SCHISIS GENE PANEL DG 3.4.0 (196 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
ACTB	100,0%	100,0%	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTG1	100,0%	100,0%	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ALX1	100,0%	100,0%	Frontonasal dysplasia 3, 613456
ALX3	100,0%	100,0%	Frontonasal dysplasia 1, 136760
AMER1	100,0%	100,0%	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	100,0%	100,0%	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANKRD11	100,0%	100,0%	KBG syndrome, 148050
ARHGAP29	100,0%	100,0%	No OMIM Disease ID
ARHGAP31	100,0%	100,0%	Adams-Oliver syndrome 1, 100300
ASXL1	99,9%	99,9%	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039
B3GALT6	99,8%	98,8%	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Al-Gazali syndrome, 609465
B3GLCT	100,0%	100,0%	Peters-plus syndrome, 261540
B4GALT7	100,0%	100,0%	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B9D2	100,0%	100,0%	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BCOR	100,0%	100,0%	Microphthalmia, syndromic 2, 300166
BMP2	100,0%	100,0%	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 Brachydactyly, type A2, 112600
BMPER	100,0%	100,0%	Diaphanospondylodysostosis, 608022
IMPAD1	100,0%	100,0%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
C2CD3	95,9%	95,9%	Orofaciodigital syndrome XIV, 615948
CC2D2A	97,1%	97,1%	COACH syndrome 2, 619111 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285

CDC45	100,0%	100,0%	Meier-Gorlin syndrome 7, 617063
CDH1	97,8%	97,8%	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	100,0%	100,0%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CHD7	100,0%	100,0%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHRNG	100,0%	100,0%	Multiple pterygium syndrome, lethal type, 253290 Escobar syndrome, 265000
CHST14	100,0%	100,0%	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CILK1	100,0%	100,0%	Endocrine-cerebroosteodysplasia, 612651
COL11A1	100,0%	100,0%	Fibrochondrogenesis 1, 228520 Stickler syndrome, type II, 604841 Marshall syndrome, 154780 Deafness, autosomal dominant 37, 618533
COL11A2	100,0%	100,0%	Deafness, autosomal dominant 13, 601868 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150 Fibrochondrogenesis 2, 614524 Deafness, autosomal recessive 53, 609706 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840
COL2A1	100,0%	100,0%	?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 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	100,0%	100,0%	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135
COLEC10	100,0%	100,0%	3MC syndrome 3, 248340
COLEC11	100,0%	100,0%	3MC syndrome 2, 265050
CPLANE1	100,0%	100,0%	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CTCF	100,0%	100,0%	Intellectual developmental disorder, autosomal dominant 21, 615502
CTNND1	100,0%	100,0%	Blepharocheilodontic syndrome 2, 617681
DDX3X	99,2%	97,6%	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958
DDX59	100,0%	100,0%	Orofaciodigital syndrome V, 174300
DHCR7	100,0%	100,0%	Smith-Lemli-Opitz syndrome, 270400
DHODH	100,0%	100,0%	Miller syndrome, 263750
DLL4	100,0%	100,0%	Adams-Oliver syndrome 6, 616589
DOCK6	100,0%	100,0%	Adams-Oliver syndrome 2, 614219
DVL1	100,0%	100,0%	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100,0%	100,0%	Robinow syndrome, autosomal dominant 3, 616894
DYNC2H1	100,0%	100,0%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	100,0%	100,0%	Short-rib thoracic dysplasia 15 with polydactyly, 617088
EBP	100,0%	100,0%	MEND syndrome, 300960 Chondrodysplasia punctata, X-linked dominant, 302960
EDN1	100,0%	100,0%	Question mark ears, isolated, 612798 Auriculocondylar syndrome 3, 615706
EDNRA	100,0%	100,0%	Mandibulofacial dysostosis with alopecia, 616367
EFNB1	100,0%	100,0%	Craniofrontonasal dysplasia, 304110
EFTUD2	100,0%	100,0%	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF2S3	100,0%	100,0%	MEHMO syndrome, 300148
EIF4A3	100,0%	100,0%	Robin sequence with cleft mandible and limb anomalies, 268305
EOGT	94,3%	90,6%	Adams-Oliver syndrome 4, 615297
EPG5	100,0%	100,0%	Vici syndrome, 242840
ESCO2	100,0%	100,0%	Juberg-Hayward syndrome, 216100 Roberts-SC phocomelia syndrome, 268300
EYA1	100,0%	100,0%	Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650 Anterior segment anomalies with or without cataract, 602588 ?Otofaciocervical syndrome, 166780
FAM20C	100,0%	100,0%	Raine syndrome, 259775

FGD1	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF8	100,0%	100,0%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100,0%	100,0%	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	100,0%	100,0%	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,
FLNA	100,0%	100,0%	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	100,0%	100,0%	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721

			Spondylocarpotarsal synostosis syndrome, 272460 Boomerang dysplasia, 112310
FOXC2	100,0%	100,0%	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	100,0%	100,0%	Bamforth-Lazarus syndrome, 241850
FRAS1	100,0%	100,0%	Fraser syndrome 1, 219000
FTO	94,2%	94,2%	Growth retardation, developmental delay, facial dysmorphism, 612938
GDF6	100,0%	100,0%	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	100,0%	100,0%	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
GLI2	100,0%	100,0%	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	100,0%	100,0%	Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700
GNAI3	100,0%	100,0%	Auriculocondylar syndrome 1, 602483
GNB1	100,0%	100,0%	Myelodysplastic syndrome, somatic, 614286 Leukemia, acute lymphoblastic, somatic, 613065 Intellectual developmental disorder, autosomal dominant 42, 616973
GPC3	100,0%	99,9%	Wilms tumor, somatic, 194070 Simpson-Golabi-Behmel syndrome, type 1, 312870
GRHL3	100,0%	100,0%	van der Woude syndrome 2, 606713
HDAC8	96,6%	96,0%	Cornelia de Lange syndrome 5, 300882
HYLS1	100,0%	100,0%	Hydrolethalus syndrome, 236680
IFT140	100,0%	100,0%	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781

IFT172	100,0%	100,0%	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT57	100,0%	100,0%	?Orofaciodigital syndrome XVIII, 617927
IFT80	100,0%	100,0%	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
INTU	100,0%	100,0%	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
IRF6	100,0%	100,0%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome 1, 119300
KANSL1	100,0%	100,0%	Koolen-De Vries syndrome, 610443
KAT6A	100,0%	100,0%	Arboleda-Tham syndrome, 616268
KCNJ2	100,0%	100,0%	Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 Short QT syndrome 3, 609622
KCNK9	97,3%	97,3%	Birk-Barel syndrome, 612292
KDM6A	100,0%	100,0%	Kabuki syndrome 2, 300867
KIAA0586	95,8%	95,8%	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490
KIF7	100,0%	100,0%	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	96,1%	96,1%	Goldberg-Shprintzen megacolon syndrome, 609460
KMT2D	100,0%	100,0%	Kabuki syndrome 1, 147920
MAP3K7	100,0%	100,0%	Frontometaphyseal dysplasia 2, 617137 Cardiospondylocarpofacial syndrome, 157800
MAPRE2	100,0%	100,0%	Symmetric circumferential skin creases, congenital, 2, 616734
MASP1	100,0%	100,0%	3MC syndrome 1, 257920
MBTPS2	100,0%	100,0%	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
MED25	100,0%	100,0%	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEIS2	100,0%	100,0%	Cleft palate, cardiac defects, and mental retardation, 600987
MID1	100,0%	100,0%	Opitz GBBB syndrome, 300000

MKS1	100,0%	100,0%	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MSX1	100,0%	100,0%	Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874
MYMK	100,0%	100,0%	Carey-Fineman-Ziter syndrome, 254940
NECTIN1	100,0%	100,0%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NEDD4L	100,0%	100,0%	Periventricular nodular heterotopia 7, 617201
NEK1	100,0%	100,0%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NIPBL	100,0%	100,0%	Cornelia de Lange syndrome 1, 122470
NOTCH1	100,0%	100,0%	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
OFD1	100,0%	100,0%	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
ORC1	100,0%	100,0%	Meier-Gorlin syndrome 1, 224690
PAX3	100,0%	100,0%	Craniofacial-deafness-hand syndrome, 122880 Waardenburg syndrome, type 3, 148820 Waardenburg syndrome, type 1, 193500 Rhabdomyosarcoma 2, alveolar, 268220
PGM1	94,2%	94,2%	Congenital disorder of glycosylation, type It, 614921
PHF8	100,0%	100,0%	Intellectual developmental disorder, X-linked, syndromic, Siderius type, 300263
PHGDH	100,0%	100,0%	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIEZO2	100,0%	100,0%	Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 3, 114300 ?Marden-Walker syndrome, 248700
PIGN	98,8%	98,8%	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGV	100,0%	100,0%	Hyperphosphatasia with mental retardation syndrome 1, 239300
PLCB4	100,0%	100,0%	Auriculocondylar syndrome 2, 614669
POLR1A	100,0%	100,0%	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	83,0%	82,8%	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	100,0%	100,0%	Treacher Collins syndrome 2, 613717

POMT1	100,0%	100,0%	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 mental retardation), type B, 1, 613155
PORCN	100,0%	100,0%	Focal dermal hypoplasia, 305600
PQBP1	100,0%	100,0%	Renpenning syndrome, 309500
PROKR2	100,0%	100,0%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PRRX1	100,0%	100,0%	Agnathia-otocephaly complex, 202650
PTCH1	100,0%	100,0%	Basal cell carcinoma, somatic, 605462 Holoprosencephaly 7, 610828 Basal cell nevus syndrome, 109400
PTCH2	100,0%	100,0%	Medulloblastoma, somatic, 155255 Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462
RBM10	100,0%	100,0%	TARP syndrome, 311900
RIPK4	100,0%	100,0%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
ROR2	97,0%	97,0%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RPGRIP1L	100,0%	99,8%	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RPL11	100,0%	100,0%	Diamond-Blackfan anemia 7, 612562
RPL26	100,0%	100,0%	?Diamond-Blackfan anemia 11, 614900
RPL5	100,0%	100,0%	Diamond-Blackfan anemia 6, 612561
RPS19	100,0%	100,0%	Diamond-Blackfan anemia 1, 105650
RPS26	100,0%	100,0%	Diamond-Blackfan anemia 10, 613309
RPS28	100,0%	100,0%	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RUNX2	100,0%	100,0%	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	100,0%	100,0%	?IVIC syndrome, 147750 Duane-radial ray syndrome, 607323
SATB2	100,0%	100,0%	Glass syndrome, 612313
SCARF2	100,0%	100,0%	Van den Ende-Gupta syndrome, 600920
SEC23A	100,0%	100,0%	Craniolenticulosutural dysplasia, 607812
SEMA3E	100,0%	100,0%	?CHARGE syndrome, 214800

SEPTIN9	100,0%	100,0%	Amyotrophy, hereditary neuralgic, 162100
SF3B4	100,0%	100,0%	Acrofacial dysostosis 1, Nager type, 154400
SHH	100,0%	100,0%	Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SIX1	100,0%	100,0%	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389
SIX3	100,0%	100,0%	Schizencephaly, 269160 Holoprosencephaly 2, 157170
SIX5	100,0%	100,0%	Branchiootorenal syndrome 2, 610896
SKI	100,0%	100,0%	Shprintzen-Goldberg syndrome, 182212
SLC10A7	100,0%	100,0%	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363
SLC26A2	100,0%	100,0%	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	100,0%	100,0%	Loeys-Dietz syndrome 3, 613795
SMAD4	100,0%	100,0%	Pancreatic cancer, somatic, 260350 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMC1A	100,0%	100,0%	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	100,0%	100,0%	Cornelia de Lange syndrome 3, 610759
SMCHD1	100,0%	100,0%	Bosma arhinia microphthalmia syndrome, 603457 Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMS	100,0%	100,0%	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNRPB	100,0%	100,0%	Cerebrocostomandibular syndrome, 117650
SON	100,0%	100,0%	ZTTK syndrome, 617140
SOX9	100,0%	100,0%	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290
SPECC1L	97,8%	96,2%	Teebi hypertelorism syndrome 1, 145420 ?Facial clefting, oblique, 1, 600251
STAC3	100,0%	100,0%	Myopathy, congenital, Baily-Bloch, 255995

STAMBP	100,0%	100,0%	Microcephaly-capillary malformation syndrome, 614261
TAPT1	100,0%	100,0%	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TBX1	98,1%	95,9%	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX15	100,0%	100,0%	Cousin syndrome, 260660
TBX2	100,0%	100,0%	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX22	100,0%	100,0%	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TCOF1	100,0%	100,0%	Treacher Collins syndrome 1, 154500
TCTN3	100,0%	100,0%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TFAP2A	100,0%	100,0%	Branchiooculofacial syndrome, 113620
TGDS	100,0%	100,0%	Catel-Manzke syndrome, 616145
TGFB3	100,0%	100,0%	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBR1	100,0%	99,9%	Loeys-Dietz syndrome 1, 609192
TGFBR2	100,0%	100,0%	Loeys-Dietz syndrome 2, 610168 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239
TGIF1	100,0%	100,0%	Holoprosencephaly 4, 142946
TMCO1	88,0%	88,0%	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM216	100,0%	100,0%	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TP63	100,0%	100,0%	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	98,7%	98,7%	Mulibrey nanism, 253250
TUBB	100,0%	99,8%	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TWIST1	100,0%	100,0%	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750

			Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TXNL4A	100,0%	100,0%	Burn-McKeown syndrome, 608572
USP9X	100,0%	100,0%	Intellectual developmental disorder, X-linked 99, 300919 Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968
WASHC5	100,0%	100,0%	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WDR35	100,0%	100,0%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WNT4	100,0%	99,8%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT5A	100,0%	100,0%	Robinow syndrome, autosomal dominant 1, 180700
XYLT1	100,0%	99,7%	Desbuquois dysplasia 2, 615777
ZEB2	97,4%	97,4%	Mowat-Wilson syndrome, 235730
ZIC2	100,0%	100,0%	Holoprosencephaly 5, 609637
ZIC3	100,0%	100,0%	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390
ZMPSTE24	100,0%	100,0%	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy 1, 275210
ZSWIM6	97,6%	96,3%	Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865 Acromelic frontonasal dysostosis, 603671

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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors