CRANIOFACIAL ANOMALIES GENE PANEL DGD20062014

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM ID
ABCA4	85,3	99%	96%	Stargardt disease 1, 248200 Retinitis pigmentosa 19, 601718 Cone-rod dystrophy 3, 604116 Macular degeneration, age-related, 2, 153800 Fundus flavimaculatus, 248200 Retinal dystrophy, early-onset severe, 248200
ALX1	161,5	100%	100%	Frontonasal dysplasia 3, 613456
ALX3	78,6	90%	77%	Frontonasal dysplasia 1, 136760
ALX4	69,8	100%	100%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMELX	54,4	100%	100%	Amelogenesis imperfecta, hypoplastic/hypomaturation type 1E, 301200
ANKRD11	107,6	90%	86%	KBG syndrome, 148050
AXIN2	88,4	98%	90%	Oligodontia-colorectal cancer syndrome, 608615 Colorectal cancer, somatic, 114500
BCOR	60,3	99%	95%	Microphthalmia, syndromic 2, 300166
BMP4	110,8	100%	100%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625 -3
C4orf26	141,9	100%	100%	Amelogenesis imperfecta, hypomaturation type, IIA4, 614832
CDON	109,7	100%	98%	Holoprosencephaly 11, 614226

COL11A1	94,5	98%	98%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis, 228520
COL11A2	13,8	53%	18%	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	80,9	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 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	101,3	100%	97%	Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	71,3	97%	92%	Epiphyseal dysplasia, multiple, 2, 600204 {Intervertebral disc disease, susceptibility to}, 603932 Stickler syndrome, type V, 614284
CTSK	116,2	100%	99%	Pycnodysostosis, 265800

DLX3	77,5	99%	86%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, hypomaturation-hypoplastic type, with taurodontism, 104510
DSPP	139,9	98%	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	45,1	97%	75%	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500
EFNB1	51,4	99%	88%	?Craniofrontonasal dysplasia, 304110
EFTUD2	82,3	99%	97%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
ENAM	117,3	100%	100%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ERF	107,1	100%	100%	Craniosynostosis 4, 600775
FAM83H	73,4	100%	95%	Amelogenesis imperfecta, type 3, 130900
FGFR1	111,9	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	116,4	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	72,9	95%	87%	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%	81%	Iridogoniodysgenesis, type 1, 601631 Rieger or Axenfeld anomalies, 602482 Axenfeld-Rieger syndrome, type 3, 602482 Iris hypoplasia and glaucoma, 601631
FOXE1	47,1	96%	91%	Bamforth-Lazarus syndrome, 241850
GLI2	99,2	99%	95%	Holoprosencephaly-9, 610829

IKBKG	6,4	14%	11%	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	86,6	100%	93%	Craniosynostosis and dental anomalies, 614188
IRF6	90,9	97%	93%	van der Woude syndrome, 119300 Popliteal pterygium syndrome 1, 119500 Orofacial cleft 6, 608864
KAT6B	137	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KLK4	136,7	100%	100%	Amelogenesis imperfecta, type IIA1, 204700
LTBP3	73,1	99%	93%	Tooth agenesis, selective, 6, 613097
MAFB	91,3	100%	100%	Multicentric carpotarsal osteolysis syndrome, 166300
MED12	60,1	94%	86%	Opitz-Kaveggia syndrome, 305450 Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895
MMP20	102,5	100%	99%	Amelogenesis imperfecta, type IIA2, 612529
MSX1	62,7	99%	93%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500
MSX2	37,7	83%	76%	Craniosynostosis, type 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
NIPBL	115,1	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	117,7	100%	99%	Sotos syndrome 1, 117550 Leukemia, acute myeloid, 601626 (1) Beckwith-Wiedemann syndrome, 130650
PAX7	83,4	99%	92%	Rhabdomyosarcoma 2, alveolar, 268220
PAX9	196,5	100%	99%	Tooth agenesis, selective, 3, 604625
PITX2	134,1	100%	100%	Axenfeld-Rieger syndrome, type 1, 180500 Iridogoniodysgenesis, type 2, 137600 Ring dermoid of cornea, 180550 Peters anomaly, 604229
PLCB4	92,4	100%	99%	Auriculocondylar syndrome 2, 614669
POLR1C	123,4	100%	97%	Treacher Collins syndrome 3, 248390
POLR1D	220,2	100%	100%	Treacher Collins syndrome 2, 613717
PTCH1	80,7	99%	94%	Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462 Holoprosencephaly-7, 610828
PVRL1	71,2	100%	98%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
RPS6KA3	48,5	99%	95%	Coffin-Lowry syndrome, 303600 Mental retardation, X-linked 19, 300844
RUNX2	115,1	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	109	100%	97%	Cleft palate and mental retardation, 119540

SHH	97,5	98%	90%	Holoprosencephaly-3, 142945 Single median maxillary central incisor, 147250 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160
SIX3	114,9	100%	98%	Holoprosencephaly-2, 157170 Schizensephaly, 269160
SMOC2	77,6	97%	83%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SPECC1L	122	100%	99%	Facial clefting, oblique, 1, 600251
SUMO1	29,5	77%	70%	Orofacial cleft 10, 613705
TBX22	75,8	100%	93%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TCF12	109,1	100%	100%	Craniosynostosis 3, 615314
TCOF1	93,1	100%	97%	Treacher Collins syndrome 1, 154500
TGFBR1	126,5	93%	93%	Loeys-Dietz syndrome, type 1A, 609192 Loeys-Dietz syndrome, type 2A, 608967 {Multiple self-healing squamous epithelioma, susceptiblity to}, 132800
TGFBR2	86,1	100%	97%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome, type 1B, 610168 Loeys-Dietz syndrome, type 2B, 610380
TP63	118,3	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	120,3	100%	99%	Saethre-Chotzen syndrome, 101400 Saethre-Chotzen syndrome with eyelid anomalies, 101400 Craniosynostosis, type 1, 123100 Robinow-Sorauf syndrome, 180750
UBB	23,8	79%	58%	Cleft palate, isolated, 119540 (2)
VAX1	72	100%	99%	Microphthalmia, syndromic 11, 614402
WDR72	118,4	100%	99%	Amelogenesis imperfecta, hypomaturation type, IIA3, 613211
WNT10A	70,1	96%	89%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
ZEB2	155,2	100%	100%	Mowat-Wilson syndrome, 235730
ZIC2	60	94%	85%	Holoprosencephaly-5, 609637

Gene symbols used follow HGCN guidelines Genomics 79(4):464-470 (2002) updated October 2013
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 : 15 october 2013
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors