Oslo universitetssykehus
Avdeling for medisinsk genetikk

Kraniofaciale malformasjoner

Genpanel, versjon v02

* 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: **UBB**

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, childhood OMIM
ALX3	449	NM_006492.2		1-4	Frontonasal dysplasia 1 OMIM
ALX4	450	NM_021926.3		1-4	{Craniosynostosis 5, susceptibility to} OMIM Parietal foramina 2 OMIM Frontonasal dysplasia 2 OMIM
ASXL1	18318	NM_015338.5		1-12	Bohring-Opitz syndrome OMIM
ATR	882	NM_001184.3		1-47	Seckel syndrome 1 OMIM
BMP4	1071	NM_001202.3		3-4	Orofacial cleft 11 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CCBE1	29426	NM_133459.3		1-11	Hennekam lymphangiectasia-lymphedema syndrome 1 OMIM
CDC45	1739	NM_001178010.2		1-19	Meier-Gorlin syndrome 7 OMIM
CEP120	26690	NM_153223.3		2-21	Short-rib thoracic dysplasia 13 with or without polydactyly OMIM
CHD7	20626	NM_017780.3		2-38	CHARGE syndrome OMIM
COL11A1	2186	NM_001854.3		1-67	Marshall syndrome OMIM Fibrochondrogenesis 1 OMIM Stickler syndrome, type II OMIM
COL11A2	2187	NM_080680.2		1-66	Fibrochondrogenesis 2 OMIM Weissenbacher-Zweymuller syndrome OMIM Stickler syndrome, type III OMIM Otospondylomegaepiphyseal dysplasia OMIM
COL2A1	2200	NM_001844.4		1-54	Achondrogenesis, type II or hypochondrogenesis OMIM SMED Strudwick type OMIM Kniest dysplasia OMIM SED congenita OMIM Stickler syndrome, type I OMIM Stickler sydrome, type I, nonsyndromic ocular OMIM Otospondylomegaepiphyseal dysplasia OMIM
COL9A1	2217	NM_001851.4		1-38	Stickler syndrome, type IV OMIM
COL9A2	2218	NM_001852.3		1-32	?Stickler syndrome, type V OMIM
COLEC11	17213	NM_024027.4		2-7	3MC syndrome 2 OMIM
CTSK	2536	NM_000396.3		2-8	Pycnodysostosis OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CYP26B1	20581	NM_019885.3		1-6	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies OMIM
DHODH	2867	NM_001361.4		1-9	Miller syndrome OMIM
DPH1	3003	NM_001383.3		1-12	Developmental delay with short stature, dysmorphic features, and sparse hair OMIM
EDN1	3176	NM_001955.4		1-5	Question mark ears, isolated OMIM Auriculocondylar syndrome 3 OMIM
EDNRA	3179	NM_001957.3		2-8	Mandibulofacial dysostosis with alopecia OMIM
EFNA4	3224	NM_005227.2		1-4	Coronal craniosynostosis PubMed
EFNB1	3226	NM_004429.4		1-5	Craniofrontonasal dysplasia OMIM
EFTUD2	30858	NM_004247.3		2-28	Mandibulofacial dysostosis, Guion-Almeida type OMIM
EIF4A3	18683	NM_014740.3		1-12	Robin sequence with cleft mandible and limb anomalies OMIM
ERF	3444	NM_006494.3		1-4	Craniosynostosis 4 OMIM
ESCO2	27230	NM_001017420.2		2-11	Roberts syndrome OMIM
EYA1	3519	NM_000503.5		3-18	?Otofaciocervical syndrome OMIM Branchiootorenal syndrome 1, with or without cataracts OMIM Branchiootic syndrome 1 OMIM Anterior segment anomalies with or without cataract OMIM
FAM20C	22140	NM_020223.3		1-10	Raine syndrome OMIM
FAM58A	28434	NM_152274.4		1-5	STAR syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
FBN1	3603	NM_000138.4		2-66	Weill-Marchesani syndrome 2, dominant OMIM Marfan syndrome OMIM
FGFR1	3688	NM_023110.2		2-18	Trigonocephaly 1 OMIM Encephalocraniocutaneous lipomatosis OMIM Pfeiffer syndrome OMIM Osteoglophonic dysplasia OMIM Jackson-Weiss syndrome OMIM Hartsfield syndrome OMIM
FGFR2	3689	NM_000141.4		2-18	Scaphocephaly, maxillary retrusion, and mental retardation OMIM Beare-Stevenson cutis gyrata syndrome OMIM Scaphocephaly and Axenfeld-Rieger anomaly PubMed Apert syndrome OMIM Crouzon syndrome OMIM Saethre-Chotzen syndrome OMIM Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis OMIM Craniosynostosis, nonspecific PubMed Pfeiffer syndrome OMIM Craniofacial-skeletal-dermatologic dysplasia OMIM LADD syndrome OMIM Jackson-Weiss syndrome OMIM
FGFR3	3690	NM_000142.4		2-18	Muenke syndrome OMIM Crouzon syndrome with acanthosis nigricans OMIM
FREM1	23399	NM_144966.5		3-38	Trigonocephaly 2 OMIM
GLI3	4319	NM_000168.5		2-15	Greig cephalopolysyndactyly syndrome OMIM
GNAI3	4387	NM_006496.3		1-8	Auriculocondylar syndrome 1 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
GNAS	4392	NM_000516.4		1-13	Pseudohypoparathyroidism Ic OMIM Pseudohypoparathyroidism Ib OMIM Pseudohypoparathyroidism Ia OMIM Osseous heteroplasia, progressive OMIM Pseudopseudohypoparathyroidism OMIM
GPC3	4451	NM_004484.3		1-8	Simpson-Golabi-Behmel syndrome, type 1 OMIM
GRHL3	25839	NM_198174.2		1-16	Van der Woude syndrome 2 OMIM
GSC	4612	NM_173849.2		1-3	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities OMIM
GTF2E2	4651	NM_002095.4		2-8	Trichothiodystrophy 6, nonphotosensitive OMIM
HNRNPK	5044	NM_031262.2	17	3-17	Au-Kline syndrome OMIM
HUWE1	30892	NM_031407.6		4-84	Mental retardation, X-linked syndromic, Turner type OMIM
IDS	5389	NM_000202.6	2-3	1-9	Mucopolysaccharidosis II OMIM
IDUA	5391	NM_000203.4		1-14	Mucopolysaccharidosis Ih/s OMIM Mucopolysaccharidosis Ih OMIM Mucopolysaccharidosis Is OMIM
IFT122	13556	NM_052985.3	15-20	1-31	Cranioectodermal dysplasia 1 OMIM
IFT140	29077	NM_014714.3		3-31	Short-rib thoracic dysplasia 9 with or without polydactyly OMIM
IFT43	29669	NM_052873.2		1-8	Cranioectodermal dysplasia 3 OMIM
IHH	5956	NM_002181.3		1-3	Acrocapitofemoral dysplasia OMIM
IL11RA	5967	NM_001142784.2		2-13	Craniosynostosis and dental anomalies OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
IMPAD1	26019	NM_017813.4		1-5	Chondrodysplasia with joint dislocations, GPAPP type OMIM
IRF6	6121	NM_006147.3		3-9	Popliteal pterygium syndrome 1 OMIM van der Woude syndrome OMIM
IRX5	14361	NM_005853.5		1-3	Hamamy syndrome OMIM
JAG1	6188	NM_000214.2		1-26	Alagille syndrome 1 OMIM
KAT6A	13013	NM_006766.3		2-17	Mental retardation, autosomal dominant 32 OMIM
KMT2D	7133	NM_003482.3		1-54	Kabuki syndrome 1 OMIM
KRAS	6407	NM_004985.4	5	2-5	Cardiofaciocutaneous syndrome 2 OMIM Noonan syndrome 3 OMIM Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic OMIM
LMX1B	6654	NM_002316.3		1-8	Nail-patella syndrome OMIM
LRP5	6697	NM_002335.3	1, 3-9	1-23	van Buchem disease, type 2 OMIM Osteosclerosis OMIM Osteoporosis-pseudoglioma syndrome OMIM Osteopetrosis, autosomal dominant 1 OMIM
MASP1	6901	NM_139125.3		1-11	3MC syndrome 1 OMIM
MEGF8	3233	NM_001410.2		1-41	Carpenter syndrome 2 OMIM
MSX1	7391	NM_002448.3		1-2	Orofacial cleft 5 OMIM Ectodermal dysplasia 3, Witkop type OMIM
MSX2	7392	NM_002449.4	2	1-2	Craniosynostosis 2 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
MYH3	7573	NM_002470.3		3-41	Arthrogryposis, distal, type 8 OMIM
P4HB	8548	NM_000918.3		1-11	Cole-Carpenter syndrome 1 OMIM
PAX1	8615	NM_006192.4		1-5	?Otofaciocervical syndrome 2 OMIM
PHEX	8918	NM_000444.5		1-22	Hypophosphatemic rickets, X-linked dominant OMIM
PLCB4	9059	NM_000933.3		1-36	Auriculocondylar syndrome 2 OMIM
POLR1A	17264	NM_015425.3		1-34	Acrofacial dysostosis, Cincinnati type OMIM
POLR1C	20194	NM_203290.2		1-9	Treacher Collins syndrome 3 OMIM Leukodystrophy, hypomyelinating, 11 OMIM
POLR1D	20422	NM_015972.3		1-2	Treacher Collins syndrome 2 OMIM
POR	9208	NM_000941.2		2-16	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis OMIM
PVRL1	9706	NM_002855.4		1-6	Orofacial cleft 7 OMIM Cleft lip/palate-ectodermal dysplasia syndrome OMIM
RAB23	14263	NM_183227.2		2-7	Carpenter syndrome OMIM
RECQL4	9949	NM_004260.3		1-22	RAPADILINO syndrome OMIM Baller-Gerold syndrome OMIM
RSPRY1	29420	NM_133368.2		2-15	Spondylopeimetaphyseal dsyplasia, Faden-Alkuraya type OMIM
RUNX2	10472	NM_001024630.3		2-9	Cleidocranial dysplasia OMIM Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
SALL1	10524	NM_002968.2	2-3	1-3	Townes-Brocks syndrome OMIM Townes-Brocks branchiootorenal-like syndrome OMIM
SALL4	15924	NM_020436.3		1-4	Duane-radial ray syndrome OMIM IVIC syndrome OMIM
SCARF2	19869	NM_153334.6		1-12	Van den Ende-Gupta syndrome OMIM
SEMA3E	10727	NM_012431.2		1-17	?CHARGE syndrome OMIM
SF3B4	10771	NM_005850.4		1-6	Acrofacial dysostosis 1, Nager type OMIM
SH3PXD2B	29242	NM_001017995.2		1-13	Frank-ter Haar syndrome OMIM
SIX1	10887	NM_005982.3		1-2	Branchiootic syndrome 3 OMIM
SIX5	10891	NM_175875.4		1-3	Branchiootorenal syndrome 2 OMIM
SKI	10896	NM_003036.3		1-7	Shprintzen-Goldberg syndrome OMIM
SMO	11119	NM_005631.4		1-12	Curry-Jones syndrome, somatic mosaic OMIM PubMed
SNRPB	11153	NM_003091.3		1-7	Cerebrocostomandibular syndrome OMIM
SON	11183	NM_032195.2		1-7	ZTTK syndrome OMIM
SPECC1L	29022	NM_015330.4	4	3-17	Opitz GBBB syndrome, type II OMIM
STAT3	11364	NM_139276.2		2-24	Hyper-IgE recurrent infection syndrome OMIM Autoimmune disease, multisystem, infantile-onset, 1 OMIM
SUMO1	12502	NM_003352.4		1-5	Orofacial cleft 10 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
TBX1	11592	NM_080647.1		2-9	Velocardiofacial syndrome OMIM DiGeorge syndrome OMIM
TCF12	11623	NM_207036.1		2-20	Craniosynostosis 3 OMIM
TCOF1	11654	NM_001135243.1		1-26	Treacher Collins syndrome 1 OMIM
TGDS	20324	NM_014305.3		1-12	Catel-Manzke syndrome OMIM
TGFBR1	11772	NM_004612.2		1-9	Loeys-Dietz syndrome 1 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
TP63	15979	NM_003722.4		1-14	Orofacial cleft 8 OMIM Limb-mammary syndrome OMIM Hay-Wells syndrome OMIM Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3 OMIM Split-hand/foot malformation 4 OMIM ADULT syndrome OMIM Rapp-Hodgkin syndrome OMIM
TWIST1	12428	NM_000474.3		1	Craniosynostosis 1 OMIM Robinow-Sorauf syndrome OMIM Saethre-Chotzen syndrome OMIM Saethre-Chotzen syndrome with eyelid anomalies OMIM
TXNL4A	30551	NM_006701.3		1-3	Burn-McKeown syndrome OMIM
UBB	12463	NM_001281720.1	2	2	Cleft palate, isolated OMIM
WDR19	18340	NM_025132.3		1-36	Senior-Loken syndrome 8 OMIM ?Short-rib thoracic dysplasia 5 with or without polydactyly OMIM ?Cranioectodermal dysplasia 4 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
ZEB2	14881	NM_014795.3	10	2-10	Mowat-Wilson syndrome OMIM
ZIC1	12872	NM_003412.3		1-3	Craniosynostosis 6 OMIM