OROFACIAL CLEFTING PANEL WITH GENOME WIDE CNV ANALYSIS DG-5.1.0 (217 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
ACTB	100%	100%	100%	99.9%	99.4%	Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620470
ACTG1	100%	100%	100%	99.9%	98.9%	Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583
ALX1	100%	100%	100%	100%	99.7%	Frontonasal dysplasia 3, 613456
ALX3	100%	100%	100%	99.5%	98.7%	Frontonasal dysplasia 1, 136760
AMER1	100%	100%	98.9%	90.5%	71.9%	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	99.9%	97.7%	99.1%	88.8%	71.2%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
AMOTL1	100%	100%	100%	100%	99.5%	Craniofaciocardiohepatic syndrome, 621192

ANKRD11	100%	100%	100%	100%	99.2%	KBG syndrome, 148050
ARHGAP29	100%	100%	100%	100%	99.8%	
ARHGAP31	100%	100%	100%	100%	99.6%	Adams-Oliver syndrome 1, 100300
ASXL1	100%	100%	100%	100%	99.8%	Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039
B3GALT6	100%	98.6%	100%	99.9%	98.6%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465
B3GLCT	100%	100%	100%	100%	99.9%	Peters-plus syndrome, 261540
B4GALT7	86.8%	86.8%	100%	100%	99.9%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B9D2	100%	100%	100%	100%	99.7%	?Meckel syndrome 10, 614175;Joubert syndrome 34, 614175
BCOR	100%	100%	98.9%	89.4%	69.8%	Microphthalmia, syndromic 2, 300166
BMP2	100%	100%	100%	99.8%	97.7%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877;Brachydactyly, type A2, 112600;{HFE hemochromatosis, modifier of}, 235200
BMPER	100%	100%	100%	100%	99.6%	Diaphanospondylodysostosis, 608022

BPNT2	94.9%	94.9%	100%	100%	99.5%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
C2CD3	96%	96%	100%	100%	99.5%	Orofaciodigital syndrome XIV, 615948
CAMTA1	100%	100%	100%	100%	99.2%	Cerebellar dysfunction with variable cognitive and behavioral abnormalities, 614756
CC2D2A	98.2%	98.2%	100%	100%	99.8%	COACH syndrome 2, 619111;Retinitis pigmentosa 93, 619845;Meckel syndrome 6, 612284;Joubert syndrome 9, 612285
CCDC32	100%	100%	100%	99.9%	99.5%	Cardiofacioneurodevelopmental syndrome, 619123
CDC45	100%	100%	100%	100%	99.5%	Meier-Gorlin syndrome 7, 617063
CDH1	100%	100%	100%	100%	99.6%	Ovarian cancer, somatic, 167000;Blepharocheilodontic syndrome 1, 119580;Diffuse gastric and lobular breast cancer syndrome with or without cleft lip and/or palate, 137215;Endometrial carcinoma, somatic, 608089;Breast cancer, lobular, somatic, 114480
CDKN1C	100%	100%	100%	100%	99%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CHD7	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800

CHRNG	100%	100%	100%	99.9%	99.6%	Multiple pterygium syndrome, lethal type, 253290;Escobar syndrome, 265000
CHST14	100%	100%	100%	99.9%	98.4%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CILK1	100%	100%	100%	100%	99.8%	Cranioectodermal dysplasia 6, 621337;{Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924;Endocrine-cerebroosteodysplasia, 612651
COL11A1	100%	100%	100%	100%	99.8%	Fibrochondrogenesis 1, 228520;Stickler syndrome, type II, 604841;Marshall syndrome, 154780;Deafness, autosomal dominant 37, 618533;{Lumbar disc herniation, susceptibility to}, 603932
COL11A2	100%	100%	100%	100%	99.1%	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%	100%	100%	99.5%	?Vitreoretinopathy with phalangeal epiphyseal dysplasia, 619248;Czech dysplasia, 609162;Achondrogenesis, type II or hypochondrogenesis, 200610;Spondyloperipheral dysplasia, 271700;SMED Strudwick type, 184250;?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450;Spondylometaphyseal dysplasia, Algerian type, 184253;SED congenita, 183900;Kniest dysplasia, 156550;Stickler syndrome, type I, nonsyndromic ocular, 609508;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%	100%	100%	99.7%	Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135
COLEC10	95%	86.3%	100%	100%	99.8%	3MC syndrome 3, 248340
COLEC11	100%	100%	100%	100%	99.6%	3MC syndrome 2, 265050

CPLANE1	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome VI, 277170;Joubert syndrome 17, 614615
CTCF	100%	100%	100%	100%	99.4%	Intellectual developmental disorder, autosomal dominant 21, 615502
CTNND1	100%	100%	100%	100%	99.9%	Blepharocheilodontic syndrome 2, 617681
DDX3X	99.4%	98.7%	99.1%	90.6%	72.1%	Intellectual developmental disorder, X-linked syndromic, Snijders Blok type, 300958
DDX59	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome V, 174300
DHCR7	96.2%	96.2%	100%	100%	99.6%	Smith-Lemli-Opitz syndrome, 270400
DHODH	89%	86.1%	100%	100%	99.5%	Miller syndrome, 263750
DLL4	100%	100%	100%	100%	99.2%	Adams-Oliver syndrome 6, 616589
DOCK6	100%	100%	100%	99.9%	99.3%	Adams-Oliver syndrome 2, 614219
DVL1	100%	100%	100%	99.9%	99.3%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100%	100%	100%	99.9%	99.1%	Robinow syndrome, autosomal dominant 3, 616894
DYNC2H1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	81.5%	81.4%	100%	100%	99.8%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
EBP	100%	100%	98.8%	88.4%	69.5%	MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960

EDN1	100%	100%	100%	100%	99.8%	Question mark ears, isolated, 612798;Auriculocondylar syndrome 3, 615706
EDNRA	100%	100%	100%	100%	99.8%	{Migraine, resistance to}, 157300;Mandibulofacial dysostosis with alopecia, 616367
EFNB1	100%	100%	98.8%	87.7%	69.3%	Craniofrontonasal dysplasia, 304110
EFTUD2	100%	100%	100%	100%	99.7%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF2S3	100%	100%	99.2%	91%	73.9%	MEHMO syndrome, 300148
EIF4A3	100%	100%	100%	100%	99.4%	Robin sequence with cleft mandible and limb anomalies, 268305
EOGT	95.8%	92.4%	100%	100%	99.6%	Adams-Oliver syndrome 4, 615297
EPG5	100%	100%	100%	100%	99.8%	Vici syndrome, 242840;Neurodevelopmental disorder with parkinsonism or other movement abnormalities, 621506
ESCO2	100%	100%	100%	100%	99.8%	Juberg-Hayward syndrome, 216100;Roberts-SC phocomelia syndrome, 268300
EYA1	100%	100%	100%	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
FAM20C	99.4%	97.2%	100%	100%	99.2%	Raine syndrome, 259775

FGD1	100%	100%	98.4%	88%	69.3%	Aarskog-Scott syndrome, 305400
FGF8	100%	100%	100%	99.8%	98.3%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100%	99.2%	100%	100%	99.4%	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%	100%	100%	99.7%	Bent bone dysplasia syndrome, 614592;LADD syndrome 1, 149730;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410;Scaphocephaly and Axenfeld-Rieger anomaly;Jackson-Weiss syndrome, 123150;Gastric cancer, somatic, 613659;Craniofacial-skeletal-dermatologic dysplasia, 101600;Apert syndrome, 101200;Pfeiffer syndrome, 101600;Craniosynostosis, nonspecific;?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579;Beare-Stevenson cutis gyrata syndrome, 123790;Crouzon syndrome, 123500;Saethre-Chotzen syndrome, 101400

FLNA	100%	100%	98.3%	87.4%	68.7%	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%	100%	100%	99.6%	Larsen syndrome, 150250;Atelosteogenesis, type I, 108720;Atelosteogenesis, type III, 108721;Spondylocarpotarsal synostosis syndrome, 272460;Boomerang dysplasia, 112310
FOXC2	100%	100%	100%	99.8%	98.2%	Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	100%	100%	100%	99.9%	97.1%	Bamforth-Lazarus syndrome, 241850;{Thyroid cancer, nonmedullary, 4}, 616534
FRAS1	100%	100%	100%	100%	99.7%	Fraser syndrome 1, 219000

FTO	94.5%	94.5%	100%	100%	99.9%	Growth retardation, developmental delay, facial dysmorphism, 612938;{Obesity, susceptibility to, BMIQ14}, 612460
GDF6	100%	100%	100%	99.9%	98.2%	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%	100%	100%	99.7%	Erythrokeratodermia variabilis et progressiva 3, 617525;Craniometaphyseal dysplasia, autosomal recessive, 218400;Oculodentodigital dysplasia, 164200;Palmoplantar keratoderma with congenital alopecia, 104100;Syndactyly, type III, 186100;Oculodentodigital dysplasia, autosomal recessive, 257850
GLI2	100%	100%	100%	99.9%	99.3%	Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829
GLI3	99.3%	99.3%	100%	99.9%	99.2%	Greig cephalopolysyndactyly syndrome, 175700;Polydactyly, postaxial, types A1 and B, 174200;Pallister-Hall syndrome, 146510;Polydactyly, preaxial, type IV, 174700
GNAI3	100%	100%	100%	100%	99.9%	Auriculocondylar syndrome 1, 602483

GNB1	94.3%	94.3%	100%	100%	99.5%	Myelodysplastic syndrome, somatic, 614286;Leukemia, acute lymphoblastic, somatic, 613065;Intellectual developmental disorder, autosomal dominant 42, 616973
GPC3	94%	94%	98.6%	88.7%	70.8%	Wilms tumor, somatic, 194070;Simpson-Golabi-Behmel syndrome, type 1, 312870
GRHL3	100%	100%	100%	100%	99.7%	van der Woude syndrome 2, 606713
HDAC8	94.6%	94.6%	99.2%	90.3%	71.3%	Cornelia de Lange syndrome 5, 300882
HYAL2	100%	100%	100%	100%	99.5%	Muggenthaler-Chowdhury-Chioza syndrome, 621063
HYLS1	100%	100%	100%	100%	99.8%	Hydrolethalus syndrome, 236680
IFT140	100%	100%	100%	100%	99.6%	{Polycystic kidney disease 9, susceptibility to}, 621164;Short-rib thoracic dysplasia 9 with or without polydactyly, 266920;Retinitis pigmentosa 80, 617781;Cranioectodermal dysplasia 5, 621180
IFT172	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT57	92.4%	92.4%	100%	100%	99.9%	?Orofaciodigital syndrome XVIII, 617927

IFT80	100%	100%	100%	100%	99.6%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
INTU	100%	100%	100%	100%	99.7%	?Orofaciodigital syndrome XVII, 617926;?Short-rib thoracic dysplasia 20 with polydactyly, 617925
IRF6	100%	100%	100%	100%	99.4%	{Orofacial cleft 6}, 608864;Popliteal pterygium syndrome 1, 119500;van der Woude syndrome 1, 119300
KANSL1	98.1%	98.1%	100%	100%	99.8%	Koolen-De Vries syndrome, 610443
KAT6A	100%	100%	100%	100%	99.7%	Arboleda-Tham syndrome, 616268
KCNJ2	100%	100%	100%	100%	99.8%	Atrial fibrillation, familial, 9, 613980;Andersen syndrome, 170390;Short QT syndrome 3, 609622
KCNK9	100%	100%	100%	100%	99.8%	Birk-Barel syndrome, 612292
KDM1A	93.5%	93.5%	100%	100%	99.6%	Cleft palate, psychomotor retardation, and distinctive facial features, 616728;{ACTH-independent macronodular adrenal hyperplasia 3}, 620990
KDM6A	100%	100%	99%	91.6%	72.4%	Kabuki syndrome 2, 300867
KIAA0586	95.6%	95.6%	100%	100%	99.7%	Short-rib thoracic dysplasia 14 with polydactyly, 616546;Joubert syndrome 23, 616490

KIF7	100%	100%	100%	100%	99.2%	Joubert syndrome 12, 200990;Acrocallosal syndrome, 200990;?Hydrolethalus syndrome 2, 614120;?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	95.9%	95.9%	100%	99.9%	99.5%	Goldberg-Shprintzen megacolon syndrome, 609460
KMT2D	100%	100%	100%	100%	99.2%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920
LRRC32	100%	100%	100%	100%	99.3%	Cleft palate, proliferative retinopathy, and developmental delay, 619074
MAP3K7	97.9%	97.9%	100%	100%	99.8%	Frontometaphyseal dysplasia 2, 617137;Cardiospondylocarpofacial syndrome, 157800
MAPRE2	100%	100%	100%	100%	99.8%	Symmetric circumferential skin creases, congenital, 2, 616734
MASP1	100%	100%	100%	100%	99.6%	3MC syndrome 1, 257920
MBTPS2	94.6%	94.6%	99.3%	91.4%	73.6%	Keratosis follicularis spinulosa decalvans, X-linked, 308800;Osteogenesis imperfecta, type XIX, 301014;IFAP syndrome with or without BRESHECK syndrome, 308205;?Olmsted syndrome, X-linked, 300918

MED12	100%	99.9%	98.1%	87.5%	68.9%	Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450
MED13L	100%	100%	100%	100%	99.6%	Impaired intellectual development and distinctive facial features with or without cardiac defects, 616789
MED25	100%	100%	100%	100%	99.4%	Basel-Vanagaite-Smirin-Yosef syndrome, 616449
MEIS2	89.6%	88.5%	100%	100%	99.6%	Cleft palate, cardiac defects, and impaired intellectual development, 600987
MID1	100%	100%	98.8%	88.7%	69.8%	Opitz GBBB syndrome, 300000
MKS1	99.1%	99%	100%	100%	99.7%	Bardet-Biedl syndrome 13, 615990;Meckel syndrome 1, 249000;Joubert syndrome 28, 617121
MSX1	100%	100%	100%	99.8%	98.4%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600;Ectodermal dysplasia 3, Witkop type, 189500;Orofacial cleft 5, 608874
MYMK	100%	100%	100%	100%	99.4%	Carey-Fineman-Ziter syndrome, 254940
MYMX	100%	100%	100%	100%	100%	Carey-Fineman-Ziter syndrome 2, 619941
NEB	99.9%	99.8%	99.6%	99.2%	98.3%	Nemaline myopathy 2, autosomal recessive, 256030;Arthrogryposis multiplex congenita 6, 619334

NECTIN1	93.4%	93.4%	100%	99.9%	99.1%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060;Orofacial cleft 7, 225060
NEDD4L	100%	100%	100%	100%	99.6%	Periventricular nodular heterotopia 7, 617201
NEK1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NIPBL	100%	100%	100%	100%	99.8%	Cornelia de Lange syndrome 1, 122470
NOTCH1	99.1%	99%	100%	99.9%	99.4%	Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730
OFD1	100%	100%	99.1%	91%	73.2%	Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804
ORC1	100%	100%	100%	100%	99.8%	Meier-Gorlin syndrome 1, 224690
PAX3	100%	100%	100%	100%	99.6%	Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220
PGAP3	100%	100%	100%	100%	99.4%	Hyperphosphatasia with impaired intellectual development syndrome 4, 615716

PGM1	87.5%	87.5%	100%	100%	99.4%	Congenital disorder of glycosylation, type It, 614921
PHF8	100%	100%	98.9%	90.4%	71.6%	Intellectual developmental disorder, X-linked syndromic, Siderius type, 300263
PHGDH	100%	100%	100%	100%	99.6%	Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815
PIEZO2	100%	100%	100%	100%	99.7%	Arthrogryposis, distal, type 5, 108145;Arthrogryposis, distal, with impaired proprioception and touch, 617146;Arthrogryposis, distal, type 3, 114300;?Marden-Walker syndrome, 248700
PIGN	95.9%	94.4%	100%	100%	99.7%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGO	100%	100%	100%	100%	99.8%	Hyperphosphatasia with impaired intellectual development syndrome 2, 614749
PIGV	100%	100%	100%	100%	99.7%	Hyperphosphatasia with impaired intellectual development syndrome 1, 239300
PLCB4	99%	99%	100%	100%	99.8%	Auriculocondylar syndrome 2B, 620458;Auriculocondylar syndrome 2A, 614669
POLR1A	100%	100%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 27, 620675;Acrofacial dysostosis, Cincinnati type, 616462

POLR1C	83.6%	83.5%	100%	100%	99.2%	Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390
POLR1D	100%	100%	100%	100%	99.8%	Treacher Collins syndrome 2, 613717
POMT1	100%	99.7%	100%	100%	99.3%	Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670;Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308;Muscular dystrophy-dystroglycanopathy (congenital with impaired intellectual development), type B, 1, 613155
PORCN	100%	100%	98.5%	89.4%	70.4%	Focal dermal hypoplasia, 305600
PQBP1	100%	100%	98.6%	86.7%	67.3%	Renpenning syndrome, 309500
PROKR2	100%	100%	100%	100%	99.3%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PRRX1	100%	100%	100%	100%	99.5%	Agnathia-otocephaly complex, 202650
PTCH1	100%	100%	100%	100%	99.2%	Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828
PTCH2	100%	100%	100%	99.9%	99.4%	Medulloblastoma, somatic, 155255;Basal cell carcinoma, somatic, 605462
RBM10	95.6%	95.6%	98.7%	88.7%	70.4%	TARP syndrome, 311900

RIPK4	100%	100%	100%	100%	99.7%	CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
ROR2	100%	100%	100%	100%	99.6%	Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive 1, 268310
RPGRIP1L	100%	100%	100%	100%	99.7%	Joubert syndrome 7, 611560;Meckel syndrome 5, 611561;?COACH syndrome 3, 619113
RPL11	96.3%	94.4%	100%	100%	99.9%	Diamond-Blackfan anemia 7, 612562
RPL26	100%	100%	100%	100%	99.9%	Diamond-Blackfan anemia 11, 614900
RPL5	100%	100%	100%	100%	99.9%	Diamond-Blackfan anemia 6, 612561
RPS19	100%	100%	100%	100%	99.2%	Diamond-Blackfan anemia 1, 105650
RPS26	100%	99.9%	100%	100%	99.7%	Diamond-Blackfan anemia 10, 613309
RPS28	100%	100%	100%	100%	99.9%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RUNX2	100%	100%	100%	99.9%	98.8%	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
SALL4	100%	100%	100%	100%	99.6%	?IVIC syndrome, 147750;Duane-radial ray syndrome, 607323

SATB2	100%	100%	100%	99.9%	99.5%	Glass syndrome, 612313
SCARF2	100%	100%	100%	99.8%	97.6%	Van den Ende-Gupta syndrome, 600920
SEC23A	100%	100%	100%	100%	99.7%	Craniolenticulosutural dysplasia, 607812
SEMA3E	100%	100%	100%	100%	99.9%	
SEPTIN9	100%	100%	100%	99.9%	98.9%	Amyotrophy, hereditary neuralgic, 162100
SF3B2	100%	100%	100%	100%	99.4%	Craniofacial microsomia, 164210
SF3B4	100%	100%	100%	100%	99.1%	Acrofacial dysostosis 1, Nager type, 154400
SHH	100%	100%	100%	99.8%	98.4%	Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945;Microphthalmia/coloboma 5, 611638
SIX1	100%	100%	100%	100%	98.8%	Deafness, autosomal dominant 23, 605192;Branchiootic syndrome 3, 608389
SIX3	100%	100%	100%	99.4%	97.2%	Schizencephaly, 269160;Holoprosencephaly 2, 157170
SIX5	100%	100%	100%	100%	99.4%	Branchiootorenal syndrome 2, 610896
SKI	100%	100%	100%	99.8%	98.8%	Shprintzen-Goldberg syndrome, 182212
SLC10A7	92.8%	92.8%	100%	100%	100%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363

SLC26A2	100%	100%	100%	100%	99.8%	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
SMAD3	100%	100%	100%	100%	99.4%	Loeys-Dietz syndrome 3, 613795
SMAD4	95.4%	95.4%	100%	100%	99.8%	Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMARCA4	100%	100%	100%	99.9%	99.3%	Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792
SMC1A	100%	100%	99.1%	88.6%	70.4%	Cornelia de Lange syndrome 2, 300590;Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	100%	100%	100%	100%	99.9%	Cornelia de Lange syndrome 3, 610759
SMCHD1	100%	100%	100%	100%	99.8%	Facioscapulohumeral muscular dystrophy 2, digenic, 158901;Bosma arhinia microphthalmia syndrome, 603457

SMS	94.4%	94.2%	99.1%	91.9%	73.3%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNRPB	90.5%	82.4%	100%	100%	99.7%	Cerebrocostomandibular syndrome, 117650
SON	100%	100%	100%	100%	99.7%	ZTTK syndrome, 617140
SOX11	100%	100%	100%	99.7%	96%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866
SOX9	100%	99.5%	100%	99.9%	97.9%	Campomelic dysplasia with autosomal sex reversal, 114290;46XY sex reversal 10, 616425;Acampomelic campomelic dysplasia, 114290;Campomelic dysplasia, 114290;46XX sex reversal 2, 278850
SPECC1L	94.6%	94.6%	100%	100%	99.7%	Teebi hypertelorism syndrome 1, 145420;?Facial clefting, oblique, 1, 600251
STAC3	100%	100%	100%	100%	99.2%	Congenital myopathy 13, 255995
STAG2	100%	100%	99.3%	91.5%	74%	Holoprosencephaly 13, X-linked, 301043;Mullegama-Klein-Martinez syndrome, 301022
STAMBP	96.3%	96.3%	100%	100%	99.8%	Microcephaly-capillary malformation syndrome, 614261
TAPT1	100%	100%	100%	100%	99.7%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897

TBC1D32	100%	100%	100%	100%	99.8%	Orofaciodigital syndrome IX, 258865;Alsahan-Harris syndrome, 621307;Retinitis pigmentosa 100, 621280
TBX1	98.6%	95.7%	100%	99.4%	96.1%	Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430
TBX15	100%	100%	100%	100%	99.8%	Cousin syndrome, 260660
TBX2	99.5%	97.7%	100%	100%	99.1%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX22	100%	100%	99%	91.2%	72.3%	Cleft palate with ankyloglossia, 303400;?Abruzzo-Erickson syndrome, 302905
TBX4	100%	100%	100%	100%	99%	Ischiocoxopodopatellar syndrome with or without pulmonary arterial hypertension, 147891;Amelia, posterior, with pelvic and pulmonary hypoplasia syndrome, 601360
TCOF1	97.9%	97.9%	100%	100%	99.6%	Treacher Collins syndrome 1, 154500
TCTN3	100%	100%	100%	100%	99.5%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TFAP2A	100%	100%	100%	100%	98.8%	Branchiooculofacial syndrome, 113620
TGDS	100%	100%	100%	100%	99.9%	Catel-Manzke syndrome, 616145

TGFB2	100%	100%	100%	99.8%	99.4%	Loeys-Dietz syndrome 4, 614816;Camurati-Engelmann disease 2, 606631
TGFB3	88.3%	88.3%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 1, 107970;Loeys-Dietz syndrome 5, 615582
TGFBR1	100%	100%	100%	100%	99.6%	{Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192
TGFBR2	100%	100%	100%	100%	99.6%	Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239
TGIF1	100%	100%	100%	100%	99.9%	Holoprosencephaly 4, 142946
TMCO1	72.3%	72.3%	100%	100%	99.6%	Craniofacial dysmorphism, skeletal anomalies, and impaired intellectual development 1, 213980
TMEM216	100%	100%	100%	100%	99.8%	Joubert syndrome 2, 608091;Retinitis pigmentosa 98, 620996;Meckel syndrome 2, 603194

TP63	100%	100%	100%	100%	99.6%	Premature ovarian failure 21, 620311;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
TRIM37	98.3%	97.6%	100%	100%	99.7%	Mulibrey nanism, 253250
TRRAP	100%	100%	100%	100%	99.5%	?Deafness, autosomal dominant 75, 618778;Developmental delay with or without dysmorphic facies and autism, 618454
TUBB	99.8%	98.9%	100%	100%	99.8%	Symmetric circumferential skin creases, congenital, 1, 156610;Cortical dysplasia, complex, with other brain malformations 6, 615771
TWIST1	100%	100%	100%	100%	98.5%	Craniosynostosis 1, 123100;Robinow-Sorauf syndrome, 180750;Sweeney-Cox syndrome, 617746;Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TXNL4A	100%	100%	100%	99.7%	99.5%	Burn-McKeown syndrome, 608572

USP9X	100%	100%	99.4%	91.8%	75%	Intellectual developmental disorder, X-linked 99, 300919;Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968
WASHC5	100%	100%	100%	100%	99.8%	Ritscher-Schinzel syndrome 1, 220210;Spastic paraplegia 8, autosomal dominant, 603563
WDR35	100%	100%	100%	100%	99.7%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610
WNT4	100%	100%	100%	100%	99.1%	Mullerian aplasia and hyperandrogenism, 158330;SERKAL syndrome, 611812
WNT5A	100%	100%	100%	99.9%	98.6%	Robinow syndrome, autosomal dominant 1, 180700
XYLT1	100%	100%	100%	99.9%	99.3%	Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800
ZEB2	100%	100%	100%	99.9%	99.4%	Mowat-Wilson syndrome, 235730
ZFHX4	100%	100%	100%	100%	99.6%	
ZIC2	100%	100%	99.9%	99.6%	97%	Holoprosencephaly 5, 609637

ZIC3	100%	100%	98.4%	87.4%	68.6%	Congenital heart defects, nonsyndromic, multiple types, 1, X-linked, 306955;Heterotaxy, visceral, 1, X-linked, 306955;VACTERL association, X-linked, 314390
ZMPSTE24	100%	100%	100%	100%	100%	Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210
ZSWIM6	97.9%	96.3%	99.2%	98.1%	96.1%	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865;Acromelic frontonasal dysostosis, 603671

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.

TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
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 : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors