMPER	24154	NM_133468.4		2-16	Diaphanospondylodysostosis OMIM
BMPR1B	1077	NM_001203.2		4-13	Acromesomelic dysplasia, Demirhan type OMIM Brachydactyly, type A1, D OMIM Brachydactyly, type A2 OMIM
C16orf62	24641	NM_020314.5		1-31	Ritscher-Schinzel syndrome 3 OMIM
C21orf2	1260	NM_004928.2		1-7	Spondylometaphyseal dysplasia, axial OMIM
C2CD3	24564	NM_001286577.1		1-33	Orofaciodigital syndrome XIV OMIM
CA2	1373	NM_000067.2		1-7	Osteopetrosis, autosomal recessive 3, with renal tubular acidosis OMIM
CANT1	19721	NM_001159773.1		3-5	Desbuquois dysplasia 1 OMIM Epiphyseal dysplasia, multiple, 7 OMIM
CASR	1514	NM_001178065.1		2-7	Hyperparathyroidism, neonatal OMIM Hypocalcemia, autosomal dominant OMIM Hypocalcemia, autosomal dominant, with Bartter syndrome OMIM Hypocalciuric hypercalcemia, type I OMIM
CC2D2A	29253	NM_001080522.2		3-38	Joubert syndrome 9 OMIM Meckel syndrome 6 OMIM
CCDC8	25367	NM_032040.5		1	3-M syndrome 3 OMIM
CDC45	1739	NM_001178010.2		1-19	Meier-Gorlin syndrome 7 OMIM PubMed
CDH3	1762	NM_001793.5		1-16	Ectodermal dysplasia, ectrodactyly, and macular dystrophy OMIM Hypotrichosis, congenital, with juvenile macular dystrophy OMIM
CDKN1C	1786	NM_000076.2		1-2	IMAGE syndrome OMIM

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CDT1	24576	NM_030928.3		1-10	Meier-Gorlin syndrome 4 OMIM
CEP120	26690	NM_153223.3		2-21	Joubert syndrome 31 OMIM Short-rib thoracic dysplasia 13 with or without polydactyly OMIM
CEP290	29021	NM_025114.3	54	2-54	?Bardet-Biedl syndrome 14 OMIM Joubert syndrome 5 OMIM Meckel syndrome 4 OMIM
CHST14	24464	NM_130468.3		1	Ehlers-Danlos syndrome, musculocontractural type 1 OMIM
CHST3	1971	NM_004273.4		2-3	Spondyloepiphyseal dysplasia with congenital joint dislocations OMIM
CHSY1	17198	NM_014918.4		1-3	Temtamy preaxial brachydactyly syndrome OMIM
CLCN5	2023	NM_001127899.3		3-15	Dent disease 1 OMIM Hypophosphatemic rickets OMIM
CLCN7	2025	NM_001287.5		1-25	Hypopigmentation, organomegaly, and delayed myelination and development OMIM Osteopetrosis, autosomal dominant 2 OMIM Osteopetrosis, autosomal recessive 4 OMIM
COG1	6545	NM_018714.2		1-14	Congenital disorder of glycosylation, type IIg OMIM
COL10A1	2185	NM_000493.3		2-3	Metaphyseal chondrodysplasia, Schmid type OMIM
COL11A1	2186	NM_080629.2		1-67	Fibrochondrogenesis 1 OMIM Marshall syndrome OMIM Stickler syndrome, type II OMIM
COL11A2	2187	NM_080681.2		1-64	Fibrochondrogenesis 2 OMIM Otospondylomegaepiphyseal dysplasia, autosomal dominant OMIM Otospondylomegaepiphyseal dysplasia, autosomal recessive OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
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
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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
COL2A1	2200	NM_033150.3		1-53	?Epiphyseal dysplasia, multiple, with myopia and deafness OMIM ?Vitreoretinopathy with phalangeal epiphyseal dysplasia OMIM Achondrogenesis, type II or hypochondrogenesis OMIM Avascular necrosis of the femoral head OMIM Czech dysplasia OMIM Kniest dysplasia OMIM Legg-Calve-Perthes disease OMIM Osteoarthritis with mild chondrodysplasia OMIM Platyspondylic skeletal dysplasia, Torrance type OMIM SED congenita OMIM SMED Strudwick type OMIM Spondyloepiphyseal dysplasia, Stanescu type OMIM Spondyloperipheral dysplasia OMIM Stickler sydrome, type I, nonsyndromic ocular OMIM Stickler syndrome, type I OMIM
COL9A1	2217	NM_001851.5		1-38	?Epiphyseal dysplasia, multiple, 6 OMIM Stickler syndrome, type IV OMIM
COL9A2	2218	NM_001852.4		1-32	?Stickler syndrome, type V OMIM Epiphyseal dysplasia, multiple, 2 OMIM
COL9A3	2219	NM_001853.3		1-32	Epiphyseal dysplasia, multiple, 3, with or without myopathy OMIM
COLEC11	17213	NM_001255983.1		2-6	3MC syndrome 2 OMIM
COMP	2227	NM_000095.3		1-19	Epiphyseal dysplasia, multiple, 1 OMIM Pseudoachondroplasia 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

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CREBBP	2348	NM_004380.3		1-31	Menke-Hennekam syndrome 1 OMIM Rubinstein-Taybi syndrome 1 OMIM
CRTAP	2379	NM_006371.4		1-7	Osteogenesis imperfecta, type VII OMIM
CSPP1	26193	NM_024790.6		1-29	Joubert syndrome 21 OMIM
CTSA	9251	NM_000308.2		1-15	Galactosialidosis OMIM
CTSC	2528	NM_001814.5		1-7	Haim-Munk syndrome OMIM Papillon-Lefevre syndrome OMIM Periodontitis 1, juvenile OMIM
CTSK	2536	NM_000396.4		2-8	Pycnodysostosis OMIM
CUL7	21024	NM_014780.4		2-26	3-M syndrome 1 OMIM
CYP27B1	2606	NM_000785.3		1-9	Vitamin D-dependent rickets, type I OMIM
CYP2R1	20580	NM_024514.4		1-5	Rickets due to defect in vitamin D 25-hydroxylation deficiency OMIM
CYP3A4	2637	NM_001202855.3	1-13	1-13	Vitamin D-dependent rickets, type 3 OMIM
DCC	2701	NM_005215.3		1-29	Gaze palsy, familial horizontal, with progressive scoliosis, 2 OMIM
DDR2	2731	NM_001014796.3		4-19	Spondylometaepiphyseal dysplasia, short limb-hand type OMIM
DHCR24	2859	NM_014762.4		1-9	Desmosterolosis OMIM
DHCR7	2860	NM_001360.2		3-9	Smith-Lemli-Opitz syndrome OMIM
DHODH	2867	NM_001361.4		1-9	Miller syndrome OMIM
DIS3L2	28648	NM_001257281.2		2-14	Perlman syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
DLL3	2909	NM_203486.2		1-9	Spondylocostal dysostosis 1, autosomal recessive OMIM
DLL4	2910	NM_019074.3		1-11	Adams-Oliver syndrome 6 OMIM
DLX3	2916	NM_005220.3		1-3	Amelogenesis imperfecta, type IV OMIM Trichodontoosseous syndrome OMIM
DLX5	2918	NM_005221.5		1-3	?Split-hand/foot malformation 1 with sensorineural hearing loss OMIM Split-hand/foot malformation 1 OMIM
DMP1	2932	NM_004407.3		2-6	Hypophosphatemic rickets, AR OMIM
DNMT3A	2978	NM_175629.2		2-23	Heyn-Sproul-Jackson syndrome OMIM Tatton-Brown-Rahman syndrome OMIM
DOCK6	19189	NM_020812.3		1-48	Adams-Oliver syndrome 2 OMIM
DPAGT1	2995	NM_001382.3		1-9	Congenital disorder of glycosylation, type Ij OMIM
DPM1	3005	NM_003859.1		1-9	Congenital disorder of glycosylation, type Ie 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
DVL1	3084	NM_004421.2		1-15	Robinow syndrome, autosomal dominant 2 OMIM
DVL3	3087	NM_004423.4		1-15	Robinow syndrome, autosomal dominant 3 OMIM
DYM	21317	NM_017653.3		2-17	Dyggve-Melchior-Clausen disease OMIM Smith-McCort dysplasia OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
DYNC2H1	2962	NM_001080463.1		1-90	Short-rib thoracic dysplasia 3 with or without polydactyly OMIM
DYNC2LI1	24595	NM_001193464.2		1-13	Short-rib thoracic dysplasia 15 with polydactyly OMIM
EBP	3133	NM_006579.2		2-5	Chondrodysplasia punctata, X-linked dominant OMIM MEND syndrome OMIM
EED	3188	NM_003797.3		1-12	Cohen-Gibson syndrome OMIM
EFTUD2	30858	NM_001142605.1		2-27	Mandibulofacial dysostosis, Guion-Almeida type OMIM
EIF2AK3	3255	NM_004836.5		1-17	Wolcott-Rallison syndrome OMIM
ENPP1	3356	NM_006208.2		1-25	Arterial calcification, generalized, of infancy, 1 OMIM Cole disease OMIM Hypophosphatemic rickets, autosomal recessive, 2 OMIM
EOGT	28526	NM_001278689.1		4-18	Adams-Oliver syndrome 4 OMIM
ERF	3444	NM_006494.3		1-4	Chitayat syndrome OMIM Craniosynostosis 4 OMIM
ESCO2	27230	NM_001017420.3		2-11	Juberg-Hayward syndrome OMIM Roberts-SC phocomelia syndrome OMIM
EVC	3497	NM_153717.2		1-21	?Weyers acrofacial dysostosis OMIM Ellis-van Creveld syndrome OMIM
EVC2	19747	NM_001166136.1		2-22	Ellis-van Creveld syndrome OMIM Weyers acrofacial dysostosis OMIM
EXT1	3512	NM_000127.2		1-11	Exostoses, multiple, type 1 OMIM
EXT2	3513	NM_207122.1		2-14	Exostoses, multiple, type 2 OMIM Seizures, scoliosis, and macrocephaly syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
EXTL3	3518	NM_001440.3		3-7	Immunoskeletal dysplasia with neurodevelopmental abnormalities OMIM
EZH2	3527	NM_004456.5		2-20	Weaver syndrome OMIM
FAH	3579	NM_000137.2		1-14	Tyrosinemia, type I OMIM
FAM111A	24725	NM_001142520.1		4-5	Gracile bone dysplasia OMIM Kenny-Caffey syndrome, type 2 OMIM
FAM20C	22140	NM_020223.3		1-10	Raine syndrome OMIM
FAM46A	18345	NM_017633.2		2-3	Osteogenesis imperfecta, type XVIII OMIM
FAM58A	28434	NM_152274.4		1-5	STAR syndrome OMIM
FBLN1	3600	NM_006486.3		1-17	Synpolydactyly, 3/3'4, associated with metacarpal and metatarsal synostoses OMIM
FBN1	3603	NM_000138.4		2-66	Acromicric dysplasia OMIM Geleophysic dysplasia 2 OMIM Marfan lipodystrophy syndrome OMIM Marfan syndrome OMIM MASS syndrome OMIM Stiff skin syndrome OMIM Weill-Marchesani syndrome 2, dominant OMIM
FBN2	3604	NM_001999.3		1-65	Contractural arachnodactyly, congenital OMIM
FERMT3	23151	NM_031471.6		2-15	Leukocyte adhesion deficiency, type III OMIM
FGF10	3666	NM_004465.1		1-3	LADD syndrome OMIM
FGF16	3672	NM_003868.2		1-2	Metacarpal 4-5 fusion OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
FGF23	3680	NM_020638.2		1-3	Hypophosphatemic rickets, autosomal dominant OMIM Tumoral calcinosis, hyperphosphatemic, familial, 2 OMIM
FGFR1	3688	NM_001174063.1		2-18	Hartsfield syndrome OMIM Hypogonadotropic hypogonadism 2 with or without anosmia OMIM Jackson-Weiss syndrome OMIM Osteoglophonic dysplasia OMIM Pfeiffer syndrome OMIM Trigonocephaly 1 OMIM
FGFR2	3689	NM_000141.4		2-18	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis OMIM Apert syndrome OMIM Beare-Stevenson cutis gyrata syndrome OMIM Bent bone dysplasia syndrome OMIM Craniofacial-skeletal-dermatologic dysplasia OMIM Crouzon syndrome OMIM Jackson-Weiss syndrome OMIM LADD syndrome OMIM Pfeiffer syndrome OMIM Saethre-Chotzen syndrome OMIM Scaphocephaly, maxillary retrusion, and mental retardation OMIM
FGFR3	3690	NM_001163213.1		2-18	Achondroplasia OMIM CATSHL syndrome OMIM Crouzon syndrome with acanthosis nigricans OMIM Hypochondroplasia OMIM LADD syndrome OMIM Muenke syndrome OMIM SADDAN OMIM Thanatophoric dysplasia, type I OMIM Thanatophoric dysplasia, type II OMIM
FIG4	16873	NM_014845.5		1-23	Yunis-Varon syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
FKBP10	18169	NM_021939.4		1-10	Bruck syndrome 1 OMIM Osteogenesis imperfecta, type XI OMIM
FLNA	3754	NM_001456.3		2-47	?FG syndrome 2 OMIM Frontometaphyseal dysplasia 1 OMIM Melnick-Needles syndrome OMIM Otopalatodigital syndrome, type I OMIM Otopalatodigital syndrome, type II OMIM Terminal osseous dysplasia OMIM
FLNB	3755	NM_001457.3		1-46	Atelosteogenesis, type I OMIM Atelosteogenesis, type III OMIM Boomerang dysplasia OMIM Larsen syndrome OMIM Spondylocarpotarsal synostosis syndrome OMIM
FN1	3778	NM_212482.1		1-46	Spondylometaphyseal dysplasia, corner fracture type OMIM
FUCA1	4006	NM_000147.4		1-8	Fucosidosis OMIM
FZD2	4040	NM_001466.4		1	Omodysplasia 2 OMIM
GALNS	4122	NM_000512.4		1-14	Mucopolysaccharidosis IVA OMIM
GALNT3	4125	NM_004482.4		2-11	Tumoral calcinosis, hyperphosphatemic, familial, 1 OMIM
GDF5	4220	NM_000557.5		1-2	?Acromesomelic dysplasia, Hunter-Thompson type OMIM Brachydactyly, type A1, C OMIM Brachydactyly, type A2 OMIM Brachydactyly, type C OMIM Chondrodysplasia, Grebe type OMIM Du Pan syndrome OMIM Multiple synostoses syndrome 2 OMIM Symphalangism, proximal, 1B OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
GDF6	4221	NM_001001557.3		1-2	Klippel-Feil syndrome 1, autosomal dominant OMIM Multiple synostoses syndrome 4 OMIM
GHR	4263	NM_001242406.2		1-9	Growth hormone insensitivity, partial OMIM Increased responsiveness to growth hormone OMIM Laron dwarfism OMIM
GJA1	4274	NM_000165.5	2	2	Craniometaphyseal dysplasia, autosomal recessive OMIM Oculodentodigital dysplasia OMIM Oculodentodigital dysplasia, autosomal recessive OMIM Syndactyly, type III OMIM
GLB1	4298	NM_000404.3		1-16	GM1-gangliosidosis, type I OMIM GM1-gangliosidosis, type II OMIM GM1-gangliosidosis, type III OMIM Mucopolysaccharidosis type IVB (Morquio) OMIM
GLI3	4319	NM_000168.5		2-15	Greig cephalopolysyndactyly syndrome OMIM Pallister-Hall syndrome OMIM Polydactyly, postaxial, types A1 and B OMIM Polydactyly, preaxial, type IV OMIM
GNAS	4392	NM_016592.4		1	Osseous heteroplasia, progressive OMIM Pseudohypoparathyroidism Ia OMIM Pseudohypoparathyroidism Ib OMIM Pseudohypoparathyroidism Ic OMIM Pseudopseudohypoparathyroidism OMIM
GNPAT	4416	NM_014236.4		1-16	Rhizomelic chondrodysplasia punctata, type 2 OMIM
GNPTAB	29670	NM_024312.5		1-21	Mucolipidosis II alpha/beta OMIM Mucolipidosis III alpha/beta OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
GNPTG	23026	NM_032520.4		1-11	Mucolipidosis III gamma OMIM
GNS	4422	NM_002076.3		1-14	Mucopolysaccharidosis type IIID OMIM
GORAB	25676	NM_152281.2		1-5	Geroderma osteodysplasticum OMIM
GPC6	4454	NM_005708.3		1-9	Omodysplasia 1 OMIM
GPX4	4556	NM_001039847.2		1-7	Spondylometaphyseal dysplasia, Sedaghatian type OMIM
GSC	4612	NM_173849.2		1-3	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities OMIM
GUSB	4696	NM_000181.3	11	1-12	Mucopolysaccharidosis VII OMIM
GZF1	15808	NM_022482.3		1-5	Joint laxity, short stature, and myopia OMIM
HDAC4	14063	NM_006037.3		2-27	Brachydactylies PubMed
HDAC8	13315	NM_018486.2		1-11	Cornelia de Lange syndrome 5 OMIM
HES7	15977	NM_032580.3		1-4	Spondylocostal dysostosis 4, autosomal recessive OMIM
HGSNAT	26527	NM_152419.3		1-18	Mucopolysaccharidosis type IIIC (Sanfilippo C) OMIM
HNRNPK	5044	NM_031262.2	17	3-17	Au-Kline syndrome OMIM
HOXA13	5102	NM_000522.5		1-2	?Guttmacher syndrome OMIM Hand-foot-uterus syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
HOXD13	5136	NM_000523.3		1-2	?Brachydactyly-syndactyly syndrome OMIM Brachydactyly, type D OMIM Brachydactyly, type E OMIM Syndactyly, type V OMIM Synpolydactyly 1 OMIM
HPGD	5154	NM_001256307.1		3-6	?Digital clubbing, isolated congenital OMIM Cranioosteoarthropathy OMIM Hypertrophic osteoarthropathy, primary, autosomal recessive 1 OMIM
HSPG2	5273	NM_005529.6		1-97	Dyssegmental dysplasia, Silverman-Handmaker type OMIM Schwartz-Jampel syndrome, type 1 OMIM
ICK	21219	NM_014920.3		2-14	Endocrine-cerebroosteodysplasia OMIM
IDS	5389	NM_001166550.3	2-3	2-9	Mucopolysaccharidosis II OMIM
IDUA	5391	NM_000203.4		1-14	Mucopolysaccharidosis Ih OMIM Mucopolysaccharidosis Ih/s OMIM Mucopolysaccharidosis Is OMIM
IFIH1	18873	NM_022168.3		1-16	Singleton-Merten syndrome 1 OMIM
IFITM5	16644	NM_001025295.2		1-2	Osteogenesis imperfecta, type V OMIM
IFT122	13556	NM_018262.3	13-18	1-29	Cranioectodermal dysplasia 1 OMIM
IFT140	29077	NM_014714.3		3-31	Short-rib thoracic dysplasia 9 with or without polydactyly OMIM
IFT172	30391	NM_015662.3		1-48	Bardet-Biedl syndrome 20 OMIM Short-rib thoracic dysplasia 10 with or without polydactyly OMIM
IFT43	29669	NM_001102564.1		1-9	?Cranioectodermal dysplasia 3 OMIM Short-rib thoracic dysplasia 18 with polydactyly OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
IFT52	15901	NM_016004.5		2-14	Short-rib thoracic dysplasia 16 with or without polydactyly OMIM
IFT80	29262	NM_020800.2		2-20	Short-rib thoracic dysplasia 2 with or without polydactyly OMIM
IFT81	14313	NM_001143779.1		2-19	Short-rib thoracic dysplasia 19 with or without polydactyly OMIM
IHH	5956	NM_002181.3		1-3	Acrocapitofemoral dysplasia OMIM Brachydactyly, type A1 OMIM
IL11RA	5967	NM_001142784.2		2-13	Craniosynostosis and dental anomalies OMIM
IL1RN	6000	NM_173843.2		3-6	Interleukin 1 receptor antagonist deficiency OMIM
IMPAD1	26019	NM_017813.5		1-5	Chondrodysplasia with joint dislocations, GPAPP type OMIM
INPPL1	6080	NM_001567.4		1-28	Opsismodysplasia OMIM
KAT6B	17582	NM_001256468.1		3-18	Genitopatellar syndrome OMIM SBBYSS syndrome OMIM
KDELR2	6305	NM_006854.3		1-5	Osteogenesis imperfecta 21 OMIM
KIAA0753	29110	NM_014804.2		2-19	?Joubert syndrome 38 OMIM ?Orofaciodigital syndrome XV OMIM Short-rib thoracic dysplasia 21 without polydactyly OMIM
KIF22	6391	NM_001256270.1		2-14	Spondyloepimetaphyseal dysplasia with joint laxity, type 2 OMIM
KIF7	30497	NM_198525.2		2-19	?Al-Gazali-Bakalinova syndrome OMIM ?Hydrolethalus syndrome 2 OMIM Acrocallosal syndrome OMIM Joubert syndrome 12 OMIM
KMT2D	7133	NM_003482.3		1-54	Kabuki syndrome 1 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
LBR	6518	NM_194442.2		2-14	Greenberg skeletal dysplasia OMIM Pelger-Huet anomaly OMIM Pelger-Huet anomaly with mild skeletal anomalies OMIM
LEMD3	28887	NM_014319.4		1-13	Buschke-Ollendorff syndrome OMIM Osteopoikilosis with or without melorheostosis OMIM
LEPRE1	19316	NM_022356.3		1-15	Osteogenesis imperfecta, type VIII OMIM
LIFR	6597	NM_002310.5		2-20	Stuve-Wiedemann syndrome/Schwartz-Jampel type 2 syndrome OMIM
LMBR1	13243	NM_022458.4		1-17	Acheiropody OMIM Hypoplastic or aplastic tibia with polydactyly OMIM Laurin-Sandrow syndrome OMIM Polydactyly, preaxial type II OMIM Syndactyly, type IV OMIM Triphalangeal thumb, type I OMIM Triphalangeal thumb-polysyndactyly syndrome OMIM
LMNA	6636	NM_001282625.1		4-13	Heart-hand syndrome, Slovenian type OMIM Hutchinson-Gilford progeria OMIM Malouf syndrome OMIM Mandibuloacral dysplasia OMIM Muscular dystrophy, congenital OMIM Restrictive dermopathy, lethal OMIM
LMX1B	6654	NM_001174146.1		1-8	Nail-patella syndrome OMIM
LONP1	9479	NM_001276480.1		3-18	CODAS syndrome OMIM
LPIN2	14450	NM_014646.2		2-20	Majeed syndrome OMIM
LRP4	6696	NM_002334.3		1-38	Cenani-Lenz syndactyly syndrome OMIM Sclerosteosis 2 OMIM

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LRP5	6697	NM_002335.3	1, 3-9	1-23	Hyperostosis, endosteal OMIM Osteopetrosis, autosomal dominant 1 OMIM Osteoporosis-pseudoglioma syndrome OMIM Osteosclerosis OMIM
LTBP3	6716	NM_001130144.2		1-28	Dental anomalies and short stature OMIM Geleophysic dysplasia 3 OMIM ?Weill-Marchesani syndrome 3, recessive OMIM
MAFB	6408	NM_005461.4		1	Multicentric carpotarsal osteolysis syndrome OMIM
MAN2B1	6826	NM_000528.4		1-24	Mannosidosis, alpha-, types I and II OMIM
MANBA	6831	NM_005908.3		1-17	Mannosidosis, beta OMIM
MAP3K7	6859	NM_003188.4		1-16	Cardiospondylocarpofacial syndrome OMIM Frontometaphyseal dysplasia 2 OMIM
MASP1	6901	NM_001879.5		1-16	3MC syndrome 1 OMIM
MATN3	6909	NM_002381.4		1-8	Epiphyseal dysplasia, multiple, 5 OMIM Spondyloepimetaphyseal dysplasia, Borochowitz-Cormier-Daire type OMIM
MEGF8	3233	NM_001410.2		1-41	Carpenter syndrome 2 OMIM
MEOX1	7013	NM_001040002.2		2-4	Klippel-Feil syndrome 2 OMIM
MESDC2	13520	NM_015154.2		1-3	Osteogenesis imperfecta, type XX OMIM
MESP2	29659	NM_001039958.1		1-2	Spondylocostal dysostosis 2, autosomal recessive OMIM
MGP	7060	NM_000900.3		1-4	Keutel syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
MKKS	7108	NM_170784.2		3-6	Bardet-Biedl syndrome 6 OMIM McKusick-Kaufman syndrome OMIM
MKS1	7121	NM_001165927.1		1-18	Bardet-Biedl syndrome 13 OMIM Joubert syndrome 28 OMIM Meckel syndrome 1 OMIM
MMP13	7159	NM_002427.4		1-10	?Spondyloepimetaphyseal dysplasia, Missouri type OMIM Metaphyseal anadysplasia 1 OMIM Metaphyseal dysplasia, Spahr type OMIM
MMP2	7166	NM_004530.5		1-13	Multicentric osteolysis, nodulosis, and arthropathy OMIM
MMP9	7176	NM_004994.3		1-13	Metaphyseal anadysplasia 2 OMIM
MNX1	4979	NM_005515.3		1-3	Currarino syndrome OMIM
MPDU1	7207	NM_004870.3		1-7	Congenital disorder of glycosylation, type lf OMIM
MSX2	7392	NM_002449.4	2	1-2	Craniosynostosis 2 OMIM Parietal foramina with cleidocranial dysplasia OMIM
MYCN	7559	NM_005378.6		2-3	Feingold syndrome 1 OMIM
NAGLU	7632	NM_000263.4		1-6	Mucopolysaccharidosis type IIIB (Sanfilippo B) OMIM
NANS	19237	NM_018946.4		1-6	Spondyloepimetaphyseal dysplasia, Camera-Genevieve type OMIM
NBAS	15625	NM_015909.4		1-52	Short stature, optic nerve atrophy, and Pelger-Huet anomaly OMIM
NEK1	7744	NM_001199398.1		3-35	Short-rib thoracic dysplasia 6 with or without polydactyly OMIM
NEU1	7758	NM_000434.3		1-6	Sialidosis, type I OMIM Sialidosis, type II OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
NF1	7765	NM_001042492.2	9-11, 13-29, 32-36	1-58	Neurofibromatosis, familial spinal OMIM Neurofibromatosis, type 1 OMIM Neurofibromatosis-Noonan syndrome OMIM Watson syndrome OMIM
NFIX	7788	NM_002501.4		1-9	Marshall-Smith syndrome OMIM Sotos syndrome 2 OMIM
NIPBL	28862	NM_133433.4		2-47	Cornelia de Lange syndrome 1 OMIM
NKX3-2	951	NM_001189.4		1-2	Spondylo-megaepiphyseal-metaphyseal dysplasia OMIM
NLRP3	16400	NM_001079821.2		3-11	CINCA syndrome OMIM
NOG	7866	NM_005450.4		1	Brachydactyly, type B2 OMIM Multiple synostoses syndrome 1 OMIM Stapes ankylosis with broad thumbs and toes OMIM Symphalangism, proximal, 1A OMIM Tarsal-carpal coalition syndrome OMIM
NOTCH1	7881	NM_017617.3		1-34	Adams-Oliver syndrome 5 OMIM Aortic valve disease 1 OMIM
NOTCH2	7882	NM_024408.4	1-4	1-34	Hajdu-Cheney syndrome OMIM
NPR2	7944	NM_003995.3		1-22	Acromesomelic dysplasia, Maroteaux type OMIM Epiphyseal chondrodysplasia, Miura type OMIM Short stature with nonspecific skeletal abnormalities OMIM
NSD1	14234	NM_172349.2		2-24	Sotos syndrome 1 OMIM
NSDHL	13398	NM_015922.3		2-8	CHILD syndrome OMIM CK syndrome OMIM

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NUDT6	8053	NM_198041.2		4-5	
NXN	18008	NM_022463.5		1-8	Robinow syndrome, autosomal recessive 2 OMIM
OBSL1	29092	NM_015311.3		1-21	3-M syndrome 2 OMIM
OCRL	8108	NM_000276.3		1-24	Dent disease 2 OMIM Lowe syndrome OMIM
OFD1	2567	NM_003611.3		1-23	Joubert syndrome 10 OMIM Orofaciodigital syndrome I OMIM Simpson-Golabi-Behmel syndrome, type 2 OMIM
ORC1	8487	NM_001190818.1		2-17	Meier-Gorlin syndrome 1 OMIM
ORC4	8490	NM_001190882.2		3-13	Meier-Gorlin syndrome 2 OMIM
ORC6	17151	NM_014321.3		1-7	Meier-Gorlin syndrome 3 OMIM
OSTM1	21652	NM_014028.3		1-6	Osteopetrosis, autosomal recessive 5 OMIM
P4HB	8548	NM_000918.3		1-11	Cole-Carpenter syndrome 1 OMIM
PAM16	29679	NM_016069.9		1-5	Spondylometaphyseal dysplasia, Megarbane-Dagher-Melike type OMIM
PAPSS2	8604	NM_004670.3		1-12	Brachyolmia 4 with mild epiphyseal and metaphyseal changes OMIM
PAX3	8617	NM_181461.3		1-8	Craniofacial-deafness-hand syndrome OMIM Waardenburg syndrome, type 1 OMIM Waardenburg syndrome, type 3 OMIM
PCNT	16068	NM_006031.5		1-47	Microcephalic osteodysplastic primordial dwarfism, type II OMIM
PCYT1A	8754	NM_005017.4		3-10	Spondylometaphyseal dysplasia with cone-rod dystrophy OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
PDE3A	8778	NM_001244683.1		1-15	Hypertension and brachydactyly syndrome OMIM
PDE4D	8783	NM_001165899.2		2-17	Acrodysostosis 2, with or without hormone resistance OMIM
PEX5	9719	NM_001131026.1		3-17	Peroxisome biogenesis disorder 2A (Zellweger) OMIM Peroxisome biogenesis disorder 2B OMIM Rhizomelic chondrodysplasia punctata, type 5 OMIM
PEX7	8860	NM_000288.4		1-10	Peroxisome biogenesis disorder 9B OMIM Rhizomelic chondrodysplasia punctata, type 1 OMIM
PGM3	8907	NM_001199918.1		2-12	Immunodeficiency 23 OMIM
PHEX	8918	NM_000444.5		1-22	Hypophosphatemic rickets, X-linked dominant OMIM
PHGDH	8923	NM_006623.4		1-12	Neu-Laxova syndrome 1 OMIM Phosphoglycerate dehydrogenase deficiency OMIM
PIGT	14938	NM_015937.5		1-12	Multiple congenital anomalies-hypotonia-seizures syndrome 3 OMIM
PIGV	26031	NM_017837.3		2-4	Hyperphosphatasia with mental retardation syndrome 1 OMIM
PIK3C2A	8971	NM_002645.2		1-32	Oculoskeletodental syndrome OMIM
PIK3R1	8979	NM_181523.3		2-16	Immunodeficiency 36 OMIM SHORT syndrome OMIM
PITX1	9004	NM_002653.4		1-3	Clubfoot, congenital, with or without deficiency of long bones and/or mirror-image polydactyly OMIM Liebenberg syndrome OMIM
PLOD2	9082	NM_182943.3		1-20	Bruck syndrome 2 OMIM

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PLS3	9091	NM_005032.7		2-16	Bone mineral density QTL18, osteoporosis OMIM
POC1A	24488	NM_001161580.1		1-10	Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis OMIM
POLR1A	17264	NM_015425.3		1-34	Acrofacial dysostosis, Cincinnati type OMIM
POLR1C	20194	NM_203290.3		1-9	Leukodystrophy, hypomyelinating, 11 OMIM Treacher Collins syndrome 3 OMIM
POLR1D	20422	NM_152705.3		1-3	Treacher Collins syndrome 2 OMIM
POP1	30129	NM_001145860.2		2-16	Anauxetic dysplasia 2 OMIM
POR	9208	NM_000941.2		2-16	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis OMIM
PPIB	9255	NM_000942.4		1-5	Osteogenesis imperfecta, type IX OMIM
PRKAR1A	9388	NM_001276290.1		1-10	Acrodysostosis 1, with or without hormone resistance OMIM Pigmented nodular adrenocortical disease, primary, 1 OMIM
PRMT7	25557	NM_019023.2		3-19	Short stature, brachydactyly, intellectual developmental disability, and seizures OMIM
PSAT1	19129	NM_058179.4	9	1-9	Neu-Laxova syndrome 2 OMIM
PSPH	9577	NM_004577.3	8	8,4-7	Phosphoserine phosphatase deficiency OMIM
PTDSS1	9587	NM_014754.3		1-13	Lenz-Majewski hyperostotic dwarfism OMIM

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PTH1R	9608	NM_000316.2		3-16	Chondrodysplasia, Blomstrand type OMIM Eiken syndrome OMIM Failure of tooth eruption, primary OMIM Metaphyseal chondrodysplasia, Murk Jansen type OMIM
PTHLH	9607	NM_198965.1		3-5	Brachydactyly, type E2 OMIM
PTPN11	9644	NM_002834.4		1-15	LEOPARD syndrome 1 OMIM Metachondromatosis OMIM Noonan syndrome 1 OMIM
PUF60	17042	NM_001271098.1		1-12	Verheij syndrome OMIM
PYCR1	9721	NM_001282279.1		2-7	Cutis laxa, autosomal recessive, type IIB OMIM Cutis laxa, autosomal recessive, type IIIB OMIM
RAB23	14263	NM_016277.5		2-7	Carpenter syndrome OMIM
RAB33B	16075	NM_031296.3		1-2	Smith-McCort dysplasia 2 OMIM
RAD21	9811	NM_006265.2	14	2-14	Cornelia de Lange syndrome 4 OMIM
RASGRP2	9879	NM_153819.1		2-16	?Bleeding disorder, platelet-type, 18 OMIM
RBM8A	9905	NM_005105.4	1-6	1-6	Thrombocytopenia-absent radius syndrome OMIM
RBPJ	5724	NM_005349.3	10-12	2-12	Adams-Oliver syndrome 3 OMIM
RECQL4	9949	NM_004260.3		1-22	Baller-Gerold syndrome OMIM RAPADILINO syndrome OMIM Rothmund-Thomson syndrome, type 2 OMIM

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RFT1	30220	NM_052859.3		1-13	Congenital disorder of glycosylation, type In OMIM
ROR2	10257	NM_004560.3		1-9	Brachydactyly, type B1 OMIM Robinow syndrome, autosomal recessive OMIM
RPGRIP1L	29168	NM_001127897.4		2-25	Joubert syndrome 7 OMIM Meckel syndrome 5 OMIM
RPL13	10303	NM_001243130.1		2-6	Spondyloepimetaphyseal dysplasia, Isidor-Toutain type OMIM
RUNX2	10472	NM_001024630.4		2-9	Cleidocranial dysplasia OMIM Cleidocranial dysplasia, forme fruste, dental anomalies only OMIM Cleidocranial dysplasia, forme fruste, with brachydactyly OMIM Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly OMIM
SALL1	10524	NM_002968.2	2-3	1-3	Townes-Brocks branchiootorenal-like syndrome OMIM Townes-Brocks syndrome 1 OMIM
SALL4	15924	NM_020436.3		1-4	?IVIC syndrome OMIM Duane-radial ray syndrome OMIM
SBDS	19440	NM_016038.4	1-5	1-5	Shwachman-Diamond syndrome OMIM
SCARF2	19869	NM_182895.5		1-14	Van den Ende-Gupta syndrome OMIM
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_001207014.1		3-6	Osteogenesis imperfecta, type X OMIM
SETD2	18420	NM_014159.6		1-21	Luscan-Lumish syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
SF3B4	10771	NM_005850.4		1-6	Acrofacial dysostosis 1, Nager type OMIM
SFRP4	10778	NM_003014.3		1-6	Pyle disease OMIM
SGSH	10818	NM_000199.4		1-8	Mucopolysaccharidosis type IIIA (Sanfilippo A) OMIM
SH3BP2	10825	NM_001122681.1		2-13	Cherubism OMIM
SH3PXD2B	29242	NM_001017995.2		1-13	Frank-ter Haar syndrome OMIM
SHOX	10853	NM_006883.2	2-6	2-6	Langer mesomelic dysplasia OMIM Leri-Weill dyschondrosteosis OMIM Short stature, idiopathic familial OMIM
SKI	10896	NM_003036.3		1-7	Shprintzen-Goldberg syndrome OMIM
SLC10A7	23088	NM_001029998.6		1-12	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis OMIM
SLC17A5	10933	NM_012434.4		1-11	Salla disease OMIM Sialic acid storage disorder, infantile OMIM
SLC26A2	10994	NM_000112.3		2-3	Achondrogenesis Ib OMIM Atelosteogenesis, type II OMIM De la Chapelle dysplasia OMIM Diastrophic dysplasia OMIM Diastrophic dysplasia, broad bone-platyspondylic variant OMIM Epiphyseal dysplasia, multiple, 4 OMIM
SLC29A3	23096	NM_018344.6		1-6	Histiocytosis-lymphadenopathy plus syndrome OMIM
SLC34A1	11019	NM_003052.5		2-13	?Fanconi renotubular syndrome 2 OMIM Hypercalcemia, infantile, 2 OMIM Nephrolithiasis/osteoporosis, hypophosphatemic, 1 OMIM

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SLC34A3	20305	NM_001177317.1		2-13	Hypophosphatemic rickets with hypercalciuria OMIM
SLC35C1	20197	NM_001145266.1		2-3	Congenital disorder of glycosylation, type IIc OMIM
SLC35D1	20800	NM_015139.3		1-12	Schneckenbecken dysplasia OMIM
SLC39A13	20859	NM_001128225.3		2-10	Ehlers-Danlos syndrome, spondylodysplastic type, 3 OMIM
SLCO2A1	10955	NM_005630.2		1-14	Hypertrophic osteoarthropathy, primary, autosomal dominant OMIM Hypertrophic osteoarthropathy, primary, autosomal recessive 2 OMIM
SMAD3	6769	NM_005902.3		1-9	Loeys-Dietz syndrome 3 OMIM
SMAD4	6770	NM_005359.5		2-12	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome OMIM Myhre syndrome OMIM
SMARCAL1	11102	NM_014140.3		3-18	Schimke immunoosseous dysplasia OMIM
SMC1A	11111	NM_001281463.1		2-26	Cornelia de Lange syndrome 2 OMIM Developmental and epileptic encephalopathy 85, with or without midline brain defects OMIM
SMC3	2468	NM_005445.3		1-29	Cornelia de Lange syndrome 3 OMIM
SMOC1	20318	NM_022137.6		1-12	Microphthalmia with limb anomalies OMIM
SNRPB	11153	NM_003091.3		1-7	Cerebrocostomandibular syndrome OMIM
SNX10	14974	NM_013322.2		2-7	Osteopetrosis, autosomal recessive 8 OMIM
SOST	13771	NM_025237.2		1-2	Craniodiaphyseal dysplasia, autosomal dominant OMIM Sclerosteosis 1 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
SOX9	11204	NM_000346.3		1-3	Acampomelic campomelic dysplasia OMIM Campomelic dysplasia OMIM Campomelic dysplasia with autosomal sex reversal 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
SUMF1	20376	NM_182760.3		1-9	Multiple sulfatase deficiency OMIM
TALDO1	11559	NM_006755.1		1-8	Transaldolase deficiency OMIM
TAPT1	26887	NM_153365.2		1-14	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type OMIM
TBCE	11582	NM_001079515.2		2-17	Hypoparathyroidism-retardation-dysmorphism syndrome OMIM Kenny-Caffey syndrome, type 1 OMIM
TBX15	11594	NM_152380.2		2-8	Cousin syndrome OMIM
TBX3	11602	NM_005996.3		1-7	Ulnar-mammary syndrome OMIM
TBX4	11603	NM_018488.2		1-8	Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome OMIM Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension OMIM
TBX5	11604	NM_000192.3		2-9	Holt-Oram syndrome OMIM
TBX6	11605	NM_004608.3		2-9	Spondylocostal dysostosis 5 OMIM
TBXAS1	11609	NM_001130966.2		5-17	Ghosal hematodiaphyseal syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
TCIRG1	11647	NM_006019.4		2-20	Osteopetrosis, autosomal recessive 1 OMIM
TCOF1	11654	NM_001135243.1		1-26	Treacher Collins syndrome 1 OMIM
TCTEX1D2	28482	NM_152773.4		1-5	Short-rib thoracic dysplasia 17 with or without polydactyly OMIM
TCTN2	25774	NM_024809.4		1-18	?Meckel syndrome 8 OMIM Joubert syndrome 24 OMIM
TCTN3	24519	NM_015631.5		1-14	Joubert syndrome 18 OMIM Orofaciodigital syndrome IV OMIM
TERT	11730	NM_198253.2		1-16	{Dyskeratosis congenita, autosomal dominant 2} OMIM {Dyskeratosis congenita, autosomal recessive 4} OMIM
TGFB1	11766	NM_000660.7		1-7	Camurati-Engelmann disease OMIM
TGFB2	11768	NM_003238.6		1-7	Loeys-Dietz syndrome 4 OMIM
TGFBR2	11773	NM_003242.5		1-7	Loeys-Dietz syndrome 2 OMIM
TMCO1	18188	NM_019026.4		1-7	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome OMIM
TMEM165	30760	NM_018475.5		1-6	Congenital disorder of glycosylation, type IIk OMIM
TMEM216	25018	NM_016499.5		3-5	Joubert syndrome 2 OMIM Meckel syndrome 2 OMIM
TMEM231	37234	NM_001077418.2		1-7	Joubert syndrome 20 OMIM Meckel syndrome 11 OMIM
TMEM38B	25535	NM_018112.2		1-6	Osteogenesis imperfecta, type XIV OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
TNFRSF11A	11908	NM_001278268.2		1-10	Osteolysis, familial expansile OMIM Osteopetrosis, autosomal recessive 7 OMIM {Paget disease of bone 2, early-onset} OMIM
TNFRSF11B	11909	NM_002546.3		1-5	Paget disease of bone 5, juvenile-onset OMIM
TNFSF11	11926	NM_033012.3		4-7	Osteopetrosis, autosomal recessive 2 OMIM
TP63	15979	NM_003722.4		1-14	ADULT syndrome OMIM Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3 OMIM Hay-Wells syndrome OMIM Limb-mammary syndrome OMIM Orofacial cleft 8 OMIM Rapp-Hodgkin syndrome OMIM Split-hand/foot malformation 4 OMIM
TRAPPC2	23068	NM_001011658.3	6	3-6	Spondyloepiphyseal dysplasia tarda OMIM
TREM2	17761	NM_001271821.1		1-4	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 2 OMIM
TRIP11	12305	NM_004239.4	21	1-21	Achondrogenesis, type IA OMIM Odontochondrodysplasia 1 OMIM
TRPS1	12340	NM_014112.5		2-7	Trichorhinophalangeal syndrome, type I OMIM Trichorhinophalangeal syndrome, type III OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
TRPV4	18083	NM_021625.5		2-16	?Avascular necrosis of femoral head, primary, 2 OMIM Brachyolmia type 3 OMIM Digital arthropathy-brachydactyly, familial OMIM Metatropic dysplasia OMIM Parastremmatic dwarfism OMIM Scapuloperoneal spinal muscular atrophy OMIM SED, Maroteaux type OMIM Spondylometaphyseal dysplasia, Kozlowski type OMIM
TRPV6	14006	NM_018646.5		1-15	Hyperparathyroidism, transient neonatal OMIM
TTC21B	25660	NM_024753.4		1-29	Short-rib thoracic dysplasia 4 with or without polydactyly OMIM
TTC8	20087	NM_198309.3		2-15	Bardet-Biedl syndrome 8 OMIM
TWIST1	12428	NM_000474.3		1	Craniosynostosis 1 OMIM Robinow-Sorauf syndrome OMIM Saethre-Chotzen syndrome with or without eyelid anomalies OMIM Sweeney-Cox syndrome OMIM
TYROBP	12449	NM_001173515.2		1-4	Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy 1 OMIM
VDR	12679	NM_001017535.1		4-11	Rickets, vitamin D-resistant, type IIA OMIM
WDPCP	28027	NM_015910.5		1-18	?Bardet-Biedl syndrome 15 OMIM Congenital heart defects, hamartomas of tongue, and polysyndactyly OMIM
WDR19	18340	NM_025132.3		1-36	?Cranioectodermal dysplasia 4 OMIM ?Short-rib thoracic dysplasia 5 with or without polydactyly OMIM Nephronophthisis 13 OMIM
WDR34	28296	NM_052844.3		1-9	Short-rib thoracic dysplasia 11 with or without polydactyly OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
WDR35	29250	NM_001006657.1		1-28	Cranioectodermal dysplasia 2 OMIM Short-rib thoracic dysplasia 7 with or without polydactyly OMIM
WDR60	21862	NM_018051.4		1-25	Short-rib thoracic dysplasia 8 with or without polydactyly OMIM
WISP3	12771	NM_198239.2		1-5	Progressive pseudorheumatoid dysplasia OMIM
WNT1	12774	NM_005430.3		1-4	Osteogenesis imperfecta, type XV OMIM
WNT10B	12775	NM_003394.4		2-5	Split-hand/foot malformation 6 OMIM Tooth agenesis, selective, 8 OMIM
WNT5A	12784	NM_003392.4		1-5	Robinow syndrome, autosomal dominant 1 OMIM
WNT7A	12786	NM_004625.4		1-4	Fuhrmann syndrome OMIM Ulna and fibula, absence of, with severe limb deficiency OMIM
XRCC4	12831	NM_003401.3		2-8	Short stature, microcephaly, and endocrine dysfunction OMIM
XYLT1	15516	NM_022166.4		1-12	Desbuquois dysplasia 2 OMIM
XYLT2	15517	NM_022167.4		1-11	Spondyloocular syndrome OMIM
YY1	12856	NM_003403.5		1-5	Gabriele-de Vries syndrome OMIM
ZMPSTE24	12877	NM_005857.4		1-10	Mandibuloacral dysplasia with type B lipodystrophy OMIM Restrictive dermopathy, lethal OMIM
ZSWIM6	29316	NM_020928.1		1-14	Acromelic frontonasal dysostosis OMIM Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features OMIM