CRANIOFACIAL ANOMALIES GENE PANEL DG 2.4.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM ID*
ADAMTSL4	97.2	100%	98%	Ectopia lentis et pupillae,225200 Ectopia lentis,isolated,autosomal recessive,225100
ALX1	177.1	100%	100%	Frontonasal dysplasia 3, 613456
ALX3	82.1	89%	82%	Frontonasal dysplasia 1, 136760
ALX4	87.2	100%	100%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451 {Craniosynostosis 5, susceptibility to},615529
AMELX	60.1	100%	99%	Amelogenesis imperfecta, hypoplastic/hypomaturation type 1E, 301200
ANKRD11	117.5	91%	87%	KBG syndrome, 148050
AXIN2	112.6	99%	96%	Oligodontia-colorectal cancer syndrome, 608615 Colorectal cancer, somatic, 114500
BCOR	68.5	98%	94%	Microphthalmia, syndromic 2, 300166
BMP4	124.6	100%	100%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C4orf26	174.3	100%	100%	Amelogenesis imperfecta, hypomaturation type, IIA4, 614832
CDON	122.6	100%	99%	Holoprosencephaly 11, 614226
CHD7	127.8	99%	99%	CHARGE syndrome,214800 Hypogonadotropic hypogonadism 5 with or without anosmia,612370
COL11A1	98.8	98%	98%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	14.4	58%	21%	Stickler syndrome, type III, 184840 Otospondylomegaepiphyseal dysplasia, 215150 Weissenbacher-Zweymuller syndrome, 277610 Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524

COL2A1	93.2	99%	95%	Stickler syndrome, type I, 108300 Kniest dysplasia, 156550 Achondrogenesis, type II or hypochondrogenesis, 200610 SED congenita, 183900 SMED Strudwick type, 184250 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Spondyloperiph
COL9A1	112.8	98%	96%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	77.8	98%	94%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
CTSK	138.3	100%	100%	Pycnodysostosis, 265800
DISP1	172.9	100%	100%	Craniofacial and neuro-developmental abnormalities (Roessler (2009) Hum Genet 125,393) Diaphragmatic hernia, congenital (Kantarci (2010) Am J Med Genet A 152A,2493) Tetralogy of Fallot (Silversides (2012) PLoS Genet 8, e1002843)
DLX3	74.4	99%	89%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, hypomaturation-hypoplastic type, with taurodontism, 104510
DLX4	150.3	99%	99%	No OMIM disease ID
DSPP	161.3	98%	97%	Dentinogenesis imperfecta, Shields type II, 125490 Deafness, autosomal dominant 36, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type III, 125500 Dentin dysplasia, type II, 125420 -3
EDA	50.1	97%	89%	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500
EDAR	85	100%	99%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 [Hair morphology 1, hair thickness], 612630
EDARADD	130.1	100%	99%	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDN1	135.5	100%	100%	auriculocondylar syndrome 3,615706 Question mark ears,isolated,612798 {High density lipoprotein cholesterol level QTL 7}
EFNB1	57.9	100%	97%	?Craniofrontonasal dysplasia, 304110

EFTUD2	95.9	100%	98%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
ENAM	134.3	100%	100%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ERF	119.1	100%	99%	Craniosynostosis 4, 600775
EZH2	89.5	99%	95%	Weaver syndrome,277590
FAM83H	91.7	100%	99%	Amelogenesis imperfecta, type 3, 130900
FGD1	48.5	95%	85%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF8	52.2	80%	62%	Hypogonadotropic hypogonadism 6 with or without anosmia,612702
FGFR1	125	100%	93%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Osteoglophonic dysplasia, 166250 Trigonocephaly 1, 190440 Hartsfield syndrome, 615465
FGFR2	127	97%	97%	Crouzon syndrome, 123500 Jackson-Weiss syndrome, 123150 Beare-Stevenson cutis gyrata syndrome, 123790 Pfeiffer syndrome, 101600 Apert syndrome, 101200 Saethre-Chotzen syndrome, 101400 Craniosynostosis, nonspecific Gastric cancer, somatic, 61
FGFR3	82.8	93%	92%	Achondroplasia, 100800 Hypochondroplasia, 146000 Thanatophoric dysplasia, type I, 187600 Crouzon syndrome with acanthosis nigricans, 612247 Muenke syndrome, 602849 Bladder cancer, somatic, 109800 Colorectal cancer, somatic, 114500
FOXC1	60	100%	94%	Iridogoniodysgenesis, type 1, 601631 Rieger or Axenfeld anomalies, 602482 Axenfeld-Rieger syndrome, type 3, 602482 Iris hypoplasia and glaucoma, 601631
FOXE1	57.9	99%	95%	Bamforth-Lazarus syndrome, 241850

GLI2	116.9	100%	97%	Holoprosencephaly-9, 610829
GLI3	121.7	100%	99%	Greig cefalopolysyndactyly
GNAI3	121.8	100%	100%	Auriculocondylar syndrome 1,602483
GRHL3	115.1	100%	98%	Van der Woude syndrome 2, 606713
IFT122	87.9	95%	95%	Cranioectodermal dysplasia 1
IFT43	88	100%	94%	Cranioectodermal dysplasia 3
IKBKG	15.5	26%	23%	Incontinentia pigmenti, type II, 308300 Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency, isolated, 300584
IL11RA	92.2	100%	96%	Craniosynostosis and dental anomalies, 614188
IRF6	104.9	98%	95%	van der Woude syndrome, 119300 Popliteal pterygium syndrome 1, 119500 Orofacial cleft 6, 608864
KAT6B	153.4	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KLK4	154.5	100%	100%	Amelogenesis imperfecta, type IIA1, 204700
KMT2D	111.8	99%	98%	Kabuki syndrome 1, 147920
LRP2	115.6	100%	99%	Donnai-Barrow syndrome,222448
LRP6	112.4	99%	98%	{Coronary artery disease,autosomal dominant,2},610947
LTBP3	80.4	100%	98%	Tooth agenesis, selective, 6, 613097
MED12	67.4	94%	86%	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895
MID1	77.8	100%	92%	Opitz GBBB syndrome, type I, 300000
MMP20	106.9	100%	99%	Amelogenesis imperfecta, type IIA2, 612529
MSX1	64.9	100%	94%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500
MSX2	30.6	82%	60%	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
NIPBL	127	98%	98%	Cornelia de Lange syndrome 1, 122470

NOG	144.7	100%	100%	Symphalangism, proximal, 185800 Multiple synostosis syndrome 1, 186500 Tarsal-carpal coalition syndrome, 186570 Stapes ankylosis with broad thumb and toes, 184460 Brachydactyly, type B2, 611377
NSD1	131.6	100%	99%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 (1) Beckwith-Wiedemann syndrome, 130650
OFD1	39.7	87%	78%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OTX2	181.6	100%	100%	Microphthalmia, syndromic 5
PAX6	99.1	100%	99%	?Morning glory disc anomaly, 120430 Aniridia, 106210 Cataract with late-onset corneal dystrohpy, 106210 Coloboma of optic nerve, 120430 Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Gillespie syndrome, 206700 Keratitis, 148190
PAX7	88.7	100%	93%	Rhabdomyosarcoma 2, alveolar, 268220
PAX9	247.1	99%	99%	Tooth agenesis, selective, 3, 604625
PITX2	128	99%	91%	Axenfeld-Rieger syndrome, type 1, 180500
PLCB4	92.4	100%	99%	Auriculocondylar syndrome 2, 614669
POLR1C	115.9	90%	90%	Treacher Collins syndrome 3, 248390
POLR1D	183.6	100%	100%	Treacher Collins syndrome 2, 613717
PTCH1	92.4	99%	96%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828
PTH1R	88.8	100%	98%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PVRL1	87.9	100%	98%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060

RAD21	93.3	100%	95%	Cornelia de Lange syndrome,614701
RPS6KA3	52.3	100%	92%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RUNX2	95.4	74%	74%	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156
SATB2	118.8	100%	99%	Cleft palate and mental retardation, 119540
SH3BP2	98.3	90%	85%	Cherubism, 118400
SHH	113.1	99%	91%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SIX3	130.1	100%	100%	Holoprosencephaly-2, 157170 Schizensephaly, 269160
SMC1A	72.3	96%	91%	Cornelia de Lange syndrome 2,300590
SMC3	122.7	99%	97%	Cornelia de Lange syndrome 3,610759
SMOC2	82.5	99%	86%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SPECC1L	140.3	100%	98%	Facial clefting, oblique, 1, 600251
SUMO1	19.5	63%	36%	Orofacial cleft 10, 613705
TBX22	76.5	98%	88%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TCF12	124.5	100%	100%	Craniosynostosis 3, 615314
TCOF1	103.1	100%	99%	Treacher Collins syndrome 1, 154500
TGFBR1	137.2	95%	93%	Loeys-Dietz syndrome, type 1A, 609192 Loeys-Dietz syndrome, type 2A, 608967 {Multiple self-healing squamous epithelioma, susceptiblity to}, 132800
TGFBR2	97.6	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331
TGIF1	170.1	100%	100%	Holoprosencephaly-4, 142946

TP63	133	100%	100%	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Split-hand/foot malformation 4, 605289 Hay-Wells syndrome, 106260 ADULT syndrome, 103285 Limb-mammary syndrome, 603543 Rapp-Hodgkin syndrome, 129400
TWIST1	136.6	100%	99%	Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750
UBB	14.2	62%	42%	Cleft palate, isolated, 119540
VAX1	91.7	100%	97%	Microphthalmia, syndromic 11, 614402
WDR35	120.2	100%	99%	Cranioectodermal dysplasia 2
WDR72	119.6	100%	99%	Amelogenesis imperfecta, hypomaturation type, IIA3, 613211
WNT10A	74.3	99%	88%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
ZEB2	160.3	100%	100%	Mowat-Wilson syndrome, 235730
ZIC2	67.8	96%	91%	Holoprosencephaly-5, 609637

Gene symbols used follow HGCN guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan;43(Database issue):D1079-85.
Median Coverage describes the average number of reads seen across 50 exomes
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x
OMIM release used for OMIM disease identifiers and descriptions : November 15th, 2015
This list is accurate for all panel versions starting with DG 2.4. (where x is a random number signifying a minor analysis patch without consequences for the panel composition or coverage information)

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors