CRANIOFACIAL ANOMALIES GENE PANEL DG 2.16 (171 genes)

Releasedate: 07-06-2019

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACP4	91,2	96.8%	88.5%	Amelogenesis imperfecta, type IJ, 617297
ADAMTSL4	122,7	100.0%	99.6%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ALX1	134,2	99.9%	98.5%	?Frontonasal dysplasia 3, 613456
ALX3	134,6	91.1%	79.0%	Frontonasal dysplasia 1, 136760
ALX4	157,1	100.0%	100.0%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMBN	175,1	99.3%	97.1%	Amelogenesis imperfecta, type IF, 616270
AMELX	85,7	98.9%	93.5%	Amelogenesis imperfecta, type 1E, 301200
AMER1	98,2	99.8%	98.9%	Osteopathia striata with cranial sclerosis, 300373
AMTN	119,3	100.0%	99.1%	?Amelogenesis imperfecta, type IIIB, 617607
ANKRD11	119,6	99.2%	97.1%	KBG syndrome, 148050
ARHGAP29	138,7	99.7%	98.8%	No OMIM phenotype Cleft lip with or without cleft palate (Leslie (2015) Am J Hum Genet 96,397)
AXIN2	124,2	100.0%	99.9%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
BCOR	102,7	98.8%	95.3%	Microphthalmia, syndromic 2, 300166
BMP2	163,4	100.0%	100.0%	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 {HFE hemochromatosis, modifier of}, 235200
BMP4	173,4	100.0%	100.0%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C4orf26	NC	NC	NC	Amelogenesis imperfecta, type IIA4, 614832
CDC45	138,9	99.6%	98.1%	Meier-Gorlin syndrome 7, 617063
CDON	107	100.0%	99.0%	Holoprosencephaly 11, 614226
CDSN	131	100.0%	100.0%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CHD7	137	99.9%	99.4%	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370

COL11A1	96,6	97.9%	94.0%	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780 Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	111,6	100.0%	99.4%	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150
COL2A1	112,2	100.0%	99.7%	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler sydrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL9A1	132,3	100.0%	99.7%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	95,2	99.9%	98.8%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	107,8	99.6%	96.8%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
CTSK	86,3	100.0%	99.8%	Pycnodysostosis, 265800
DHODH	98,8	100.0%	100.0%	Miller syndrome, 263750
DISP1	164,3	99.9%	99.7%	No OMIM phenotype 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	146,7	100.0%	99.0%	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320

DLX4	228,4	100.0%	100.0%	?Orofacial cleft 15, 616788
DSPP	79	98.4%	93.8%	Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentin dysplasia, type II, 125420 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
EDA	102	95.6%	85.7%	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500
EDAR	126,6	100.0%	100.0%	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	89,8	99.7%	98.3%	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDN1	156,5	100.0%	99.9%	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}, 0
EDNRA	150,7	100.0%	99.8%	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EFNA4	152,5	100.0%	100.0%	No OMIM phenotype Craniosynostosis 1 (Merrill et al. (2006) Hum Molec Genet 15)
EFNB1	116,7	100.0%	99.9%	Craniofrontonasal dysplasia, 304110
EFTUD2	103,2	100.0%	99.2%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF4A3	87,5	100.0%	99.4%	Robin sequence with cleft mandible and limb anomalies, 268305
ENAM	139,5	100.0%	100.0%	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ERF	146,8	100.0%	99.2%	Chitayat syndrome, 617180 Craniosynostosis 4, 600775
EYA1	120,2	99.9%	99.8%	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
EZH2	130	99.4%	97.6%	Weaver syndrome, 277590
FAM20A	111,1	100.0%	99.4%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM83H	120,2	100.0%	99.9%	Amelogenesis imperfecta, type IIIA, 130900
FGD1	86,7	98.4%	93.0%	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF10	120,5	100.0%	99.6%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF3	139,5	100.0%	100.0%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706

FGF8	130	97.9%	86.8%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	153,5	100.0%	100.0%	Multiple synostoses syndrome 3, 612961
FGFR1	122,6	100.0%	99.6%	Encephalocraniocutaneous lipomatosis, 613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440
FGFR2	113,1	97.7%	96.8%	Antley-Bixler syndrome without 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, 0 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, 0 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FGFR3	138,5	100.0%	99.6%	Achondroplasia, 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	80,4	99.7%	97.5%	Anterior segment dysgenesis 3, multiple subtypes, 601631

				Axenfeld-Rieger syndrome, type 3, 602482
FOXE1	87,5	100.0%	99.7%	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
GDF3	127,9	100.0%	100.0%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF6	156,3	100.0%	100.0%	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Multiple synostoses syndrome 4, 617898
GJA1	156,2	100.0%	100.0%	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJB6	140,9	100.0%	100.0%	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GLI2	158,2	100.0%	100.0%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	139,5	100.0%	99.3%	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510 Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700 {Hypothalamic hamartomas, somatic}, 241800
GNAI3	88,3	98.9%	94.5%	Auriculocondylar syndrome 1, 602483
GPR68	165,4	99.9%	99.0%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GRHL3	133,2	100.0%	99.8%	Van der Woude syndrome 2, 606713
GSC	133,8	100.0%	98.9%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
HOXA2	83	100.0%	99.9%	?Microtia, hearing impairment, and cleft palate (AR), 612290 Microtia with or without hearing impairment (AD), 612290
HUWE1	79,3	99.1%	94.3%	Mental retardation, X-linked syndromic, Turner type, 300706
HYAL2	175,3	100.0%	99.8%	No OMIM phenotype

				Orofacial clefting (Muggenthaler (2017) PLoS Genet 13,e1006470) ?Hypertelorism and high myopia (Shaheen (2016) Genet Med 18,686)
IFT122	120,5	99.9%	99.0%	Cranioectodermal dysplasia 1, 218330
IFT43	112,4	100.0%	100.0%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT88	94,7	99.6%	97.4%	No OMIM phenotype ?Cleft lip and palate (Tian (2017) Hum Mol Genet 26,860)
IKBKG	60,1	88.1%	78.8%	Ectodermal dysplasia and immunodeficiency 1, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IL11RA	131,5	100.0%	99.6%	Craniosynostosis and dental anomalies, 614188
INTU	115,4	99.8%	98.7%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
IRF6	90,3	99.4%	95.0%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300 {Orofacial cleft 6}, 608864
ITGB6	127,5	96.7%	95.0%	Amelogenesis imperfecta, type IH, 616221
KAT6B	155,7	99.9%	99.1%	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KDF1	110,3	100.0%	99.9%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	130,9	100.0%	98.8%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM6A	97,7	95.3%	87.8%	Kabuki syndrome 2, 300867
KLK4	164,3	100.0%	100.0%	Amelogenesis imperfecta, type IIA1, 204700
KMT2D	136,2	100.0%	99.7%	Kabuki syndrome 1, 147920
KREMEN1	143,4	99.7%	97.3%	Ectodermal dysplasia 13, hair/tooth type, 617392
LAMB3	116,9	100.0%	99.4%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LRP2	139,2	100.0%	99.9%	Donnai-Barrow syndrome, 222448
LRP6	136,8	99.9%	99.2%	Tooth agenesis, selective, 7, 616724 {Coronary artery disease, autosomal dominant, 2}, 610947
LTBP3	147,5	100.0%	99.6%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
MASP1	131,1	100.0%	99.3%	3MC syndrome 1, 257920

MED12	85,1	99.5%	95.5%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MEGF8	144	100.0%	99.5%	Carpenter syndrome 2, 614976
MEIS2	123,6	100.0%	99.8%	Cleft palate, cardiac defects, and mental retardation, 600987
MEOX1	105	99.9%	97.4%	Klippel-Feil syndrome 2, 214300
MID1	124,1	99.8%	97.4%	Opitz GBBB syndrome, type I, 300000
MITF	141,1	100.0%	99.8%	COMMAD syndrome, 617306 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MMP20	90,8	99.8%	97.6%	Amelogenesis imperfecta, type IIA2, 612529
MSX1	143,3	99.9%	98.6%	Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	101,1	100.0%	100.0%	Craniosynostosis 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
NAA10	105	100.0%	98.8%	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NECTIN1	134	100.0%	99.9%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NFKBIA	134,6	95.3%	89.4%	Ectodermal dysplasia and immunodeficiency 2, 612132
NIPBL	124,9	98.8%	96.9%	Cornelia de Lange syndrome 1, 122470
NOG	233,6	100.0%	100.0%	Brachydactyly, type B2, 611377 Multiple synostoses syndrome 1, 186500 Stapes ankylosis with broad thumbs and toes, 184460 Symphalangism, proximal, 1A, 185800 Tarsal-carpal coalition syndrome, 186570
NSD1	147	100.0%	99.8%	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
OFD1	51,9	85.8%	70.8%	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OTX2	127,4	100.0%	99.3%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986

				Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
PAX3	106,9	100.0%	99.7%	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX6	116,5	100.0%	99.8%	?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 ?Morning glory disc anomaly, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550
PAX7	131,1	100.0%	100.0%	Rhabdomyosarcoma 2, alveolar, 268220
PAX9	236,1	99.8%	99.6%	Tooth agenesis, selective, 3, 604625
PGM1	128,8	100.0%	99.8%	Congenital disorder of glycosylation, type It, 614921
PITX2	164,8	100.0%	99.5%	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550
PLCB4	102,5	99.8%	98.0%	Auriculocondylar syndrome 2, 614669
POLR1C	98,3	98.9%	94.9%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	183,1	91.6%	91.6%	Treacher Collins syndrome 2, 613717
PORCN	111,2	99.9%	98.8%	Focal dermal hypoplasia, 305600
PTCH1	110,2	99.9%	98.4%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTH1R	106,6	100.0%	99.1%	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
RAB23	107,4	100.0%	99.2%	Carpenter syndrome, 201000
RAD21	83	97.8%	93.4%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RBM10	111,6	99.8%	97.6%	TARP syndrome, 311900
RECQL4	159,9	100.0%	99.8%	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280

				Rothmund-Thomson syndrome, 268400
RIPK4	167,5	100.0%	100.0%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RUNX2	102,8	73.4%	72.2%	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510
SALL1	113,3	99.9%	98.9%	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480
SALL4	135	99.9%	98.1%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SATB2	107,4	99.8%	97.7%	Glass syndrome, 612313
SEMA3E	130,9	100.0%	99.6%	?CHARGE syndrome, 214800
SF3B4	75,5	99.9%	98.3%	Acrofacial dysostosis 1, Nager type, 154400
SH3BP2	139,3	91.9%	91.4%	Cherubism, 118400
SHH	147,1	100.0%	100.0%	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250
SIX1	131,1	99.9%	98.7%	Branchiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX3	206	100.0%	99.9%	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SKI	132,9	100.0%	99.3%	Shprintzen-Goldberg syndrome, 182212
SLC24A4	103,5	100.0%	99.8%	Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750 [Skin/hair/eye pigmentation 6, blue/green eyes], 210750
SLC26A2	205,1	100.0%	99.9%	Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Epiphyseal dysplasia, multiple, 4, 226900
SMAD6	180,5	98.8%	89.1%	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439
SMC1A	87,8	99.9%	97.8%	Cornelia de Lange syndrome 2, 300590
SMC3	84	96.0%	89.7%	Cornelia de Lange syndrome 3, 610759
SMO	140,4	99.9%	98.3%	Basal cell carcinoma, somatic, 605462

				Curry-Jones syndrome, somatic mosaic, 601707
SMOC2	88,7	77.0%	75.7%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNAI2	102,7	99.9%	99.1%	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SOX10	88,2	100.0%	99.1%	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX6	91,8	99.9%	98.5%	No OMIM phenotype Developmental delay and spinal syrinx (Scott (2014) J Child Neurol 29, NP164) Dystonia, dopa-responsive (Ebrahimi-Fakhari (2015) Pediatr Neurol 52,115) ?Craniosynostosis (Tagariello (2006) J Med Genet 43,534)
SOX9	159,9	100.0%	100.0%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SPECC1L	127,5	100.0%	99.8%	?Facial clefting, oblique, 1, 600251 Hypertelorism, Teebi type, 145420 Opitz GBBB syndrome, type II, 145410
SUMO1	20	62.3%	46.0%	?Orofacial cleft 10, 613705
TBX1	101,2	93.0%	86.9%	Conotruncal anomaly face syndrome, 217095 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Velocardiofacial syndrome, 192430
TBX22	105	99.4%	94.8%	?Abruzzo-Erickson syndrome, 302905 Cleft palate with ankyloglossia, 303400
TCF12	137,7	99.9%	99.9%	Craniosynostosis 3, 615314
TCOF1	111,6	99.9%	99.1%	Treacher Collins syndrome 1, 154500
TFAP2A	112,7	99.8%	98.0%	Branchiooculofacial syndrome, 113620
TGFBR1	156,4	95.4%	93.8%	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	156,8	100.0%	100.0%	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGIF1	141,2	100.0%	100.0%	Holoprosencephaly 4, 142946
TP63	162,8	100.0%	100.0%	ADULT syndrome, 103285 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Limb-mammary syndrome, 603543 Orofacial cleft 8, 618149

				Rapp-Hodgkin syndrome, 129400 Split-hand/foot malformation 4, 605289
TRAF6	75	96.3%	85.8%	No OMIM phenotype Ectodermal dysplasia, hypohidrotic (Wisniewski (2012) Br J Dermatol 166,1353)
TSHZ1	147,6	98.9%	98.7%	Aural atresia, congenital, 607842
TSPEAR	139,3	100.0%	99.8%	?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TWIST1	160,2	100.0%	99.6%	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 Sweeney-Cox syndrome, 617746
UBB	37,6	100.0%	93.3%	Cleft palate, isolated, 119540
VAX1	95,1	99.5%	95.7%	?Microphthalmia, syndromic 11, 614402
WDR19	126,8	100.0%	99.2%	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR35	141,8	99.7%	98.4%	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR72	123,8	96.8%	96.1%	Amelogenesis imperfecta, type IIA3, 613211
WNT10A	141,8	100.0%	99.9%	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
WNT10B	157	100.0%	100.0%	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
ZEB2	140,1	99.7%	98.4%	Mowat-Wilson syndrome, 235730
ZIC1	279,6	100.0%	100.0%	Craniosynostosis 6, 616602
ZIC2	165,6	97.5%	95.4%	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 : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors