CRANIOFACIAL ANOMALIES GENE PANEL DG 2.18 (171 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ACP4	97,20%	88,80%	100%	100%	Amelogenesis imperfecta, type IJ, 617297
ADAMTSL4	100%	99,20%	100%	100%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ALX1	99,70%	97,10%	100%	100%	Frontonasal dysplasia 3, 613456
ALX3	77,90%	73,30%	100%	100%	Frontonasal dysplasia 1, 136760
ALX4	100%	99,30%	100%	100%	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597
AMBN	99,80%	98,50%	100%	100%	Amelogenesis imperfecta, type IF, 616270
AMELX	99,90%	96,80%	100%	100%	Amelogenesis imperfecta, type 1E, 301200
AMER1	99,90%	98,50%	100%	100%	Osteopathia striata with cranial sclerosis, 300373
AMTN	99,60%	98,60%	100%	100%	?Amelogenesis imperfecta, type IIIB, 617607
ANKRD11	97,50%	94,80%	100%	100%	KBG syndrome, 148050
ARHGAP29	99,50%	98,00%	100%	100%	No OMIM disease ID
AXIN2	100%	99,90%	100%	99,90%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
BCOR	99,60%	97,40%	100%	99,90%	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
BMP4	100%	100%	100%	100%	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C4orf26	100%	100%	100%	100%	Amelogenesis imperfecta, type IIA4, 614832
CDC45	99,80%	98,50%	100%	100%	Meier-Gorlin syndrome 7, 617063
CDON	100%	99,60%	100%	100%	Holoprosencephaly 11, 614226
CDSN	100%	100%	100%	100%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CHD7	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
COL11A1	99,20%	95,70%	100%	100%	Stickler syndrome, type II, 604841 Marshall syndrome, 154780

					?Deafness, autosomal dominant 37, 618533 Fibrochondrogenesis 1, 228520
COL11A2	100%	99,50%	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%	99,70%	100%	100%	Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 Kniest dysplasia, 156550 Stickler syndrome, type I, 108300 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 Avascular necrosis of the femoral head, 608805 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%	99,20%	100%	100%	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	99,90%	99,00%	100%	100%	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	98,70%	95,50%	99,70%	98,60%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969
CTSK	100%	99,90%	100%	100%	Pycnodysostosis, 265800
DHODH	100%	100%	100%	100%	Miller syndrome, 263750
DISP1	100%	99,90%	100%	100%	No OMIM disease ID
DLX3	99,90%	98,40%	100%	100%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510
DLX4	100%	100%	100%	100%	?Orofacial cleft 15, 616788
DSPP	96,80%	86,10%	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	98,10%	91,60%	100%	99,90%	Tooth agenesis, selective, X-linked 1, 313500 Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	100%	99,90%	100%	100%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900
EDARADD	99,90%	98,80%	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
EDNRA	100%	100%	100%	100%	Mandibulofacial dysostosis with alopecia, 616367
EFNA4	100%	100%	100%	100%	No OMIM disease ID
EFNB1	100%	100%	100%	100%	Craniofrontonasal dysplasia, 304110
EFTUD2	100%	99,80%	100%	100%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF4A3	100%	99,50%	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	99,90%	98,50%	100%	100%	Craniosynostosis 4, 600775 Chitayat syndrome, 617180
EYA1	99,90%	99,70%	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%	99,50%	100%	100%	Weaver syndrome, 277590
FAM20A	99,60%	94,70%	100%	100%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM83H	99,00%	95,00%	100%	100%	Amelogenesis imperfecta, type IIIA, 130900
FGD1	97,30%	92,80%	100%	100%	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF10	100%	99,80%	100%	100%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF3	99,80%	95,10%	100%	100%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF8	98,20%	88,90%	100%	99,60%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGF9	100%	100%	100%	100%	Multiple synostoses syndrome 3, 612961
FGFR1	100%	99,90%	100%	100%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 HEARTsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001

FGFR2	97,70%	97,10%	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	99,80%	97,70%	100%	99,80%	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
FOXC1	98,00%	89,60%	99,90%	98,50%	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXE1	96,90%	78,50%	99,90%	99,10%	Bamforth-Lazarus syndrome, 241850
GDF3	100%	100%	100%	100%	Microphthalmia, isolated 7, 613704 Microphthalmia with coloboma 6, 613703 Klippel-Feil syndrome 3, autosomal dominant, 613702
GDF6	100%	99,90%	100%	99,40%	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	99,10%	97,40%	100%	99,80%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	100%	99,50%	100%	100%	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GNAI3	99,30%	95,20%	100%	100%	Auriculocondylar syndrome 1, 602483
GPR68	99,50%	96,70%	100%	100%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GRHL3	100%	100%	100%	100%	Van der Woude syndrome 2, 606713
GSC	99,20%	92,40%	100%	100%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
HOXA2	100%	99,90%	100%	100%	Microtia with or without hearing impairment (AD), 612290 ?Microtia, hearing impairment, and cleft palate (AR), 612290
HUWE1	99,20%	95,80%	100%	100%	Mental retardation, X-linked syndromic, Turner type, 309590
HYAL2	100%	100%	100%	100%	No OMIM disease ID
IFT122	100%	99,60%	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	99,60%	97,30%	100%	100%	No OMIM disease ID
IKBKG	84,10%	77,20%	100%	100%	Immunodeficiency 33, 300636 Incontinentia pigmenti, 308300 Immunodeficiency, isolated, 300584 Ectodermal dysplasia and immunodeficiency 1, 300291

					Invasive pneumococcal disease, recurrent isolated, 2, 300640 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301
IL11RA	100%	99,90%	100%	100%	Craniosynostosis and dental anomalies, 614188
INTU	99,70%	98,10%	100%	100%	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
IRF6	99,60%	95,90%	100%	100%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300
ITGB6	97,20%	95,80%	100%	100%	Amelogenesis imperfecta, type IH, 616221
KAT6B	99,90%	99,00%	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KDF1	100%	99,80%	100%	100%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	98,20%	95,20%	100%	100%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM6A	96,10%	88,70%	100%	99,90%	Kabuki syndrome 2, 300867
KLK4	100%	100%	100%	100%	Amelogenesis imperfecta, type IIA1, 204700
KMT2D	100%	99,40%	100%	100%	Kabuki syndrome 1, 147920
KREMEN1	97,70%	94,40%	99,50%	97,90%	Ectodermal dysplasia 13, hair/tooth type, 617392
LAMB3	100%	99,60%	100%	100%	Amelogenesis imperfecta, type IA, 104530 Epidermolysis bullosa, junctional, Herlitz type, 226700 Epidermolysis bullosa, junctional, non-Herlitz type, 226650
LRP2	100%	99,90%	100%	100%	Donnai-Barrow syndrome, 222448
LRP6	100%	99,90%	100%	100%	Tooth agenesis, selective, 7, 616724
LTBP3	99,60%	98,10%	100%	100%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
MASP1	100%	99,90%	100%	100%	3MC syndrome 1, 257920
MED12	99,80%	96,70%	100%	100%	Ohdo syndrome, X-linked, 300895 Lujan-Fryns syndrome, 309520 Opitz-Kaveggia syndrome, 305450
MEGF8	99,90%	99,00%	100%	100%	Carpenter syndrome 2, 614976
MEIS2	100%	100%	100%	100%	Cleft palate, cardiac defects, and mental retardation, 600987
MEOX1	100%	98,90%	100%	100%	Klippel-Feil syndrome 2, 214300
MID1	99,80%	98,70%	100%	100%	Opitz GBBB syndrome, type I, 300000
MITF	100%	99,90%	100%	100%	COMMAD syndrome, 617306 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

MSX1	96,90%	89,30%	100%	100%	Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	100%	99,40%	100%	100%	Parietal foramina 1, 168500 Craniosynostosis 2, 604757 Parietal foramina with cleidocranial dysplasia, 168550
NAA10	99,70%	98,50%	99,90%	99,90%	Ogden syndrome, 300855 ?Microphthalmia, syndromic 1, 309800
NECTIN1	100%	99,90%	100%	100%	Orofacial cleft 7, 225060 Cleft lip/palate-ectodermal dysplasia syndrome, 225060
NFKBIA	95,20%	88,00%	100%	100%	Ectodermal dysplasia and immunodeficiency 2, 612132
NIPBL	98,90%	97,00%	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%	99,90%	100%	100%	Sotos syndrome 1, 117550
OFD1	88,00%	73,70%	100%	99,90%	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
OTX2	100%	99,70%	100%	100%	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
PAX3	100%	99,90%	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 ?Coloboma of optic nerve, 120430 ?Morning glory disc anomaly, 120430 Cataract with late-onset corneal dystrophy, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229

PAX7	100%	100%	100%	100%	Myopathy, congenital, progressive, with scoliosis, 618578 Rhabdomyosarcoma 2, alveolar, 268220
PAX9	99,70%	99,60%	100%	100%	Tooth agenesis, selective, 3, 604625
PGM1	100%	100%	100%	100%	Congenital disorder of glycosylation, type It, 614921
PITX2	99,90%	97,70%	100%	100%	Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550 Anterior segment dysgenesis 4, 137600
PLCB4	99,90%	98,80%	100%	100%	Auriculocondylar syndrome 2, 614669
POLR1C	99,30%	95,50%	90,70%	90,70%	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	91,60%	91,60%	100%	100%	Treacher Collins syndrome 2, 613717
PORCN	100%	99,10%	100%	100%	Focal dermal hypoplasia, 305600
PTCH1	99,20%	97,60%	99,90%	99,80%	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTH1R	100%	98,70%	100%	100%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Failure of tooth eruption, primary, 125350 Eiken syndrome, 600002 Chondrodysplasia, Blomstrand type, 215045
RAB23	100%	99,50%	100%	100%	Carpenter syndrome, 201000
RAD21	99,20%	96,60%	100%	100%	?Mungan syndrome, 611376 Cornelia de Lange syndrome 4, 614701
RBM10	99,50%	97,10%	100%	100%	TARP syndrome, 311900
RECQL4	99,80%	98,10%	100%	99,90%	RAPADILINO syndrome, 266280 Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2,, 268400
RIPK4	100%	99,90%	100%	100%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
RUNX2	72,20%	72,20%	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	99,90%	99,00%	100%	100%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	98,60%	96,70%	100%	100%	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SATB2	99,70%	97,40%	100%	100%	Glass syndrome, 612313
SEMA3E	100%	99,60%	100%	100%	?CHARGE syndrome, 214800

SF3B4	99,90%	97,30%	100%	100%	Acrofacial dysostosis 1, Nager type, 154400
SH3BP2	91,40%	91,20%	97,00%	95,30%	Cherubism, 118400
SHH	100%	99,50%	100%	100%	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SIX1	100%	99,20%	100%	100%	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389
SIX3	99,90%	98,60%	100%	100%	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SKI	99,30%	94,90%	100%	99,40%	Shprintzen-Goldberg syndrome, 182212
SLC24A4	100%	99,80%	100%	100%	Amelogenesis imperfecta, type IIA5, 615887
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	90,90%	81,00%	100%	99,60%	Aortic valve disease 2, 614823
SMC1A	100%	98,70%	100%	99,80%	Cornelia de Lange syndrome 2, 300590 Epileptic encephalopathy, early infantile, 85, with or without midline brain defects, 301044
SMC3	95,20%	91,00%	100%	100%	Cornelia de Lange syndrome 3, 610759
SMO	97,80%	94,70%	100%	100%	Curry-Jones syndrome, somatic mosaic, 601707 Basal cell carcinoma, somatic, 605462
SMOC2	77,00%	76,70%	100%	100%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNAI2	100%	99,10%	100%	100%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SOX10	99,90%	97,90%	100%	100%	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SOX6	99,90%	99,40%	100%	100%	No OMIM disease ID
SOX9	100%	98,60%	100%	100%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SPECC1L	100%	99,60%	100%	100%	Hypertelorism, Teebi type, 145420 ?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
SUMO1	67,20%	49,90%	69,40%	69,40%	?Orofacial cleft 10, 613705

TBX1	86,90%	79,50%	94,10%	90,80%	Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500 Conotruncal anomaly face syndrome, 217095
TBX22	99,20%	95,70%	100%	100%	?Abruzzo-Erickson syndrome, 302905 Cleft palate with ankyloglossia, 303400
TCF12	100%	99,90%	100%	100%	Craniosynostosis 3, 615314
TCOF1	99,70%	98,60%	100%	100%	Treacher Collins syndrome 1, 154500
TFAP2A	99,40%	94,30%	100%	100%	Branchiooculofacial syndrome, 113620
TGFBR1	93,70%	93,60%	99,00%	96,30%	Loeys-Dietz syndrome 1, 609192
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
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	97,10%	88,90%	100%	100%	No OMIM disease ID
TSHZ1	98,80%	98,80%	100%	100%	Aural atresia, congenital, 607842
TSPEAR	100%	99,20%	97,90%	97,90%	Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 ?Deafness, autosomal recessive 98, 614861
TWIST1	100%	98,90%	97,20%	92,30%	Robinow-Sorauf syndrome, 180750 Craniosynostosis 1, 123100 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
UBB	100%	99,40%	100%	100%	No OMIM disease ID
VAX1	97,50%	91,50%	95,70%	91,70%	?Microphthalmia, syndromic 11, 614402
WDR19	100%	99,40%	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	99,80%	98,90%	100%	100%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR72	96,80%	96,40%	96,90%	96,90%	Amelogenesis imperfecta, type IIA3, 613211

WNT10A	100%	99,40%	100%	100%	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	100%	99,40%	100%	100%	Split-hand/foot malformation 6, 225300 Tooth agenesis, selective, 8, 617073
ZEB2	99,90%	99,10%	97,40%	97,40%	Mowat-Wilson syndrome, 235730
ZIC1	100%	100%	100%	100%	Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 ?Craniosynostosis 6, 616602
ZIC2	100%	98,70%	98,50%	95,70%	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 : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors