CRANIOFACIAL ANOMALIES GENE PANEL DG 3.3.0 (189 genes)

Releasedate: 13-01-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
ACP4	100%	100%	Amelogenesis imperfecta, type IJ, 617297
ACTG1	100%	100%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ADAMTSL4	100%	100%	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ALX1	100%	100%	Frontonasal dysplasia 3, 613456
ALX3	100%	100%	Frontonasal dysplasia 1, 136760
ALX4	100%	100%	Parietal foramina 2, 609597 Frontonasal dysplasia 2, 613451
AMBN	100%	100%	Amelogenesis imperfecta, type IF, 616270
AMELX	100%	100%	Amelogenesis imperfecta, type 1E, 301200
AMER1	100%	100%	Osteopathia striata with cranial sclerosis, 300373
AMTN	100%	100%	?Amelogenesis imperfecta, type IIIB, 617607
ANKRD11	100%	100%	KBG syndrome, 148050
ARHGAP29	100%	100%	No OMIM disease ID
AXIN2	100%	100%	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
BCOR	100%	100%	Microphthalmia, syndromic 2, 300166
BMP2	100%	100%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 Brachydactyly, type A2, 112600
BMP4	100%	100%	Orofacial cleft 11, 600625 Microphthalmia, syndromic 6, 607932
BPNT2	100%	100%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
CCBE1	100%	100%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CDC45	100%	100%	Meier-Gorlin syndrome 7, 617063
CDON	100%	100%	Holoprosencephaly 11, 614226
CDSN	100%	100%	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300

CHD7	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
COL11A1	100%	100%	Fibrochondrogenesis 1, 228520 Stickler syndrome, type II, 604841 Marshall syndrome, 154780 Deafness, autosomal dominant 37, 618533
COL11A2	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%	100%	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248 Czech dysplasia, 609162 Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 SMED Strudwick type, 184250 Stickler sydrome, type I, nonsyndromic ocular, 609508 ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 SED congenita, 183900 Kniest dysplasia, 156550 Osteoarthritis with mild chondrodysplasia, 604864 Stickler syndrome, type I, 108300 Platyspondylic skeletal dysplasia, Torrance type, 151210 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Avascular necrosis of the femoral head, 608805 Legg-Calve-Perthes disease, 150600
COL9A1	100%	100%	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135
COL9A2	100%	100%	Epiphyseal dysplasia, multiple, 2, 600204 ?Stickler syndrome, type V, 614284
COL9A3	100%	100%	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969
COLEC11	100%	100%	3MC syndrome 2, 265050
CTSK	100%	100%	Pycnodysostosis, 265800
CYP26B1	100%	100%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
DHODH	100%	100%	Miller syndrome, 263750
DISP1	100%	100%	No OMIM disease ID
DLX3	100%	100%	Trichodontoosseous syndrome, 190320 Amelogenesis imperfecta, type IV, 104510

DLX4	100%	100%	?Orofacial cleft 15, 616788
DSPP	100%	100%	Dentinogenesis imperfecta, Shields type III, 125500 Dentinogenesis imperfecta, Shields type II, 125490 Dentin dysplasia, type II, 125420 Deafness, autosomal dominant 39, with dentinogenesis, 605594
EDA	100%	100%	Tooth agenesis, selective, X-linked 1, 313500 Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	100%	100%	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900
EDARADD	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%	Question mark ears, isolated, 612798 Auriculocondylar syndrome 3, 615706
EDNRA	100%	100%	Mandibulofacial dysostosis with alopecia, 616367
EFNA4	100%	100%	No OMIM disease ID
EFNB1	100%	100%	Craniofrontonasal dysplasia, 304110
EFTUD2	100%	100%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF4A3	100%	100%	Robin sequence with cleft mandible and limb anomalies, 268305
ENAM	100%	100%	Amelogenesis imperfecta, type IC, 204650 Amelogenesis imperfecta, type IB, 104500
ERF	100%	100%	Craniosynostosis 4, 600775 Chitayat syndrome, 617180
ESCO2	100%	100%	Juberg-Hayward syndrome, 216100 Roberts-SC phocomelia syndrome, 268300
EYA1	100%	100%	Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 602588 ?Otofaciocervical syndrome, 166780
EZH2	100%	100%	Weaver syndrome, 277590
FAM20A	100%	100%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM83H	100%	100%	Amelogenesis imperfecta, type IIIA, 130900
FGD1	100%	100%	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF10	100%	100%	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF3	100%	100%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF8	100%	100%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702

FGF9	100%	100%	Multiple synostoses syndrome 3, 612961
FGFR1	100%	100%	Pfeiffer syndrome, 101600 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Hartsfield syndrome, 615465 Trigonocephaly 1, 190440 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	100%	100%	Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Jackson-Weiss syndrome, 123150 Gastric cancer, somatic, 613659 Craniofacial-skeletal-dermatologic dysplasia, 101600 Apert syndrome, 101200 Pfeiffer syndrome, 101600 Beare-Stevenson cutis gyrata syndrome, 123790 Crouzon syndrome, 123500 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, Craniosynostosis, nonspecific,
FGFR3	100%	100%	Muenke syndrome, 602849 SADDAN, 616482 Hypochondroplasia, 146000 LADD syndrome, 149730 Thanatophoric dysplasia, type II, 187601 Nevus, epidermal, somatic, 162900 CATSHL syndrome, 610474 Thanatophoric dysplasia, type I, 187600 Spermatocytic seminoma, somatic, 273300 Bladder cancer, somatic, 109800 Achondroplasia, 100800 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247
FLNA	100%	100%	Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048

			Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Melnick-Needles syndrome, 309350 Terminal osseous dysplasia, 300244 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type I, 311300 Heterotopia, periventricular, 1, 300049 Frontometaphyseal dysplasia 1, 305620
FLNB	100%	100%	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Spondylocarpotarsal synostosis syndrome, 272460 Boomerang dysplasia, 112310
FOXC1	100%	100%	Axenfeld-Rieger syndrome, type 3, 602482 Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXE1	100%	100%	Bamforth-Lazarus syndrome, 241850
GDF3	100%	100%	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF6	100%	100%	Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Leber congenital amaurosis 17, 615360 Multiple synostoses syndrome 4, 617898 Klippel-Feil syndrome 1, autosomal dominant, 118100
GJA1	100%	100%	Erythrokeratodermia variabilis et progressiva 3, 617525 Craniometaphyseal dysplasia, autosomal recessive, 218400 Oculodentodigital dysplasia, 164200 Hypoplastic left heart syndrome 1, 241550 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100 Oculodentodigital dysplasia, autosomal recessive, 257850 Atrioventricular septal defect 3, 600309
GJB6	100%	100%	Ectodermal dysplasia 2, Clouston type, 129500 Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290
GLI2	100%	100%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829

GLI3	100%	100%	Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700
GNAI3	100%	100%	Auriculocondylar syndrome 1, 602483
GNPTAB	100%	100%	Mucolipidosis III alpha/beta, 252600 Mucolipidosis II alpha/beta, 252500
GPR68	100%	100%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217
GRHL3	100%	100%	Van der Woude syndrome 2, 606713
GSC	100%	100%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
HOXA2	100%	100%	Microtia with or without hearing impairment (AD), 612290 ?Microtia, hearing impairment, and cleft palate (AR), 612290
HUWE1	100%	100%	Intellectual developmental disorder, X-linked, Turner type, 309590
HYAL2	100%	100%	No OMIM disease ID
IFT122	100%	100%	Cranioectodermal dysplasia 1, 218330
IFT43	100%	100%	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT88	100%	100%	No OMIM disease ID
IKBKG	100%	100%	Incontinentia pigmenti, 308300 Ectodermal dysplasia and immunodeficiency 1, 300291 Immunodeficiency 33, 300636
IL11RA	100%	100%	Craniosynostosis and dental anomalies, 614188
IL6ST	100%	100%	Hyper-IgE recurrent infection syndrome 4, autosomal recessive, 618523
INTU	100%	100%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
IRF6	100%	100%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300
ITGB6	100%	100%	Amelogenesis imperfecta, type IH, 616221
KAT6B	100%	100%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KDF1	100%	100%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337
KDM1A	100%	100%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728
KDM6A	100%	100%	Kabuki syndrome 2, 300867
KLK4	100%	100%	Amelogenesis imperfecta, type IIA1, 204700
KMT2D	100%	100%	Kabuki syndrome 1, 147920
KREMEN1	100%	100%	Ectodermal dysplasia 13, hair/tooth type, 617392

LAMB3	100%	100%	Epidermolysis bullosa, junctional, non-Herlitz type, 226650 Epidermolysis bullosa, junctional, Herlitz type, 226700 Amelogenesis imperfecta, type IA, 104530
LRP2	100%	100%	Donnai-Barrow syndrome, 222448
LRP6	100%	100%	Tooth agenesis, selective, 7, 616724
LTBP3	100%	100%	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
MASP1	100%	100%	3MC syndrome 1, 257920
MED12	100%	100%	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MEGF8	100%	100%	Carpenter syndrome 2, 614976
MEIS2	100%	100%	Cleft palate, cardiac defects, and mental retardation, 600987
MEOX1	100%	100%	Klippel-Feil syndrome 2, 214300
MID1	100%	100%	Opitz GBBB syndrome, type I, 300000
MITF	100%	100%	Waardenburg syndrome, type 2A, 193510 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome/ocular albinism, digenic, 103470 COMMAD syndrome, 617306
MMP20	100%	100%	Amelogenesis imperfecta, type IIA2, 612529
MN1	100%	100%	CEBALID syndrome, 618774 Meningioma, 607174
MSX1	100%	100%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874
MSX2	100%	100%	Parietal foramina with cleidocranial dysplasia, 168550 Craniosynostosis 2, 604757 Parietal foramina 1, 168500
NAA10	100%	100%	Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NECTIN1	100%	100%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NFKBIA	100%	100%	Ectodermal dysplasia and immunodeficiency 2, 612132
NIPBL	100%	100%	Cornelia de Lange syndrome 1, 122470
NOG	100%	100%	Symphalangism, proximal, 1A, 185800 Brachydactyly, type B2, 611377 Stapes ankylosis with broad thumbs and toes, 184460

			Tarsal-carpal coalition syndrome, 186570 Multiple synostoses syndrome 1, 186500
NSD1	100%	100%	Sotos syndrome 1, 117550
ODAPH	100%	100%	Amelogenesis imperfecta, type IIA4, 614832
OFD1	100%	100%	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
OTX2	100%	100%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 Pituitary hormone deficiency, combined, 6, 613986 Microphthalmia, syndromic 5, 610125
P4HB	100%	100%	Cole-Carpenter syndrome 1, 112240
PAX3	100%	100%	Craniofacial-deafness-hand syndrome, 122880 Waardenburg syndrome, type 3, 148820 Waardenburg syndrome, type 1, 193500 Rhabdomyosarcoma 2, alveolar, 268220
PAX6	100%	100%	Optic nerve hypoplasia, 165550 Cataract with late-onset corneal dystrophy, 106210 ?Coloboma, ocular, 120200 ?Coloboma of optic nerve, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 ?Morning glory disc anomaly, 120430 Foveal hypoplasia 1, 136520 Keratitis, 148190
PAX7	100%	100%	Rhabdomyosarcoma 2, alveolar, 268220 Myopathy, congenital, progressive, with scoliosis, 618578
PAX9	100%	100%	Tooth agenesis, selective, 3, 604625
PGM1	94%	94%	Congenital disorder of glycosylation, type It, 614921
PITX2	100%	100%	Ring dermoid of cornea, 180550 Axenfeld-Rieger syndrome, type 1, 180500 Anterior segment dysgenesis 4, 137600
PLCB4	100%	100%	Auriculocondylar syndrome 2, 614669
POLR1C	83%	82%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	100%	100%	Treacher Collins syndrome 2, 613717
POR	100%	100%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571

PORCN	100%	100%	Focal dermal hypoplasia, 305600
PTCH1	100%	100%	Basal cell carcinoma, somatic, 605462 Holoprosencephaly 7, 610828 Basal cell nevus syndrome, 109400
PTH1R	100%	100%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Chondrodysplasia, Blomstrand type, 215045
RAB23	100%	100%	Carpenter syndrome, 201000
RAD21	100%	100%	Cornelia de Lange syndrome 4, 614701 ?Mungan syndrome, 611376
RBM10	100%	100%	TARP syndrome, 311900
RECQL4	100%	100%	Baller-Gerold syndrome, 218600 Rothmund-Thomson syndrome, type 2, 268400 RAPADILINO syndrome, 266280
RIPK4	100%	100%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RUNX2	100%	100%	Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, 119600
SALL1	100%	100%	Townes-Brocks syndrome 1, 107480 Townes-Brocks branchiootorenal-like syndrome, 107480
SALL4	100%	100%	?IVIC syndrome, 147750 Duane-radial ray syndrome, 607323
SATB2	100%	100%	Glass syndrome, 612313
SCARF2	100%	100%	Van den Ende-Gupta syndrome, 600920
SEC24D	100%	100%	Cole-Carpenter syndrome 2, 616294
SEMA3E	100%	100%	?CHARGE syndrome, 214800
SF3B2	100%	100%	No OMIM disease ID
SF3B4	100%	100%	Acrofacial dysostosis 1, Nager type, 154400
SH3BP2	99%	98%	Cherubism, 118400
SHH	100%	100%	Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SIX1	100%	100%	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389

SIX3	100%	100%	Schizencephaly, 269160 Holoprosencephaly 2, 157170
SIX5	100%	100%	Branchiootorenal syndrome 2, 610896
SKI	100%	100%	Shprintzen-Goldberg syndrome, 182212
SLC24A4	100%	100%	Amelogenesis imperfecta, type IIA5, 615887
SLC26A2	100%	100%	Epiphyseal dysplasia, multiple, 4, 226900 De la Chapelle dysplasia, 256050 Diastrophic dysplasia, 222600 Diastrophic dysplasia, broad bone-platyspondylic variant, 222600 Achondrogenesis Ib, 600972 Atelosteogenesis, type II, 256050
SMAD6	100%	100%	Aortic valve disease 2, 614823
SMC1A	100%	100%	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	100%	100%	Cornelia de Lange syndrome 3, 610759
SMO	100%	100%	Pallister-Hall-like syndrome, 241800 Basal cell carcinoma, somatic, 605462 Curry-Jones syndrome, somatic mosaic, 601707
SMOC2	100%	100%	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNAI2	100%	100%	Waardenburg syndrome, type 2D, 608890 Piebaldism, 172800
SOX10	100%	100%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX6	100%	100%	Tolchin-Le Caignec syndrome, 618971
SOX9	100%	100%	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290
SPECC1L	97%	96%	Opitz GBBB syndrome, type II, 145410 Teebi hypertelorism syndrome, 145420 ?Facial clefting, oblique, 1, 600251
SUMO1	69%	69%	?Orofacial cleft 10, 613705
TBX1	97%	94%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX22	100%	100%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905

TCF12	100%	100%	Craniosynostosis 3, 615314
TCOF1	100%	100%	Treacher Collins syndrome 1, 154500
TFAP2A	100%	100%	Branchiooculofacial syndrome, 113620
TGFBR1	100%	99%	Loeys-Dietz syndrome 1, 609192
TGFBR2	100%	100%	Loeys-Dietz syndrome 2, 610168 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239
TGIF1	100%	100%	Holoprosencephaly 4, 142946
TLK2	100%	100%	Mental retardation, autosomal dominant 57, 618050
TP63	100%	100%	Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Split-hand/foot malformation 4, 605289 Orofacial cleft 8, 618149 Rapp-Hodgkin syndrome, 129400 ADULT syndrome, 103285 Limb-mammary syndrome, 603543
TRAF6	100%	100%	No OMIM disease ID
TSHZ1	100%	100%	Aural atresia, congenital, 607842
TSPEAR	100%	100%	?Deafness, autosomal recessive 98, 614861 Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180
TWIST1	100%	99%	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
UBB	100%	100%	No OMIM disease ID
VAX1	99%	99%	?Microphthalmia, syndromic 11, 614402
WDR19	100%	100%	Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 ?Cranioectodermal dysplasia 4, 614378
WDR35	100%	100%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WDR72	96%	96%	Amelogenesis imperfecta, type IIA3, 613211
WNT10A	100%	100%	Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400 Odontoonychodermal dysplasia, 257980
WNT10B	100%	100%	Tooth agenesis, selective, 8, 617073 Split-hand/foot malformation 6, 225300

ZEB2	97%	97%	Mowat-Wilson syndrome, 235730
ZIC1	100%	100%	?Craniosynostosis 6, 616602 Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736
ZIC2	100%	99%	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.
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TWIST is the chemistry used for 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-protein coding genes.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : January 13th , 2022.
This list is accurate for panel version DG 3.3.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} s ignify risk factors