CRANIOFACIAL ANOMALIES GENE PANEL DG 3.00 (186 genes)

Releasedate: 02-12-2020

Gene	Agilent V5 covered > 10x	Agilent V5 covered > 20x	TWIST covered > 10x	TWIST covered 20x	Associated Phenotype description and OMIM disease ID
ACP4	97,2	88,8	100	100	Amelogenesis imperfecta, type IJ, 617297
ADAMTSL4	100	99,2	100	100	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ALX1	99,7	97,1	100	100	Frontonasal dysplasia 3, 613456
ALX3	77,9	73,3	100	100	Frontonasal dysplasia 1, 136760
ALX4	100	99,3	100	100	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMBN	99,8	98,5	100	100	Amelogenesis imperfecta, type IF, 616270
AMELX	99,9	96,8	100	100	Amelogenesis imperfecta, type 1E, 301200
AMER1	99,9	98,5	100	100	Osteopathia striata with cranial sclerosis, 300373
AMTN	99,6	98,6	100	100	?Amelogenesis imperfecta, type IIIB, 617607
ANKRD11	96,1	93,5	100	100	KBG syndrome, 148050
ARHGAP29	99,5	98	100	100	No OMIM disease ID
AXIN2	100	99,9	100	99,9	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
BCOR	99,6	97,4	100	99,9	Microphthalmia, syndromic 2, 300166
BMP2	100	100	100	100	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 {HFE hemochromatosis, modifier of}, 235200
BMP4	100	100	100	100	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
IMPAD1	100	100	100	100	Chondrodysplasia with joint dislocations, GPAPP type, 614078
CCBE1	99,8	98,8	100	100	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510

CDC45	99,8	98,5	100	100	Meier-Gorlin syndrome 7, 617063
CDON	100	99,6	100	100	Holoprosencephaly 11, 614226
CDSN	100	100	100	100	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CHD7	100	99,5	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
COL11A1	96,2	92,8	100	100	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis 1, 228520
COL11A2	100	99,7	100	100	Deafness, autosomal dominant 13, 601868 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 Fibrochondrogenesis 2, 614524 Deafness, autosomal recessive 53, 609706 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL2A1	100	99,7	100	100	Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 Kniest dysplasia, 156550 Stickler syndrome, type I, 108300 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 Avascular necrosis of the femoral head, 608805 ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 SED congenita, 183900 Legg-Calve-Perthes disease, 150600 SMED Strudwick type, 184250 Czech dysplasia, 609162 Stickler sydrome, type I, nonsyndromic ocular, 609508 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL9A1	100	99,2	100	100	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	99,9	99	100	100	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	98,7	95,5	99,7	98,6	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932

COLEC11	100	100	100	100	3MC syndrome 2, 265050
CTSK	100	99,9	100	100	Pycnodysostosis, 265800
CYP26B1	100	99,9	100	100	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
DHODH	100	100	100	100	Miller syndrome, 263750
DISP1	100	99,9	100	100	No OMIM disease ID
DLX3	99,9	98,4	100	100	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX4	100	100	100	100	?Orofacial cleft 15, 616788
DSPP	96,8	86,1	100	100	Dentin dysplasia, type II, 125420 Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
EDA	98,1	91,6	100	99,9	Tooth agenesis, selective, X-linked 1, 313500 Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	100	99,9	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	99,9	98,8	100	100	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941 Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
EDN1	100	100	100	100	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}, 0
EDNRA	100	100	100	100	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EFNA4	100	100	100	100	No OMIM disease ID
EFNB1	100	100	100	100	Craniofrontonasal dysplasia, 304110
EFTUD2	100	99,8	100	100	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF4A3	100	99,5	100	100	Robin sequence with cleft mandible and limb anomalies, 268305
ENAM	100	100	100	100	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ERF	99,9	98,5	100	100	Craniosynostosis 4, 600775 Chitayat syndrome, 617180

ESCO2	98,7	95,2	100	100	Roberts syndrome, 268300 SC phocomelia syndrome, 269000
EYA1	99,9	99,7	100	100	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Branchiootic syndrome 1, 602588
EZH2	100	99,5	100	100	Weaver syndrome, 277590
FAM20A	99,6	94,7	100	100	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM83H	84,9	81,5	100	100	Amelogenesis imperfecta, type IIIA, 130900
FGD1	97,3	92,8	100	100	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF10	100	99,8	100	100	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF3	99,8	95,1	100	100	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF8	98,2	88,9	100	99,6	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	100	100	100	100	Multiple synostoses syndrome 3, 612961
FGFR1	100	99,9	100	100	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Hartsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	97,7	97,1	100	100	Apert syndrome, 101200 Jackson-Weiss syndrome, 123150 Saethre-Chotzen syndrome, 101400 Gastric cancer, somatic, 613659 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Craniofacial-skeletal-dermatologic dysplasia, 101600 Pfeiffer syndrome, 101600 Crouzon syndrome, 123500 Beare-Stevenson cutis gyrata syndrome, 123790 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410

					Craniosynostosis, nonspecific, 0 Scaphocephaly and Axenfeld-Rieger anomaly, 0
FGFR3	99,8	97,7	100	99,8	Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 Thanatophoric dysplasia, type II, 187601 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000 LADD syndrome, 149730 Achondroplasia, 100800 Thanatophoric dysplasia, type I, 187600 Colorectal cancer, somatic, 114500 Spermatocytic seminoma, somatic, 273300 Cervical cancer, somatic, 603956 SADDAN, 616482
FLNA	100	99,9	100	100	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620
FLNB	99,5	98,8	100	100	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Boomerang dysplasia, 112310 Spondylocarpotarsal synostosis syndrome, 272460 Atelosteogenesis, type III, 108721
FOXC1	98	89,6	99,9	98,5	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXE1	96,9	78,5	99,9	99,1	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
GDF3	100	100	100	100	Microphthalmia, isolated 7, 613704 Microphthalmia with coloboma 6, 613703 Klippel-Feil syndrome 3, autosomal dominant, 613702

GDF6	100	99,9	100	99,4	Leber congenital amaurosis 17, 615360 Klippel-Feil syndrome 1, autosomal dominant, 118100 Multiple synostoses syndrome 4, 617898 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094
GJA1	100	100	100	100	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Atrioventricular septal defect 3, 600309 Oculodentodigital dysplasia, 164200 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100
GJB6	100	100	100	100	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GLI2	99,1	97,4	100	99,8	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	98,5	98	100	100	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GNAI3	99,3	95,2	100	100	Auriculocondylar syndrome 1, 602483
GNPTAB	100	99,9	100	100	Mucolipidosis II alpha/beta, 252500 Mucolipidosis III alpha/beta, 252600
GPR68	99,5	96,7	100	100	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GRHL3	100	100	100	100	Van der Woude syndrome 2, 606713
GSC	99,2	92,4	100	100	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
HOXA2	100	99,9	100	100	Microtia with or without hearing impairment (AD), 612290 ?Microtia, hearing impairment, and cleft palate (AR), 612290
HUWE1	99,2	95,8	100	100	Mental retardation, X-linked syndromic, Turner type, 309590
HYAL2	100	100	100	100	No OMIM disease ID
IFT122	100	99,6	100	100	Cranioectodermal dysplasia 1, 218330

IFT43	100	100	100	100	?Cranioectodermal dysplasia 3, 614099 Short-rib thoracic dysplasia 18 with polydactyly, 617866 ?Retinitis pigmentosa 81, 617871
IFT88	99,6	97,3	100	100	No OMIM disease ID
IKBKG	84,1	77,2	100	100	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291
IL11RA	100	99,9	100	100	Craniosynostosis and dental anomalies, 614188
IL6ST	96,4	90,3	100	100	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
INTU	99,7	98,1	100	100	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
IRF6	99,6	95,9	100	100	Popliteal pterygium syndrome 1, 119500 {Orofacial cleft 6}, 608864 van der Woude syndrome, 119300
ITGB6	97,2	95,8	100	100	Amelogenesis imperfecta, type IH, 616221
KAT6B	99,6	98,3	100	100	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KDF1	100	99,8	100	100	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	98,2	95,3	100	100	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM6A	96,1	88,7	100	99,9	Kabuki syndrome 2, 300867
KLK4	100	100	100	100	Amelogenesis imperfecta, type IIA1, 204700
KMT2D	100	99,4	100	100	Kabuki syndrome 1, 147920
KREMEN1	97,7	94,4	99,5	97,9	Ectodermal dysplasia 13, hair/tooth type, 617392
LAMB3	100	99,6	100	100	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LRP2	100	99,9	100	100	Donnai-Barrow syndrome, 222448
LRP6	100	99,9	100	100	{Coronary artery disease, autosomal dominant, 2}, 610947 Tooth agenesis, selective, 7, 616724
LTBP3	99,6	98,1	100	100	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
MASP1	100	99,9	100	100	3MC syndrome 1, 257920

MED12	99,8	96,7	100	100	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
MEGF8	99,9	99	100	100	Carpenter syndrome 2, 614976
MEIS2	100	100	100	100	Cleft palate, cardiac defects, and mental retardation, 600987
MEOX1	100	98,9	100	100	Klippel-Feil syndrome 2, 214300
MID1	99,8	98,7	100	100	Opitz GBBB syndrome, type I, 300000
MITF	100	99,9	100	100	COMMAD syndrome, 617306 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 Tietz albinism-deafness syndrome, 103500
MMP20	100	100	100	100	Amelogenesis imperfecta, type IIA2, 612529
MN1	100	99,3	100	100	Meningioma, 607174 CEBALID syndrome, 618774
MSX1	96,9	89,3	100	100	Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	100	99,4	100	100	Parietal foramina 1, 168500 Craniosynostosis 2, 604757 Parietal foramina with cleidocranial dysplasia, 168550
NAA10	99,7	98,5	99,9	99,9	Ogden syndrome, 300855 Microphthalmia, syndromic 1, 309800
NECTIN1	100	99,9	100	100	Orofacial cleft 7, 225060 Cleft lip/palate-ectodermal dysplasia syndrome, 225060
NFKBIA	95,2	88	100	100	Ectodermal dysplasia and immunodeficiency 2, 612132
NIPBL	98,9	97	100	100	Cornelia de Lange syndrome 1, 122470
NOG	100	100	100	100	Tarsal-carpal coalition syndrome, 186570 Symphalangism, proximal, 1A, 185800 Stapes ankylosis with broad thumbs and toes, 184460 Multiple synostoses syndrome 1, 186500 Brachydactyly, type B2, 611377
NSD1	100	99,9	100	100	Sotos syndrome 1, 117550

ODAPH	100	100	100	100	Amelogenesis imperfecta, type IIA4, 614832
OFD1	88	73,7	100	99,9	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OTX2	100	99,7	100	100	Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 Microphthalmia, syndromic 5, 610125
P4HB	94,6	94	100	100	Cole-Carpenter syndrome 1, 112240
PAX3	100	99,9	100	100	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX6	100	100	100	100	Optic nerve hypoplasia, 165550 ?Coloboma, ocular, 120200 Foveal hypoplasia 1, 136520 Aniridia, 106210 Keratitis, 148190 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma of optic nerve, 120430 ?Morning glory disc anomaly, 120430
PAX7	100	100	100	100	Myopathy, congenital, progressive, with scoliosis, 618578 Rhabdomyosarcoma 2, alveolar, 268220
PAX9	99,7	99,6	100	100	Tooth agenesis, selective, 3, 604625
PGM1	94,2	94,2	94,2	94,2	Congenital disorder of glycosylation, type It, 614921
PITX2	99,9	97,7	100	100	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PLCB4	99,9	98,8	100	100	Auriculocondylar syndrome 2, 614669
POLR1C	90,5	87	82,8	82,8	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	91,6	91,6	100	100	Treacher Collins syndrome 2, 613717
POR	99,8	98,6	100	100	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750

PORCN	100	99,1	100	100	Focal dermal hypoplasia, 305600
PTCH1	99,2	97,6	99,9	99,8	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTH1R	100	98,7	100	100	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Failure of tooth eruption, primary, 125350 Eiken syndrome, 600002 Chondrodysplasia, Blomstrand type, 215045
RAB23	100	99,5	100	100	Carpenter syndrome, 201000
RAD21	99,2	96,6	100	100	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RBM10	99,5	97,1	100	100	TARP syndrome, 311900
RECQL4	99,8	98,1	100	99,9	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
RIPK4	100	99,9	100	100	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
RUNX2	72,2	72,2	100	100	Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600
SALL1	99,9	99	100	100	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	98,6	96,7	100	100	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SATB2	99,7	97,4	100	100	Glass syndrome, 612313
SCARF2	95,4	86,2	99,8	99,2	Van den Ende-Gupta syndrome, 600920
SEC24D	100	99,7	100	100	Cole-Carpenter syndrome 2, 616294
SEMA3E	99,2	98,9	100	100	?CHARGE syndrome, 214800
SF3B4	99,9	97,3	100	100	Acrofacial dysostosis 1, Nager type, 154400
SH3BP2	91,4	91,2	97	95,3	Cherubism, 118400
SHH	100	99,5	100	100	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638

					Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SIX1	100	99,2	100	100	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389
SIX3	99,9	98,6	100	100	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SKI	99,3	94,9	100	99,4	Shprintzen-Goldberg syndrome, 182212
SLC24A4	100	99,8	100	100	[Skin/hair/eye pigmentation 6, blue/green eyes], 210750 Amelogenesis imperfecta, type IIA5, 615887 [Skin/hair/eye pigmentation 6, blond/brown hair], 210750
SLC26A2	100	100	100	100	De la Chapelle dysplasia, 256050 Atelosteogenesis, type II, 256050 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Diastrophic dysplasia, 222600 Achondrogenesis Ib, 600972 Epiphyseal dysplasia, multiple, 4, 226900
SMAD6	90,9	81	100	99,6	Aortic valve disease 2, 614823 {Radioulnar synostosis, nonsyndromic}, 179300 {Craniosynostosis 7, susceptibility to}, 617439
SMC1A	100	98,7	100	99,8	Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 Cornelia de Lange syndrome 2, 300590
SMC3	95,2	91	100	100	Cornelia de Lange syndrome 3, 610759
SMO	97,8	94,7	100	100	Curry-Jones syndrome, somatic mosaic, 601707 Pallister-Hall-like syndrome, 241800 Basal cell carcinoma, somatic, 605462
SMOC2	76,8	76,6	100	100	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNAI2	100	99,1	100	100	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SOX10	99,9	97,9	100	100	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SOX6	99,9	99,4	100	100	Tolchin-Le Caignec syndrome, 618971
SOX9	100	98,6	100	100	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290

SPECC1L	96	95,7	97,8	96,2	Hypertelorism, Teebi type, 145420 ?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
SUMO1	67,2	49,9	69,4	69,4	?Orofacial cleft 10, 613705
TBX1	87	77,5	94	89,9	Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500
TBX22	99,2	95,7	100	100	?Abruzzo-Erickson syndrome, 302905 Cleft palate with ankyloglossia, 303400
TCF12	100	99,9	100	100	Craniosynostosis 3, 615314
TCOF1	99,7	98,6	100	100	Treacher Collins syndrome 1, 154500
TFAP2A	99,4	94,3	100	100	Branchiooculofacial syndrome, 113620
TGFBR1	93,7	93,6	99	96,3	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	100	100	100	100	Esophageal cancer, somatic, 133239 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Loeys-Dietz syndrome 2, 610168
TGIF1	100	100	100	100	Holoprosencephaly 4, 142946
TLK2	99,1	95,1	100	100	Mental retardation, autosomal dominant 57, 618050
TP63	100	100	100	100	Limb-mammary syndrome, 603543 Orofacial cleft 8, 618149 Split-hand/foot malformation 4, 605289 Hay-Wells syndrome, 106260 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Rapp-Hodgkin syndrome, 129400 ADULT syndrome, 103285
TRAF6	97,1	88,9	100	100	No OMIM disease ID
TSHZ1	98,8	98,8	100	100	Aural atresia, congenital, 607842
TSPEAR	100	99,2	100	100	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 ?Deafness, autosomal recessive 98, 614861
TWIST1	100	98,9	97,2	92,3	Robinow-Sorauf syndrome, 180750 Craniosynostosis 1, 123100

					Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
UBB	100	99,4	100	100	No OMIM disease ID
VAX1	97,5	91,5	95,7	91,7	?Microphthalmia, syndromic 11, 614402
WDR19	100	99,4	100	100	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376
WDR35	99,8	98,9	100	100	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR72	96,8	96,4	96,9	96,9	Amelogenesis imperfecta, type IIA3, 613211
WNT10A	100	99,4	100	100	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	100	99,4	100	100	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
ZEB2	99,9	99,1	97,4	97,4	Mowat-Wilson syndrome, 235730
ZIC1	100	100	100	100	Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 ?Craniosynostosis 6, 616602
ZIC2	100	98,7	98,5	95,7	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.
Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.
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 coverage denoting NC are non-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors