CRANIOFACIAL ANOMALIES GENE PANEL DGD141114

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ALX1	183.2	100%	100%	Frontonasal dysplasia 3, 613456
ALX3	90.8	91%	83%	Frontonasal dysplasia 1, 136760
ALX4	95.5	100%	99%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMELX	76.1	100%	100%	Amelogenesis imperfecta, hypoplastic/hypomaturation type 1E, 301200
ANKRD11	136.7	91%	89%	KBG syndrome, 148050
AXIN2	114.8	98%	95%	Oligodontia-colorectal cancer syndrome, 608615 Colorectal cancer, somatic, 114500
BCOR	74.1	99%	98%	Microphthalmia, syndromic 2, 300166
BMP4	130.8	100%	100%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C4orf26	205.3	100%	100%	Amelogenesis imperfecta, hypomaturation type, IIA4, 614832
CDON	123.4	100%	99%	Holoprosencephaly 11, 614226
COL11A1	111.7	98%	98%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	15.0	60%	25%	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	98.3	99%	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	119.6	100%	97%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	84.5	97%	93%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
CTSK	144.5	100%	100%	Pycnodysostosis, 265800
DISP1	187.6	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	93.0	98%	95%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, hypomaturation-hypoplastic type, with taurodontism, 104510
DSPP	190.1	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	49.7	100%	94%	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500

EDAR	88.0	100%	100%	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	159.3	100%	100%	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EFNB1	60.1	100%	96%	?Craniofrontonasal dysplasia, 304110
EFTUD2	100.9	100%	99%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
ENAM	139.7	100%	100%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ERF	137.2	100%	99%	Craniosynostosis 4, 600775
FAM83H	103.5	100%	98%	Amelogenesis imperfecta, type 3, 130900
FGD1	56.6	100%	92%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGFR1	135.1	100%	97%	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	134.2	100%	100%	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, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Scaphocephaly and Axenfeld-Rieger anomaly LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592

FGFR3	84.7	97%	90%	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	71.8	100%	98%	Iridogoniodysgenesis, type 1, 601631 Rieger or Axenfeld anomalies, 602482 Axenfeld-Rieger syndrome, type 3, 602482 Iris hypoplasia and glaucoma, 601631
FOXE1	57.9	100%	97%	Bamforth-Lazarus syndrome, 241850
GLI2	119.5	100%	96%	Holoprosencephaly-9, 610829
GRHL3	118.8	99%	97%	Van der Woude syndrome 2, 606713
IKBKG	10.2	16%	16%	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	107.5	100%	99%	Craniosynostosis and dental anomalies, 614188
IRF6	103.1	100%	96%	van der Woude syndrome, 119300 Popliteal pterygium syndrome 1, 119500 Orofacial cleft 6, 608864
KAT6B	164.6	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KLK4	161.5	100%	100%	Amelogenesis imperfecta, type IIA1, 204700
KMT2D	124.1	99%	98%	Kabuki syndrome 1, 147920
LTBP3	88.0	100%	99%	Tooth agenesis, selective, 6, 613097

MAFB	124.4	100%	100%	Multicentric carpotarsal osteolysis syndrome, 166300
MED12	74.5	96%	90%	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895
MID1	86.8	100%	95%	Opitz GBBB syndrome, type I, 300000
MMP20	108.6	100%	97%	Amelogenesis imperfecta, type IIA2, 612529
MSX1	74.3	95%	85%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500
MSX2	36.7	84%	77%	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
NIPBL	137.9	99%	98%	Cornelia de Lange syndrome 1, 122470
NOG	143.7	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	139.9	100%	99%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 (1) Beckwith-Wiedemann syndrome, 130650
OFD1	39.2	88%	79%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
PAX6	97.3	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	98.8	100%	97%	Rhabdomyosarcoma 2, alveolar, 268220

PAX9	237.7	99%	99%	Tooth agenesis, selective, 3, 604625
PITX2	174.5	100%	100%	Axenfeld-Rieger syndrome, type 1, 180500
PLCB4	109.8	100%	99%	Auriculocondylar syndrome 2, 614669
POLR1C	142.9	100%	99%	Treacher Collins syndrome 3, 248390
POLR1D	269.4	100%	100%	Treacher Collins syndrome 2, 613717
PTCH1	94.3	100%	98%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828
PTH1R	89.0	100%	97%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
PVRL1	90.3	100%	98%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
RPS6KA3	57.5	99%	92%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RUNX2	134.6	100%	100%	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	136.0	100%	99%	Cleft palate and mental retardation, 119540
SH3BP2	113.8	100%	96%	Cherubism, 118400
SHH	128.4	99%	96%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SIX3	151.5	100%	100%	Holoprosencephaly-2, 157170 Schizensephaly, 269160
SMOC2	89.3	100%	96%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SPECC1L	134.1	100%	99%	Facial clefting, oblique, 1, 600251
SUMO1	28.0	77%	63%	Orofacial cleft 10, 613705
TBX22	82.7	98%	96%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TCF12	134.9	100%	100%	Craniosynostosis 3, 615314
TCOF1	115.3	100%	99%	Treacher Collins syndrome 1, 154500

TGFBR1	136.2	95%	93%	Loeys-Dietz syndrome, type 1A, 609192 Loeys-Dietz syndrome, type 2A, 608967 {Multiple self-healing squamous epithelioma, susceptiblity to}, 132800
TGFBR2	103.0	100%	99%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331
TGIF1	258.0	100%	100%	Holoprosencephaly-4, 142946
TP63	142.3	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	141.9	100%	99%	Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750
UBB	22.8	57%	51%	Cleft palate, isolated, 119540 (2)
VAX1	86.0	100%	99%	Microphthalmia, syndromic 11, 614402
WDR72	135.2	100%	100%	Amelogenesis imperfecta, hypomaturation type, IIA3, 613211
WNT10A	73.5	97%	94%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
ZEB2	178.7	100%	100%	Mowat-Wilson syndrome, 235730
ZIC2	85.3	96%	89%	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 : 31 october 2014
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors