SCHISIS GENE PANEL DG 3.1.0 (195 genes)

Releasedate: 23-03-2021

Gene	Agilent V5 covered >10x	Agilent V5 covered >20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
ACTB	99,7	96,1	100	100	?Dystonia, juvenile-onset, 607371 Baraitser-Winter syndrome 1, 243310
ACTG1	100	100	100	100	Baraitser-Winter syndrome 2, 614583 Deafness, autosomal dominant 20/26, 604717
ALX1	99,7	97,1	100	100	Frontonasal dysplasia 3, 613456
ALX3	77,9	73,3	100	100	Frontonasal dysplasia 1, 136760
AMER1	99,9	98,5	100	100	Osteopathia striata with cranial sclerosis, 300373
AMMECR1	100	99,1	100	100	Midface hypoplasia, hearing impairment, elliptocytosis, and nephrocalcinosis, 300990
ANKRD11	96,1	93,5	100	100	KBG syndrome, 148050
ARHGAP29	99,5	98	100	100	No OMIM disease ID
ARHGAP31	99,9	98,8	100	100	Adams-Oliver syndrome 1, 100300
ASXL1	99,8	99,3	99,8	99,8	Bohring-Opitz syndrome, 605039 Myelodysplastic syndrome, somatic, 614286
B3GALT6	75,7	69,7	89,8	81,6	Al-Gazali syndrome, 609465 Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349 Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640
B3GLCT	99,6	96,3	99,9	99,2	Peters-plus syndrome, 261540
B4GALT7	99,8	97,4	99,9	98,6	Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070
B9D2	100	100	100	100	Joubert syndrome 34, 614175 ?Meckel syndrome 10, 614175
BCOR	99,6	97,4	100	99,9	Microphthalmia, syndromic 2, 300166
BMP2	100	100	100	100	Brachydactyly, type A2, 112600 {HFE hemochromatosis, modifier of}, 235200 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies 1, 617877
BMPER	100	99,8	100	100	Diaphanospondylodysostosis, 608022
C2CD3	95,8	95,6	95,9	95,9	Orofaciodigital syndrome XIV, 615948
CC2D2A	98,5	96,5	97,1	97,1	Meckel syndrome 6, 612284 Joubert syndrome 9, 612285 COACH syndrome 2, 619111

CDC45	99,8	98,5	100	100	Meier-Gorlin syndrome 7, 617063
CDH1	99,2	99,1	96,1	96	Endometrial carcinoma, somatic, 608089 {Prostate cancer, susceptibility to}, 176807 Blepharocheilodontic syndrome 1, 119580 Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate, 137215 {Breast cancer, lobular}, 114480 Ovarian cancer, somatic, 167000
CDKN1C	88	77,8	99,3	97,3	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CHD7	100	99,5	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CHRNG	100	100	100	100	Escobar syndrome, 265000 Multiple pterygium syndrome, lethal type, 253290
CHST14	99,9	98,9	100	100	Ehlers-Danlos syndrome, musculocontractural type 1, 601776
CILK1	99,9	98,7	100	100	Endocrine-cerebroosteodysplasia, 612651 {Epilepsy, juvenile myoclonic, susceptibility to, 10}, 617924
COL11A1	96,2	92,8	100	100	Stickler syndrome, type II, 604841 Marshall syndrome, 154780 ?Deafness, autosomal dominant 37, 618533 {Lumbar disc herniation, susceptibility to}, 603932 Fibrochondrogenesis 1, 228520
COL11A2	100	99,7	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	99,7	100	100	Achondrogenesis, type II or hypochondrogenesis, 200610 Spondyloperipheral dysplasia, 271700 Kniest dysplasia, 156550 Stickler syndrome, type I, 108300 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 Avascular necrosis of the femoral head, 608805 ?Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 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	99,2	100	100	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COLEC10	100	100	100	100	3MC syndrome 3, 248340
COLEC11	100	100	100	100	3MC syndrome 2, 265050
CPLANE1	99,7	98,4	100	100	Joubert syndrome 17, 614615 Orofaciodigital syndrome VI, 277170
CTCF	100	99,3	100	100	Mental retardation, autosomal dominant 21, 615502
CTNND1	100	100	100	100	Blepharocheilodontic syndrome 2, 617681
DDX3X	81,2	78,9	98	96,1	Intellectual developmental disorder, X-linked, syndrome, Snijders Blok type, 300958
DDX59	100	100	100	100	Orofaciodigital syndrome V, 174300
DHCR7	100	100	100	100	Smith-Lemli-Opitz syndrome, 270400
DHODH	100	100	100	100	Miller syndrome, 263750
DLL4	100	99,2	100	100	Adams-Oliver syndrome 6, 616589
DOCK6	99,3	98,9	100	100	Adams-Oliver syndrome 2, 614219
DVL1	97,2	95	100	100	Robinow syndrome, autosomal dominant 2, 616331
DVL3	100	100	100	100	Robinow syndrome, autosomal dominant 3, 616894
DYNC2H1	98,8	95,5	100	100	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2LI1	99,7	97,6	100	100	Short-rib thoracic dysplasia 15 with polydactyly, 617088
EBP	99,7	95,8	100	100	Chondrodysplasia punctata, X-linked dominant, 302960 MEND syndrome, 300960
EDN1	100	100	100	100	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}, 0
EDNRA	100	100	100	100	{Migraine, resistance to}, 157300 Mandibulofacial dysostosis with alopecia, 616367
EFNB1	100	100	100	100	Craniofrontonasal dysplasia, 304110
EFTUD2	100	99,8	100	100	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF2S3	95,4	89,1	100	100	MEHMO syndrome, 300148
EIF4A3	100	99,5	100	100	Robin sequence with cleft mandible and limb anomalies, 268305
EOGT	79,4	78,4	91,9	89	Adams-Oliver syndrome 4, 615297
EPG5	99,5	98,5	100	100	Vici syndrome, 242840
ESCO2	98,7	95,2	100	100	Roberts-SC phocomelia syndrome, 268300
EYA1	99,9	99,7	100	100	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588

					Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
FAM20C	100	100	100	99,8	Raine syndrome, 259775
FGD1	97,3	92,8	100	100	Mental retardation, X-linked syndromic 16, 305400 Aarskog-Scott syndrome, 305400
FGF8	98,2	88,9	100	99,6	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100	99,9	100	100	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Hartsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	97,7	97,1	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
FLNA	100	99,9	100	100	Otopalatodigital syndrome, type I, 311300 Congenital short bowel syndrome, 300048 Otopalatodigital syndrome, type II, 304120 Intestinal pseudoobstruction, neuronal, 300048 Melnick-Needles syndrome, 309350 Cardiac valvular dysplasia, X-linked, 314400 ?FG syndrome 2, 300321 Heterotopia, periventricular, 1, 300049 Terminal osseous dysplasia, 300244 Frontometaphyseal dysplasia 1, 305620

FLNB	99,5	98,8	100	100	Larsen syndrome, 150250 Atelosteogenesis, type I, 108720 Boomerang dysplasia, 112310 Spondylocarpotarsal synostosis syndrome, 272460 Atelosteogenesis, type III, 108721
FOXC2	100	96,7	100	99,8	Lymphedema-distichiasis syndrome, 153400 Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400
FOXE1	96,9	78,5	99,9	99,1	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
FRAS1	100	99,4	100	100	Fraser syndrome 1, 219000
FTO	83,8	83,7	94,2	94,2	{Obesity, susceptibility to, BMIQ14}, 612460 Growth retardation, developmental delay, facial dysmorphism, 612938
GDF6	100	99,9	100	99,4	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
GLI2	99,1	97,4	100	99,8	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	98,5	98	100	100	Polydactyly, postaxial, types A1 and B, 174200 Greig cephalopolysyndactyly syndrome, 175700 Polydactyly, preaxial, type IV, 174700 Pallister-Hall syndrome, 146510
GNAI3	99,3	95,2	100	100	Auriculocondylar syndrome 1, 602483
GNB1	100	100	100	100	Myelodysplastic syndrome, somatic, 614286 Mental retardation, autosomal dominant 42, 616973 Leukemia, acute lymphoblastic, somatic, 613065
GPC3	99,1	94,7	100	100	Simpson-Golabi-Behmel syndrome, type 1, 312870 Wilms tumor, somatic, 194070
GRHL3	100	100	100	100	Van der Woude syndrome 2, 606713

HDAC8	86,5	85,1	96,3	94,8	Cornelia de Lange syndrome 5, 300882
HYLS1	100	100	100	100	Hydrolethalus syndrome, 236680
IFT140	99,8	98,8	100	100	Retinitis pigmentosa 80, 617781 Short-rib thoracic dysplasia 9 with or without polydactyly, 266920
IFT172	99,9	99,1	100	100	Retinitis pigmentosa 71, 616394 Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT57	99,9	99,1	100	100	?Orofaciodigital syndrome XVIII, 617927
IFT80	97,6	88,2	100	100	Short-rib thoracic dysplasia 2 with or without polydactyly, 611263
IMPAD1	100	100	100	100	Chondrodysplasia with joint dislocations, GPAPP type, 614078
INTU	99,7	98,1	100	100	?Short-rib thoracic dysplasia 20 with polydactyly, 617925 ?Orofaciodigital syndrome XVII, 617926
IRF6	99,6	95,9	100	100	Popliteal pterygium syndrome 1, 119500 {Orofacial cleft 6}, 608864 van der Woude syndrome, 119300
KANSL1	99,9	99,2	100	100	Koolen-De Vries syndrome, 610443
KAT6A	100	99,8	100	100	Arboleda-Tham syndrome, 616268
KCNJ2	100	100	100	100	Short QT syndrome 3, 609622 Atrial fibrillation, familial, 9, 613980 Andersen syndrome, 170390
KCNK9	97,3	97,3	97,3	97,3	Birk-Barel syndrome, 612292
KDM6A	96,1	88,7	100	99,9	Kabuki syndrome 2, 300867
KIAA0586	97,3	93,1	95,8	95,8	Joubert syndrome 23, 616490 Short-rib thoracic dysplasia 14 with polydactyly, 616546
KIF7	93,6	90,6	99,1	97,8	?Hydrolethalus syndrome 2, 614120 Acrocallosal syndrome, 200990 Joubert syndrome 12, 200990 ?Al-Gazali-Bakalinova syndrome, 607131
KIFBP	96,1	96,1	96,1	96,1	Goldberg-Shprintzen megacolon syndrome, 609460
KMT2D	100	99,4	100	100	Kabuki syndrome 1, 147920
MAP3K7	100	99,6	100	100	Cardiospondylocarpofacial syndrome, 157800 Frontometaphyseal dysplasia 2, 617137
MAPRE2	100	99,3	100	100	Symmetric circumferential skin creases, congenital, 2, 616734
MASP1	100	99,9	100	100	3MC syndrome 1, 257920
MBTPS2	100	99	100	100	IFAP syndrome with or without BRESHECK syndrome, 308205 Keratosis follicularis spinulosa decalvans, X-linked, 308800 Osteogenesis imperfecta, type XIX, 301014 ?Olmsted syndrome, X-linked, 300918

MED25	100	99,8	100	100	Basel-Vanagait-Smirin-Yosef syndrome, 616449
MEIS2	100	100	100	100	Cleft palate, cardiac defects, and mental retardation, 600987
MID1	99,8	98,7	100	100	Opitz GBBB syndrome, type I, 300000
MKS1	99,8	97,9	100	100	Bardet-Biedl syndrome 13, 615990 Joubert syndrome 28, 617121 Meckel syndrome 1, 249000
MSX1	96,9	89,3	100	100	Orofacial cleft 5, 608874 Ectodermal dysplasia 3, Witkop type, 189500 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MYMK	100	100	100	100	Carey-Fineman-Ziter syndrome, 254940
NECTIN1	100	99,9	100	100	Orofacial cleft 7, 225060 Cleft lip/palate-ectodermal dysplasia syndrome, 225060
NEDD4L	72	71,5	100	100	Periventricular nodular heterotopia 7, 617201
NEK1	99,8	98	100	100	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NIPBL	98,9	97	100	100	Cornelia de Lange syndrome 1, 122470
NOTCH1	99,2	97,2	100	100	Aortic valve disease 1, 109730 Adams-Oliver syndrome 5, 616028
OFD1	88	73,7	100	99,9	Orofaciodigital syndrome I, 311200 ?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Simpson-Golabi-Behmel syndrome, type 2, 300209
ORC1	100	99,4	100	100	Meier-Gorlin syndrome 1, 224690
PAX3	100	99,9	100	100	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PGM1	94,2	94,2	94,2	94,2	Congenital disorder of glycosylation, type It, 614921
PHF8	99,7	96,8	100	100	Mental retardation syndrome, X-linked, Siderius type, 300263
PHGDH	99,9	98,8	100	100	Neu-Laxova syndrome 1, 256520 Phosphoglycerate dehydrogenase deficiency, 601815
PIEZO2	100	99,5	100	100	Arthrogryposis, distal, with impaired proprioception and touch, 617146 Arthrogryposis, distal, type 5, 108145 ?Marden-Walker syndrome, 248700 Arthrogryposis, distal, type 3, 114300
PIGN	93,8	91,5	98,8	98,8	Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080
PIGV	100	100	100	100	Hyperphosphatasia with mental retardation syndrome 1, 239300
PLCB4	99,9	98,8	100	100	Auriculocondylar syndrome 2, 614669

POLR1A	100	99,4	100	100	Acrofacial dysostosis, Cincinnati type, 616462
POLR1C	90,5	87	82,8	82,8	Treacher Collins syndrome 3, 248390 Leukodystrophy, hypomyelinating, 11, 616494
POLR1D	91,6	91,6	100	100	Treacher Collins syndrome 2, 613717
POMT1	99,3	97,5	100	100	Muscular dystrophy-dystroglycanopathy (limb-girdle), type C, 1, 609308 Muscular dystrophy-dystroglycanopathy (congenital with brain and eye anomalies), type A, 1, 236670 Muscular dystrophy-dystroglycanopathy (congenital with mental retardation), type B, 1, 613155
PORCN	100	99,1	100	100	Focal dermal hypoplasia, 305600
PQBP1	100	100	100	100	Renpenning syndrome, 309500
PROKR2	100	100	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PRRX1	100	99,7	100	100	Agnathia-otocephaly complex, 202650
PTCH1	99,2	97,6	99,9	99,8	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTCH2	99,9	99	100	100	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Medulloblastoma, somatic, 155255
RBM10	99,5	97,1	100	100	TARP syndrome, 311900
RIPK4	100	99,9	100	100	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
ROR2	100	99,9	97	97	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RPGRIP1L	96,7	95,7	100	99,5	?COACH syndrome 3, 619113 Meckel syndrome 5, 611561 Joubert syndrome 7, 611560
RPL11	100	100	100	100	Diamond-Blackfan anemia 7, 612562
RPL26	97,2	84,4	100	100	?Diamond-Blackfan anemia 11, 614900
RPL5	86,2	70	100	100	Diamond-Blackfan anemia 6, 612561
RPS19	100	99,6	100	100	Diamond-Blackfan anemia 1, 105650
RPS26	95,7	84,9	100	100	Diamond-Blackfan anemia 10, 613309
RPS28	100	94,8	100	100	Diamond Blackfan anemia 15 with mandibulofacial dysostosis, 606164
RUNX2	72,2	72,2	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
SALL4	98,6	96,7	100	100	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750

SATB2	99,7	97,4	100	100	Glass syndrome, 612313
SCARF2	95,4	86,2	99,8	99,2	Van den Ende-Gupta syndrome, 600920
SEC23A	99,7	98,2	100	100	Craniolenticulosutural dysplasia, 607812
SEMA3E	99,2	98,9	100	100	?CHARGE syndrome, 214800
SEPTIN9	100	99,9	100	100	Amyotrophy, hereditary neuralgic, 162100
SF3B4	99,9	97,3	100	100	Acrofacial dysostosis 1, Nager type, 154400
SHH	100	99,5	100	100	Schizencephaly, 269160 Microphthalmia with coloboma 5, 611638 Single median maxillary central incisor, 147250 Holoprosencephaly 3, 142945
SIX1	100	99,2	100	100	Deafness, autosomal dominant 23, 605192 Branchiootic syndrome 3, 608389
SIX3	99,9	98,6	100	100	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SIX5	95,4	88,2	100	100	Branchiootorenal syndrome 2, 610896
SKI	99,3	94,9	100	99,4	Shprintzen-Goldberg syndrome, 182212
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
SMAD3	99,9	99	100	100	Loeys-Dietz syndrome 3, 613795
SMAD4	100	99,9	100	100	Polyposis, juvenile intestinal, 174900 Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050 Myhre syndrome, 139210 Pancreatic cancer, somatic, 260350
SMC1A	100	98,7	100	99,8	Cornelia de Lange syndrome 2, 300590 Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044
SMC3	95,2	91	100	100	Cornelia de Lange syndrome 3, 610759
SMCHD1	99,5	96,3	100	100	Fascioscapulohumeral muscular dystrophy 2, digenic, 158901 Bosma arhinia microphthalmia syndrome, 603457
SMS	91,5	78,5	100	99,9	Mental retardation, X-linked, Snyder-Robinson type, 309583
SNRPB	100	99,3	100	100	Cerebrocostomandibular syndrome, 117650
SON	98,8	94,9	100	100	ZTTK syndrome, 617140
SOX9	100	98,6	100	100	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290

SPECC1L	96	95,7	97,8	96,2	Hypertelorism, Teebi type, 145420 ?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
STAC3	100	100	100	100	Myopathy, congenital, Baily-Bloch, 255995
STAMBP	100	99,4	100	100	Microcephaly-capillary malformation syndrome, 614261
TAPT1	91,7	86,9	98,5	94,8	Osteochondrodysplasia, complex lethal, Symoens-Barnes-Gistelinck type, 616897
TBX1	87	77,5	94	89,9	Conotruncal anomaly face syndrome, 217095 Velocardiofacial syndrome, 192430 DiGeorge syndrome, 188400 Tetralogy of Fallot, 187500
TBX15	100	99,9	100	100	Cousin syndrome, 260660
TBX2	99,9	97,5	99	96,9	Vertebral anomalies and variable endocrine and T-cell dysfunction, 618223
TBX22	99,2	95,7	100	100	?Abruzzo-Erickson syndrome, 302905 Cleft palate with ankyloglossia, 303400
TCOF1	99,7	98,6	100	100	Treacher Collins syndrome 1, 154500
TCTN3	100	100	100	100	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TFAP2A	99,4	94,3	100	100	Branchiooculofacial syndrome, 113620
TGDS	99,4	96,8	100	100	Catel-Manzke syndrome, 616145
TGFB3	100	100	100	100	Loeys-Dietz syndrome 5, 615582 Arrhythmogenic right ventricular dysplasia 1, 107970
TGFBR1	93,7	93,6	99	96,3	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
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
TMCO1	88	87,4	88	88	Craniofacial dysmorphism, skeletal anomalies, and mental retardation syndrome, 213980
TMEM216	99,9	98,1	100	100	Meckel syndrome 2, 603194 Joubert syndrome 2, 608091
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
TRIM37	98,6	98,1	98,7	98,7	Mulibrey nanism, 253250

TUBB	97,3	93,9	99,8	99,8	Symmetric circumferential skin creases, congenital, 1, 156610 Cortical dysplasia, complex, with other brain malformations 6, 615771
TWIST1	100	98,9	97,2	92,3	Robinow-Sorauf syndrome, 180750 Craniosynostosis 1, 123100 Sweeney-Cox syndrome, 617746 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400
TXNL4A	100	99,4	100	100	Burn-McKeown syndrome, 608572
USP9X	98,2	92,9	100	100	Mental retardation, X-linked 99, 300919 Mental retardation, X-linked 99, syndromic, female-restricted, 300968
WASHC5	100	99,8	100	100	Ritscher-Schinzel syndrome 1, 220210 Spastic paraplegia 8, autosomal dominant, 603563
WDR35	99,8	98,9	100	100	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091 Cranioectodermal dysplasia 2, 613610
WNT4	99,1	94,8	98,9	96,2	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WNT5A	100	100	100	100	Robinow syndrome, autosomal dominant 1, 180700
XYLT1	97,4	89,6	98,1	94,8	{Pseudoxanthoma elasticum, modifier of severity of}, 264800 Desbuquois dysplasia 2, 615777
ZEB2	99,9	99,1	97,4	97,4	Mowat-Wilson syndrome, 235730
ZIC2	100	98,7	98,5	95,7	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	100	99,9	100	100	Restrictive dermopathy, lethal, 275210 Mandibuloacral dysplasia with type B lipodystrophy, 608612
ZSWIM6	95,5	91,9	94,9	92,1	Acromelic frontonasal dysostosis, 603671 Neurodevelopmental disorder with movement abnormalities, abnormal gait, and autistic features, 617865

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 : March 23rd , 2021.
This list is accurate for panel version DG 3.1.0

Ad 1. “No OMIM Disease ID” signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors