Kraniofaciale malformasjoner

Genpanel, versjon v01

Tabellen er sortert på gennavn (HGNC gensymbol)
Navn på gen er iht. HGNC
Kolonnen **>x10** viser andel av genet som vi forventer blir lest med tilfredstillende kvalitet flere enn 10 ganger under sekvensering

Gen	Transkript	>10x	Fenotype
ASXL1	NM_015338.5	100%	Bohring-Opitz syndrome OMIM
BMP4	NM_001202.3	100%	Orofacial cleft 11 OMIM Microphthalmia, syndromic 6 OMIM
CDC45	NM_001178010.2	100%	Saethre-Chotzen syndrome PubMed
CHD7	NM_017780.3	100%	CHARGE syndrome OMIM
COL11A1	NM_001854.3	99%	Stickler syndrome, type II OMIM Marshall syndrome OMIM
COL11A2	NM_080680.2	100%	Weissenbacher-Zweymuller syndrome OMIM Stickler syndrome, type III OMIM
COL2A1	NM_001844.4	100%	Kniest dysplasia OMIM Stickler syndrome, type I OMIM Stickler sydrome, type I, nonsyndromic ocular OMIM Otospondylomegaepiphyseal dysplasia OMIM Spondyloperipheral dysplasia OMIM
COL9A1	NM_001851.4	100%	Stickler syndrome, type IV OMIM
COL9A2	NM_001852.3	100%	?Stickler syndrome, type V OMIM
COLEC11	NM_024027.4	100%	3MC syndrome 2 OMIM
DHODH	NM_001361.4	100%	Miller syndrome OMIM
EDN1	NM_001955.4	100%	Auriculocondylar syndrome 3 OMIM
EDNRA	NM_001957.3	100%	Mandibulofacial dysostosis with alopecia OMIM
EFNB1	NM_004429.4	100%	Craniofrontonasal dysplasia OMIM

Gen	Transkript	>10x	Fenotype
EFTUD2	NM_004247.3	100%	Mandibulofacial dysostosis, Guion-Almeida type OMIM
EIF4A3	NM_014740.3	100%	Robin sequence with cleft mandible and limb anomalies OMIM
ERF	NM_006494.3	100%	Craniosynostosis 4 OMIM
EYA1	NM_000503.5	100%	Anterior segment anomalies with or without cataract OMIM ?Otofaciocervical syndrome OMIM Branchiootorenal syndrome 1, with or without cataracts OMIM
FGFR1	NM_023110.2	100%	Hartsfield syndrome OMIM Pfeiffer syndrome OMIM Jackson-Weiss syndrome OMIM
FGFR2	NM_000141.4	100%	Crouzon syndrome OMIM Saethre-Chotzen syndrome OMIM Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis OMIM Pfeiffer syndrome OMIM Craniofacial-skeletal-dermatologic dysplasia OMIM LADD syndrome OMIM Bent bone dysplasia syndrome OMIM Jackson-Weiss syndrome OMIM Scaphocephaly, maxillary retrusion, and mental retardation OMIM Beare-Stevenson cutis gyrata syndrome OMIM Apert syndrome OMIM
FGFR3	NM_000142.4	100%	Muenke syndrome OMIM LADD syndrome OMIM Crouzon syndrome with acanthosis nigricans OMIM
GNAI3	NM_006496.3	100%	Auriculocondylar syndrome 1 OMIM
GRHL3	NM_198174.2	100%	Van der Woude syndrome 2 OMIM
GSC	NM_173849.2	100%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities OMIM
IHH	NM_002181.3	100%	Acrocapitofemoral dysplasia OMIM
IL11RA	NM_001142784.2	100%	Craniosynostosis and dental anomalies OMIM
IRF6	NM_006147.3	100%	van der Woude syndrome OMIM
MEGF8	NM_001410.2	100%	Carpenter syndrome 2 OMIM
MSX1	NM_002448.3	100%	Orofacial cleft 5 OMIM

Gen	Transkript	>10x	Fenotype
MSX2	NM_002449.4	100%	Craniosynostosis, type 2 OMIM Parietal foramina with cleidocranial dysplasia OMIM
PAX1	NM_006192.4	98%	?Otofaciocervical syndrome 2 OMIM
PLCB4	NM_000933.3	100%	Auriculocondylar syndrome 2 OMIM
POLR1A	NM_015425.3	100%	Acrofacial dysostosis, Cincinnati type OMIM
POLR1C	NM_203290.2	100%	Treacher Collins syndrome 3 OMIM
POLR1D	NM_015972.3	100%	Treacher Collins syndrome 2 OMIM
POR	NM_000941.2	100%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis OMIM
PVRL1	NM_002855.4	100%	Orofacial cleft 7 OMIM Cleft lip/palate-ectodermal dysplasia syndrome OMIM
RAB23	NM_183227.2	100%	Carpenter syndrome OMIM
RUNX2	NM_001024630.3	100%	Cleidocranial dysplasia OMIM Cleidocranial dysplasia, forme fruste, with brachydactyly OMIM Cleidocranial dysplasia, forme fruste, dental anomalies only OMIM
SALL1	NM_002968.2	100%	Townes-Brocks syndrome OMIM Townes-Brocks branchiootorenal-like syndrome OMIM
SALL4	NM_020436.3	99%	Duane-radial ray syndrome OMIM
SEMA3E	NM_012431.2	100%	?CHARGE syndrome OMIM
SF3B4	NM_005850.4	100%	Acrofacial dysostosis 1, Nager type OMIM
SIX1	NM_005982.3	100%	Brachiootic syndrome 3 OMIM
SIX5	NM_175875.4	100%	Branchiootorenal syndrome 2 OMIM
SKI	NM_003036.3	100%	Shprintzen-Goldberg syndrome OMIM
SNRPB	NM_003091.3	100%	Cerebrocostomandibular syndrome OMIM

Gen	Transkript	>10x	Fenotype
SUMO1	NM_003352.4	96%	Orofacial cleft 10 OMIM
TBX1	NM_080647.1	86%	Velocardiofacial syndrome OMIM
TCF12	NM_207036.1	100%	Craniosynostosis 3 OMIM
TCOF1	NM_001135243.1	100%	Treacher Collins syndrome 1 OMIM
TGDS	NM_014305.3	100%	Catel-Manzke syndrome OMIM
TP63	NM_003722.4	100%	Rapp-Hodgkin syndrome OMIM Orofacial cleft 8 OMIM Hay-Wells syndrome OMIM Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3 OMIM
TWIST1	NM_000474.3	100%	Craniosynostosis, type 1 OMIM Saethre-Chotzen syndrome with eyelid anomalies OMIM Saethre-Chotzen syndrome OMIM
TXNL4A	NM_006701.3	100%	Burn-McKeown syndrome OMIM
UBB	NM_001281720.1	82%	Cleft palate, isolated OMIM
WDR35	NM_001006657.1	99%	Cranioectodermal dysplasia 2 OMIM
ZIC1	NM_003412.3	100%	Craniosynostosis 6 OMIM