CRANIOFACIAL ANOMALIES GENE PANEL DG 2.5/2.6

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
ADAMTSL4	78.4	100%	97%	Ectopia lentis et pupillae,225200 Ectopia lentis,isolated,autosomal recessive,225100
ALX1	147.3	99%	95%	Frontonasal dysplasia 3, 613456
ALX3	92.5	73%	69%	Frontonasal dysplasia 1, 136760
ALX4	112.6	97%	85%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451 {Craniosynostosis 5, susceptibility to},615529
AMELX	47.2	100%	90%	Amelogenesis imperfecta, hypoplastic/hypomaturation type 1E, 301200
ANKRD11	85.4	96%	91%	KBG syndrome, 148050
AXIN2	111.4	100%	99%	Oligodontia-colorectal cancer syndrome, 608615 Colorectal cancer, somatic, 114500
BCOR	71	98%	95%	Microphthalmia, syndromic 2, 300166
BMP4	120.8	100%	98%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C4orf26	183.7	100%	100%	Amelogenesis imperfecta, hypomaturation type, IIA4, 614832
CDON	132.5	100%	98%	Holoprosencephaly 11, 614226
CHD7	136.1	99%	96%	CHARGE syndrome,214800 Hypogonadotropic hypogonadism 5 with or without anosmia,612370
COL11A1	85.5	92%	83%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	11	46%	13%	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	93.5	99%	96%	Achondrogenesis, type II or hypochondrogenesis,200610 Avascular necrosis of the femoral head,608805 Czech dysplasia,609162 Epiphyseal dysplasia,multiple,myopia and deafness,132450 Kniest dysplasia,156550 Legg-Calve-Perthes disease,150600 Osteoarthritis with mild chondrodysplasia,604864 Otospondylomegaloepiphyseal dysplasia, 215150 Platylospondylic skeletal dysplasia,Torrance type,151210 SED congenita,183900 SMED Strudwick type,184250 Spondyloepiphyseal dysplasia,Stanescu type,616583 Spondyloperipheral dysplasia,271700 Stickler syndrome,type 1,nonsyndromic ocular,609508 Stickler syndrome,type I,108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia
COL9A1	107.6	99%	95%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	55.5	98%	86%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
CTSK	105.1	100%	100%	Pycnodysostosis, 265800
DISP1	191.9	99%	98%	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	112.7	100%	96%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, hypomaturation-hypoplastic type, with taurodontism, 104510
DLX4	101.4	99%	99%	?Orofacial cleft 15,616788
DSPP	182.5	100%	98%	Dentinogenesis imperfecta, Shields type II, 125490 Deafness, autosomal dominant 36, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type III, 125500 Dentin dysplasia, type II, 125420
EDA	47.4	80%	73%	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500

EDAR	118.2	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	75.6	91%	91%	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDN1	138.6	100%	100%	auriculocondylar syndrome 3,615706 Question mark ears,isolated,612798 {High density lipoprotein cholesterol level QTL 7}
EFNB1	82.8	100%	98%	?Craniofrontonasal dysplasia, 304110
EFTUD2	114.2	99%	98%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
ENAM	124.6	100%	100%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ERF	93.8	100%	95%	Craniosynostosis 4, 600775
EZH2	131.8	99%	96%	Weaver syndrome,277590
FAM83H	60.9	92%	86%	Amelogenesis imperfecta, type 3, 130900
FGD1	53.8	89%	77%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF8	90.6	81%	74%	Hypogonadotropic hypogonadism 6 with or without anosmia,612702
FGFR1	135.5	100%	95%	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	133.9	96%	95%	Antley-Bixler syndrome wo genital anomalies or disordered steroidogenesis,207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome,123790 Bent bone dysplasia syndrome,614592 Craniofacial-skeletal-dermatologic dysplasia,101600 Craniosynostosis,nonspecific Crouzon syndrome,123500 Gastric cancer,somatic,613659 Jackson-Weiss syndrome,123150 LADD syndrome,149730 Pfeiffer syndrome,101600 Saethre-Chotzen syndrome,101400 Scaphocephaly and Axenfeld-Rieger anomaly Scaphocephaly,maxillary retrusion,mental retardation,609579
FGFR3	91.3	100%	98%	Achondrodysplasia,100800 Bladder cancer,somatic,109800 CATSHL syndrome,610474 Cervical cancer,somatic,603956 Colorectal cancer,somatic,114500 Crouzon syndrome with acanthosis nigricans,612247 Hypochondroplasia,146000 LADD syndrome,149730 Muenke syndrome,602849 Nevus,epidermal,somatic,162900 SADDAN,616482 Spermatocytic seminoma, somatic,273300 Thanatophoric dysplasia,type I,187600 Thanatophoric dysplasia,type II,187601
FOXC1	31.1	95%	72%	Iridogoniodysgenesis, type 1, 601631 Rieger or Axenfeld anomalies, 602482 Axenfeld-Rieger syndrome, type 3, 602482 Iris hypoplasia and glaucoma, 601631
FOXE1	27.1	71%	57%	Bamforth-Lazarus syndrome, 241850
GLI2	110.3	97%	93%	Holoprosencephaly-9, 610829
GLI3	138.5	100%	100%	Greig cefalopolysyndactyly

GNAI3	124.4	100%	98%	Auriculocondylar syndrome 1,602483
GRHL3	127.9	100%	100%	Van der Woude syndrome 2, 606713
IFT122	145.6	100%	99%	Cranioectodermal dysplasia 1
IFT43	110.9	100%	100%	Cranioectodermal dysplasia 3
IKBKG	25.6	68%	55%	Ectodermal dysplasia,hypohidrotic,with immune deficiency,300291 Ectodermal,dysplasia,anhidrotic,lymphedema and immunodeficiency,300301 Immunodeficiency 33,300636 Immunodeficiency,isolated,300584 Uncontinentia pigmenti,308300 Invasive pneumococcal disease,recurrent isolated,2,300640
IL11RA	126.7	100%	97%	Craniosynostosis and dental anomalies, 614188
IRF6	121.7	99%	93%	van der Woude syndrome, 119300 Popliteal pterygium syndrome 1, 119500 Orofacial cleft 6, 608864
KAT6B	167.3	98%	97%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KDM1A	130.9	95%	92%	Cleft palate,psychomotor retardation,distinctive facial features,616728
KLK4	180	100%	94%	Amelogenesis imperfecta, type IIA1, 204700
KMT2D	133.1	99%	99%	Kabuki syndrome 1, 147920
LRP2	176.4	100%	99%	Donnai-Barrow syndrome,222448
LRP6	164.7	100%	99%	{Coronary artery disease,autosomal dominant,2},610947
LTBP3	99.2	98%	96%	Tooth agenesis, selective, 6, 613097
MED12	64.3	95%	89%	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895
MID1	111.1	100%	99%	Opitz GBBB syndrome, type I, 300000
MMP20	101.1	99%	97%	Amelogenesis imperfecta, type IIA2, 612529
MSX1	75.7	94%	87%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500
MSX2	82.3	100%	92%	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
NIPBL	111.4	96%	93%	Cornelia de Lange syndrome 1, 122470

NOG	145.3	100%	99%	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	142.2	100%	100%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 (1) Beckwith-Wiedemann syndrome, 130650
OFD1	29.6	74%	53%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OTX2	116.2	100%	97%	Microphthalmia, syndromic 5
PAX6	129.3	100%	100%	?Morning glory disc anomaly,120430 Aniridia,106210 Cataract with late-onset corneal dystrophy,106210 Coloboma of optic nerve,120430 Coloboma, ocular,120200 Foveal hypoplasia 1,136520 Keratitis,148190 Optic nerve hypoplasia,165550 Peters Anomaly,604229
PAX7	92.6	100%	100%	Rhabdomyosarcoma 2, alveolar, 268220
PAX9	228.6	99%	93%	Tooth agenesis, selective, 3, 604625
PITX2	128.1	96%	93%	Axenfeld-Rieger syndrome, type 1, 180500
PLCB4	125.6	100%	97%	Auriculocondylar syndrome 2, 614669
POLR1C	107.8	97%	94%	Treacher Collins syndrome 3, 248390
POLR1D	171.4	100%	100%	Treacher Collins syndrome 2, 613717
PTCH1	116.3	98%	96%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828
PTH1R	92.9	99%	97%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400

PVRL1	134.6	100%	99%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
RAD21	89.1	99%	96%	Cornelia de Lange syndrome,614701
RPS6KA3	55.2	88%	80%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RUNX2	94.4	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, 156510
SATB2	98	98%	91%	Cleft palate and mental retardation, 119540
SH3BP2	100.1	91%	90%	Cherubism, 118400
SHH	84.8	95%	91%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SIX3	122.8	97%	89%	Holoprosencephaly-2, 157170 Schizensephaly, 269160
SMC1A	67.1	99%	95%	Cornelia de Lange syndrome 2,300590
SMC3	77.3	94%	77%	Cornelia de Lange syndrome 3,610759
SMOC2	98.8	97%	92%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SPECC1L	141.1	100%	100%	Facial clefting, oblique, 1, 600251
SUMO1	22.5	66%	45%	Orofacial cleft 10, 613705
TBX22	66.2	96%	90%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TCF12	147.6	100%	99%	Craniosynostosis 3, 615314
TCOF1	75.9	98%	93%	Treacher Collins syndrome 1, 154500
TGFBR1	196.2	93%	93%	Loeys-Dietz syndrome, type 1A, 609192 Loeys-Dietz syndrome, type 2A, 608967 {Multiple self-healing squamous epithelioma, susceptiblity to}, 132800
TGFBR2	184.5	100%	100%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331
TGIF1	126.4	100%	97%	Holoprosencephaly-4, 142946

TP63	186.5	100%	100%	ADULT syndrome,103285 Ectrodactyly,ectodermal dysplasia,cleft lip/palate syndrome 3,604292 Hay-Wells syndrome,106260 Limb-mammary syndrome,603543 Orofacial cleft 8,129400 Rapp-Hodgkin syndrome,129400 Split-hand/foot malformation 4,605289
TWIST1	104.6	97%	87%	Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750
UBB	54.6	100%	100%	Cleft palate, isolated, 119540
VAX1	44.7	87%	70%	Microphthalmia, syndromic 11, 614402
WDR35	143.9	97%	94%	Cranioectodermal dysplasia 2
WDR72	145.6	99%	98%	Amelogenesis imperfecta, hypomaturation type, IIA3, 613211
WNT10A	90.5	100%	96%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
ZEB2	149.6	100%	98%	Mowat-Wilson syndrome, 235730
ZIC2	82.9	88%	76%	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
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

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