SCHISIS GENE PANEL DG 3.2.0 (196 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
ACTB	99,9	97,2	100	100	Baraitser-Winter syndrome 1, 243310 ?Dystonia, juvenile-onset, 607371
ACTG1	100	100	100	100	Deafness, autosomal dominant 20/26, 604717 Baraitser-Winter syndrome 2, 614583
ALX1	99,6	95,2	100	100	Frontonasal dysplasia 3, 613456
ALX3	80,2	72,8	100	100	Frontonasal dysplasia 1, 136760
AMER1	99,6	96,6	100	100	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	99,9	98,4	100	100	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANKRD11	97	94	100	100	KBG syndrome, 148050
ARHGAP29	98,9	97,6	100	99,9	No OMIM disease ID
ARHGAP31	99,7	98,2	100	100	Adams-Oliver syndrome 1, 100300
ASXL1	99,8	98,9	100	100	Myelodysplastic syndrome, somatic, 614286 Bohring-Opitz syndrome, 605039
B3GALT6	77	73	91,7	81	Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640 Al-Gazali syndrome, 609465
B3GLCT	99,7	98,2	100	99,4	Peters-plus syndrome, 261540
B4GALT7	99,7	96,8	100	99,4	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B9D2	100	100	100	100	?Meckel syndrome 10, 614175 Joubert syndrome 34, 614175
BCOR	99,2	95,8	100	100	Microphthalmia, syndromic 2, 300166
BMP2	100	100	100	100	Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877 Brachydactyly, type A2, 112600
BMPER	100	99,6	100	100	Diaphanospondylodysostosis, 608022
IMPAD1	100	99,9	100	100	Chondrodysplasia with joint dislocations, GPAPP type, 614078
C2CD3	95,8	95,4	95,9	95,9	Orofaciodigital syndrome XIV, 615948

CC2D2A	98,3	96,6	97,1	97	COACH syndrome 2, 619111 Meckel syndrome 6, 612284 Joubert syndrome 9, 612285
CDC45	99,8	98,5	100	100	Meier-Gorlin syndrome 7, 617063
CDH1	99,2	98,6	99,2	99,1	Ovarian cancer, somatic, 167000 Blepharocheilodontic syndrome 1, 119580 Endometrial carcinoma, somatic, 608089 Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215
CDKN1C	89,9	81,6	98,9	95,8	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CHD7	100	99,2	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHRNG	100	100	100	100	Multiple pterygium syndrome, lethal type, 253290 Escobar syndrome, 265000
CHST14	99,9	98,8	100	100	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CILK1	99,6	98	100	99,8	Endocrine-cerebroosteodysplasia, 612651
COL11A1	96	92,7	100	99,9	Fibrochondrogenesis 1, 228520 Stickler syndrome, type II, 604841 Marshall syndrome, 154780 Deafness, autosomal dominant 37, 618533
COL11A2	100	99,6	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	99,8	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	99,9	98,6	100	100	Stickler syndrome, type IV, 614134 ?Epiphyseal dysplasia, multiple, 6, 614135
COLEC10	100	99,9	100	100	3MC syndrome 3, 248340
COLEC11	100	100	100	100	3MC syndrome 2, 265050
CPLANE1	99,4	98,2	100	100	Orofaciodigital syndrome VI, 277170 Joubert syndrome 17, 614615
CTCF	99,7	98,4	100	100	Mental retardation, autosomal dominant 21, 615502
CTNND1	100	99,8	100	100	Blepharocheilodontic syndrome 2, 617681
DDX3X	81,1	78,6	98,3	96,1	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958
DDX59	100	99,8	100	100	Orofaciodigital syndrome V, 174300
DHCR7	100	100	100	100	Smith-Lemli-Opitz syndrome, 270400
DHODH	100	99,9	100	100	Miller syndrome, 263750
DLL4	100	99,4	100	100	Adams-Oliver syndrome 6, 616589
DOCK6	99,4	98,7	100	100	Adams-Oliver syndrome 2, 614219
DVL1	97,2	95,1	100	100	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100	100	100	100	Robinow syndrome, autosomal dominant 3, 616894
DYNC2H1	98,6	95,2	100	99,8	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	99,6	98,4	100	100	Short-rib thoracic dysplasia 15 with polydactyly, 617088
EBP	99,5	94,3	100	100	MEND syndrome, 300960 Chondrodysplasia punctata, X-linked dominant, 302960
EDN1	100	99,4	100	100	Question mark ears, isolated, 612798 Auriculocondylar syndrome 3, 615706
EDNRA	99,8	99,8	100	99,9	Mandibulofacial dysostosis with alopecia, 616367
EFNB1	100	100	100	100	Craniofrontonasal dysplasia, 304110
EFTUD2	100	99,2	100	100	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF2S3	95	86,8	100	100	MEHMO syndrome, 300148
EIF4A3	100	99,2	100	100	Robin sequence with cleft mandible and limb anomalies, 268305
EOGT	79,3	77,8	91,8	88,3	Adams-Oliver syndrome 4, 615297

EPG5	99,2	97,8	100	100	Vici syndrome, 242840
ESCO2	98,5	94,6	100	99,7	Juberg-Hayward syndrome, 216100 Roberts-SC phocomelia syndrome, 268300
EYA1	99,9	99,5	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	100	100	100	99,7	Raine syndrome, 259775
FGD1	97,2	91,2	100	100	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF8	97,1	87,2	100	99,9	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100	99,3	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	97,6	97	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,
FLNA	100	99,9	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	99,4	98,7	100	100	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Atelosteogenesis, type III, 108721 Spondylocarpotarsal synostosis syndrome, 272460 Boomerang dysplasia, 112310
FOXC2	100	98,1	100	99,6	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	97,9	82,2	100	98,9	Bamforth-Lazarus syndrome, 241850
FRAS1	100	99,2	100	100	Fraser syndrome 1, 219000
FTO	83,8	83,7	94,2	94,2	Growth retardation, developmental delay, facial dysmorphism, 612938
GDF6	100	100	100	99,6	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	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	99,8	98,6	100	99,9	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	98,5	97,7	100	100	Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, postaxial, types A1 and B, 174200 Pallister-Hall syndrome, 146510 Polydactyly, preaxial, type IV, 174700
GNAI3	98,4	93,2	100	100	Auriculocondylar syndrome 1, 602483

GNB1	100	100	100	100	Myelodysplastic syndrome, somatic, 614286 Leukemia, acute lymphoblastic, somatic, 613065 Mental retardation, autosomal dominant 42, 616973
GPC3	98,8	92,9	100	99,9	Wilms tumor, somatic, 194070 Simpson-Golabi-Behmel syndrome, type 1, 312870
GRHL3	100	99,9	100	100	Van der Woude syndrome 2, 606713
HDAC8	85,7	83,7	96,4	95,2	Cornelia de Lange syndrome 5, 300882
HYLS1	100	100	100	100	Hydrolethalus syndrome, 236680
IFT140	99,9	99,2	100	100	Short-rib thoracic dysplasia 9 with or without polydactyly, 266920 Retinitis pigmentosa 80, 617781
IFT172	99,6	98,6	100	100	Retinitis pigmentosa 71, 616394 Bardet-Biedl syndrome 20, 619471 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT57	99,9	99	100	99,7	?Orofaciodigital syndrome XVIII, 617927
IFT80	97,2	85,7	100	99,9	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
INTU	99,9	98,6	100	100	?Orofaciodigital syndrome XVII, 617926 ?Short-rib thoracic dysplasia 20 with polydactyly, 617925
IRF6	99,4	93	100	100	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300
KANSL1	99,8	98,2	100	100	Koolen-De Vries syndrome, 610443
KAT6A	100	99,2	100	100	Arboleda-Tham syndrome, 616268
KCNJ2	100	100	100	100	Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390 Short QT syndrome 3, 609622
KCNK9	97,3	97,3	97,3	97,3	Birk-Barel syndrome, 612292
KDM6A	94,2	85,9	100	99,9	Kabuki syndrome 2, 300867
KIAA0586	97,1	92	95,8	95,7	Short-rib thoracic dysplasia 14 with polydactyly, 616546 Joubert syndrome 23, 616490
KIF7	93,6	91,9	99,7	98,6	Joubert syndrome 12, 200990 Acrocallosal syndrome, 200990 ?Hydrolethalus syndrome 2, 614120 ?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	96,1	96	96,1	96,1	Goldberg-Shprintzen megacolon syndrome, 609460
KMT2D	99,9	99	100	100	Kabuki syndrome 1, 147920

MAP3K7	99,8	99,6	100	100	Frontometaphyseal dysplasia 2, 617137 Cardiospondylocarpofacial syndrome, 157800
MAPRE2	100	98,5	100	100	Symmetric circumferential skin creases, congenital, 2, 616734
MASP1	100	99,6	100	100	3MC syndrome 1, 257920
MBTPS2	99,9	98,5	100	100	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	99,9	100	99,9	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEIS2	100	99,7	100	100	Cleft palate, cardiac defects, and mental retardation, 600987
MID1	99,6	97,7	100	100	Opitz GBBB syndrome, type I, 300000
MKS1	99,4	96,3	100	100	Bardet-Biedl syndrome 13, 615990 Meckel syndrome 1, 249000 Joubert syndrome 28, 617121
MSX1	97,7	92,6	100	100	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	Carey-Fineman-Ziter syndrome, 254940
NECTIN1	100	99,7	100	100	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NEDD4L	71,9	71,7	100	99,9	Periventricular nodular heterotopia 7, 617201
NEK1	99,5	98,2	100	99,9	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NIPBL	98,4	96,3	100	99,9	Cornelia de Lange syndrome 1, 122470
NOTCH1	99,3	97,9	100	100	Adams-Oliver syndrome 5, 616028 Aortic valve disease 1, 109730
OFD1	87,1	71,3	100	99,8	Simpson-Golabi-Behmel syndrome, type 2, 300209 ?Retinitis pigmentosa 23, 300424 Orofaciodigital syndrome I, 311200 Joubert syndrome 10, 300804
ORC1	99,9	97,9	100	100	Meier-Gorlin syndrome 1, 224690
PAX3	100	99,8	100	100	Craniofacial-deafness-hand syndrome, 122880 Waardenburg syndrome, type 3, 148820 Waardenburg syndrome, type 1, 193500 Rhabdomyosarcoma 2, alveolar, 268220

PGM1	94,2	94,1	94,2	94,2	Congenital disorder of glycosylation, type It, 614921
PHF8	98,9	94,4	100	100	Intellectual developmental disorder, X-linked, syndromic, Siderius type, 300263
PHGDH	99,9	98,2	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIEZO2	99,8	99,2	100	100	Arthrogryposis, distal, type 5, 108145 Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 3, 114300 ?Marden-Walker syndrome, 248700
PIGN	93,1	89,6	98,8	98,6	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGV	100	100	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PLCB4	99,8	98,7	100	100	Auriculocondylar syndrome 2, 614669
POLR1A	99,9	98,8	100	100	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	89,6	84,8	82,8	82,8	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	91,6	91,6	100	99,8	Treacher Collins syndrome 2, 613717
POMT1	99,5	97,3	100	100	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	99,1	100	100	Focal dermal hypoplasia, 305600
PQBP1	100	99,3	100	100	Renpenning syndrome, 309500
PROKR2	100	100	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PRRX1	100	99,4	100	100	Agnathia-otocephaly complex, 202650
PTCH1	99,3	96,6	100	99,9	Basal cell carcinoma, somatic, 605462 Holoprosencephaly 7, 610828 Basal cell nevus syndrome, 109400
PTCH2	99,9	98,4	100	100	Medulloblastoma, somatic, 155255 Basal cell nevus syndrome, 109400 Basal cell carcinoma, somatic, 605462
RBM10	99,8	97,3	100	100	TARP syndrome, 311900
RIPK4	100	99,9	100	100	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
ROR2	100	99,4	97	97	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310

RPGRIP1L	96,5	95,3	100	99,4	Joubert syndrome 7, 611560 Meckel syndrome 5, 611561 ?COACH syndrome 3, 619113
RPL11	99,9	97,9	100	100	Diamond-Blackfan anemia 7, 612562
RPL26	94,2	75,5	100	100	?Diamond-Blackfan anemia 11, 614900
RPL5	81,9	59,7	100	100	Diamond-Blackfan anemia 6, 612561
RPS19	100	99,9	100	100	Diamond-Blackfan anemia 1, 105650
RPS26	93,2	81,2	100	100	Diamond-Blackfan anemia 10, 613309
RPS28	99,7	86,3	100	100	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RUNX2	72,2	72,2	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
SALL4	99,1	96,4	100	100	?IVIC syndrome, 147750 Duane-radial ray syndrome, 607323
SATB2	99,5	96,5	100	100	Glass syndrome, 612313
SCARF2	97,4	88,9	99,8	99,2	Van den Ende-Gupta syndrome, 600920
SEC23A	99,7	97	100	100	Craniolenticulosutural dysplasia, 607812
SEMA3E	99,1	98,9	100	100	?CHARGE syndrome, 214800
SEPTIN9	100	99,5	100	100	Amyotrophy, hereditary neuralgic, 162100
SF3B4	99,8	94,1	100	100	Acrofacial dysostosis 1, Nager type, 154400
SHH	100	100	100	100	Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SIX1	100	99,7	100	100	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389
SIX3	99,3	96,9	100	99,8	Schizencephaly, 269160 Holoprosencephaly 2, 157170
SIX5	96,9	90,1	100	100	Branchiootorenal syndrome 2, 610896
SKI	99,7	97,1	100	99,7	Shprintzen-Goldberg syndrome, 182212
SLC10A7	99,5	98,1	100	100	Short stature, amelogenesis imperfecta, and skeletal dysplasia with scoliosis, 618363

SLC26A2	100	100	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
SMAD3	99,9	98,4	100	100	Loeys-Dietz syndrome 3, 613795
SMAD4	99,9	99,9	100	100	Pancreatic cancer, somatic, 260350 Myhre syndrome, 139210 Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
SMC1A	99,6	97,1	100	99,9	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	94,5	89	100	99,9	Cornelia de Lange syndrome 3, 610759
SMCHD1	99,3	96,4	100	99,9	Bosma arhinia microphthalmia syndrome, 603457 Fascioscapulohumeral muscular dystrophy 2, digenic, 158901
SMS	87,9	72,1	100	99,5	Intellectual developmental disorder, X-linked syndromic, Snyder-Robinson type, 309583
SNRPB	100	98,6	100	100	Cerebrocostomandibular syndrome, 117650
SON	97,6	92,6	100	100	ZTTK syndrome, 617140
SOX9	100	99,9	100	100	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290
SPECC1L	96	95	97,1	96,1	Opitz GBBB syndrome, type II, 145410 Teebi hypertelorism syndrome, 145420 ?Facial clefting, oblique, 1, 600251
STAC3	100	100	100	100	Myopathy, congenital, Baily-Bloch, 255995
STAMBP	99,4	96,4	100	100	Microcephaly-capillary malformation syndrome, 614261
TAPT1	93	87,1	98,5	94,1	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TBX1	87,4	77,6	93,7	90,2	Tetralogy of Fallot, 187500 DiGeorge syndrome, 188400 Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430
TBX15	100	99,7	100	100	Cousin syndrome, 260660
TBX2	99,9	97,8	98,4	95,7	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223

TBX22	98,4	93,8	100	99,9	Cleft palate with ankyloglossia, 303400 ?Abruzzo-Erickson syndrome, 302905
TCOF1	99,7	98,7	100	100	Treacher Collins syndrome 1, 154500
TCTN3	100	100	100	100	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TFAP2A	98,1	92,1	100	100	Branchiooculofacial syndrome, 113620
TGDS	99,4	95,9	100	99,9	Catel-Manzke syndrome, 616145
TGFB3	100	100	100	100	Arrhythmogenic right ventricular dysplasia 1, 107970 Loeys-Dietz syndrome 5, 615582
TGFBR1	93,6	93,6	98,8	97,6	Loeys-Dietz syndrome 1, 609192
TGFBR2	100	99,9	100	100	Loeys-Dietz syndrome 2, 610168 Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239
TGIF1	100	100	100	100	Holoprosencephaly 4, 142946
TMCO1	87,8	87	88	87,9	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM216	98,5	92,8	100	100	Joubert syndrome 2, 608091 Meckel syndrome 2, 603194
TP63	100	100	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
TRIM37	98,3	97,1	98,7	98,6	Mulibrey nanism, 253250
TUBB	96,8	93,7	99,9	99,8	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TWIST1	100	99,4	96,7	90,6	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TXNL4A	99,3	99,1	100	100	Burn-McKeown syndrome, 608572
USP9X	98,1	91,7	100	99,8	Intellectual developmental disorder, X-linked 99, 300919 Intellectual developmental disorder, X-linked 99, syndromic, female-restricted, 300968

WASHC5	99,8	99,7	100	100	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WDR35	99,6	98,4	100	100	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WNT4	97,8	93,6	99,3	96,5	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT5A	100	100	100	100	Robinow syndrome, autosomal dominant 1, 180700
XYLT1	97,8	91,1	97,7	94,1	Desbuquois dysplasia 2, 615777
ZEB2	99,7	98,5	97,4	97,4	Mowat-Wilson syndrome, 235730
ZIC2	100	99,3	97,7	94,2	Holoprosencephaly 5, 609637
ZIC3	100	99,9	100	100	Congenital heart defects, nonsyndromic, 1, X-linked, 306955 Heterotaxy, visceral, 1, X-linked, 306955 VACTERL association, X-linked, 314390
ZMPSTE24	99,6	99,4	100	99,9	Mandibuloacral dysplasia with type B lipodystrophy, 608612 Restrictive dermopathy, lethal, 275210
ZSWIM6	95,1	91,6	94,3	91	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.
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-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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors