CRANIOFACIAL ANOMALIES GENE PANEL DG 2.3.x

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM ID*
ADAMTSL4	98,5	99%	96%	Ectopia lentis et pupillae,225200 Ectopia lentis,isolated,autosomal recessive,225100
ALX1	157,5	100%	100%	Frontonasal dysplasia 3, 613456
ALX3	74,4	85%	79%	Frontonasal dysplasia 1, 136760
ALX4	70,4	100%	97%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451 {Craniosynostosis 5, susceptibility to},615529
AMELX	118,5	100%	100%	Amelogenesis imperfecta, hypoplastic/hypomaturation type 1E, 301200
ANKRD11	110,4	92%	88%	KBG syndrome, 148050
AXIN2	98,4	98%	95%	Oligodontia-colorectal cancer syndrome, 608615 Colorectal cancer, somatic, 114500
BCOR	127,1	99%	99%	Microphthalmia, syndromic 2, 300166
BMP4	122,5	100%	100%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C4orf26	161,4	100%	100%	Amelogenesis imperfecta, hypomaturation type, IIA4, 614832
CDON	125,7	100%	97%	Holoprosencephaly 11, 614226
CHD7	127,7	100%	99%	CHARGE syndrome,214800 Hypogonadotropic hypogonadism 5 with or without anosmia,612370
COL11A1	102,1	98%	98%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	14,6	56%	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	91,7	100%	97%	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 Spondyloperipheral dysplasia, 271700 SED, Namaqualand type Osteoarthritis with mild chondrodysplasia, 604864 Vitreoretinopathy with phalangeal epiphyseal dysplasia Platyspondylic skeletal dysplasia, Torrance type, 151210 Otospondylomegaepiphyseal dysplasia, 215150 Avascular necrosis of the femoral head, 608805 Legg-Calve-Perthes disease, 150600 Stickler sydrome, type I, nonsyndromic ocular, 609508 Czech dysplasia, 609162
COL9A1	108,9	100%	95%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	70,6	96%	90%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
CTSK	137,4	100%	100%	Pycnodysostosis, 265800
DISP1	177,8	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	57,4	98%	90%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, hypomaturation-hypoplastic type, with taurodontism, 104510
DSPP	154,6	99%	96%	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	83,4	99%	98%	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500

EDAR	86,9	100%	98%	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	127,1	98%	95%	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDN1	136,9	100%	100%	auriculocondylar syndrome 3,615706 Question mark ears,isolated,612798
EFNB1	103,6	100%	100%	?Craniofrontonasal dysplasia, 304110
EFTUD2	95,6	99%	98%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
ENAM	137,8	100%	100%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ERF	100,1	100%	99%	Craniosynostosis 4, 600775
EZH2	90,5	99%	95%	Weaver syndrome,277590
FAM83H	74,2	99%	94%	Amelogenesis imperfecta, type 3, 130900
FGD1	99,7	100%	95%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGFR1	123,1	100%	99%	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	123,6	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 Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592

FGFR3	65,9	94%	88%	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 Cervical cancer, somatic, 603956 LADD syndrome, 149730 CATSHL syndrome, 610474 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Spermatocytic seminoma, somatic, 273300
FOXC1	46,2	94%	79%	Iridogoniodysgenesis, type 1, 601631 Rieger or Axenfeld anomalies, 602482 Axenfeld-Rieger syndrome, type 3, 602482 Iris hypoplasia and glaucoma, 601631
FOXE1	39,3	100%	78%	Bamforth-Lazarus syndrome, 241850
GLI2	109	96%	93%	Holoprosencephaly-9, 610829
GLI3	116,7	100%	99%	Greig cefalopolysyndactyly
GNAI3	112,5	100%	100%	Auriculocondylar syndrome 1,602483
GRHL3	109,7	100%	98%	Van der Woude syndrome 2, 606713
IFT122	87	96%	95%	Cranioectodermal dysplasia 1
IFT43	94,2	100%	100%	Cranioectodermal dysplasia 3
IKBKG	27,4	26%	26%	Incontinentia pigmenti, type II, 308300 Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency, isolated, 300584 {Atypical mycobacteriosis, familial}, 300636 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IL11RA	103,2	100%	94%	Craniosynostosis and dental anomalies, 614188
IRF6	98,6	98%	92%	van der Woude syndrome, 119300 Popliteal pterygium syndrome 1, 119500 Orofacial cleft 6, 608864

KAT6B	151,9	100%	99%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KLK4	152	100%	100%	Amelogenesis imperfecta, type IIA1, 204700
KMT2D	110,1	99%	98%	Kabuki syndrome 1, 147920
LTBP3	72,7	99%	95%	Tooth agenesis, selective, 6, 613097
MED12	134,2	98%	95%	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895
MID1	156,3	100%	99%	Opitz GBBB syndrome, type I, 300000
MMP20	101,1	100%	99%	Amelogenesis imperfecta, type IIA2, 612529
MSX1	59,5	93%	80%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500
MSX2	30,9	83%	58%	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
NIPBL	129,3	99%	98%	Cornelia de Lange syndrome 1, 122470
NOG	127,3	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	131,5	100%	100%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 (1) Beckwith-Wiedemann syndrome, 130650
OFD1	72,2	93%	90%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OTX2	173,8	100%	100%	Microphthalmia, syndromic 5

PAX6	97,9	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 Optic nerve hypoplasia, 165550 Peters anomaly, 604229
PAX7	79,8	99%	95%	Rhabdomyosarcoma 2, alveolar, 268220
PAX9	227,4	99%	99%	Tooth agenesis, selective, 3, 604625
PITX2	123,7	97%	92%	Axenfeld-Rieger syndrome, type 1, 180500
PLCB4	95,8	100%	99%	Auriculocondylar syndrome 2, 614669
POLR1C	121,8	90%	89%	Treacher Collins syndrome 3, 248390
POLR1D	171,9	100%	100%	Treacher Collins syndrome 2, 613717
PTCH1	85,6	97%	94%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828
PTH1R	78,8	95%	93%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PVRL1	82,7	100%	97%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
RAD21	98,5	100%	94%	Cornelia de Lange syndrome,614701
RPS6KA3	105,6	100%	99%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RUNX2	94,6	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	117,3	99%	97%	Cleft palate and mental retardation, 119540
SH3BP2	85,9	88%	86%	Cherubism, 118400

SHH	103,2	99%	90%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SIX3	109,4	100%	100%	Holoprosencephaly-2, 157170 Schizensephaly, 269160
SMC1A	150,2	99%	98%	Cornelia de Lange syndrome 2,300590
SMC3	109,3	99%	97%	Cornelia de Lange syndrome 3,610759
SMOC2	82,3	98%	87%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SPECC1L	130,1	100%	98%	Facial clefting, oblique, 1, 600251
SUMO1	20,9	58%	34%	Orofacial cleft 10, 613705
TBX22	163	97%	95%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TCF12	116,9	100%	100%	Craniosynostosis 3, 615314
TCOF1	94,5	99%	96%	Treacher Collins syndrome 1, 154500
TGFBR1	124,9	93%	93%	Loeys-Dietz syndrome, type 1A, 609192 Loeys-Dietz syndrome, type 2A, 608967 {Multiple self-healing squamous epithelioma, susceptiblity to}, 132800
TGFBR2	92,6	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331
TGIF1	168,7	100%	100%	Holoprosencephaly-4, 142946
TP63	140,2	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 Orofacial cleft 8, 129400
TWIST1	108,8	100%	85%	Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750
UBB	17,1	80%	39%	Cleft palate, isolated, 119540
VAX1	80,2	96%	90%	Microphthalmia, syndromic 11, 614402
WDR35	118,1	100%	99%	Cranioectodermal dysplasia 2

WDR72	123,8	100%	99%	Amelogenesis imperfecta, hypomaturation type, IIA3, 613211
WNT10A	70,4	96%	86%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
ZEB2	158,3	100%	100%	Mowat-Wilson syndrome, 235730
ZIC2	63,7	91%	74%	Holoprosencephaly-5, 609637

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated February 2014
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 : June 30th, 2015
This list is accurate for all panel versions starting with DG 2.3. (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