Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS DG-5.1.0 (195 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
ACP4	100%	100%	100%	99.9%	99.4%	Amelogenesis imperfecta, type IJ, 617297
ACTG1	100%	100%	100%	99.9%	98.9%	Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583
ADAMTSL4	100%	100%	100%	100%	99.4%	Ectopia lentis et pupillae, 225200;Ectopia lentis, isolated, autosomal recessive, 225100
ALX1	100%	100%	100%	100%	99.7%	Frontonasal dysplasia 3, 613456
ALX3	100%	100%	100%	99.5%	98.7%	Frontonasal dysplasia 1, 136760
ALX4	100%	100%	100%	100%	99.4%	Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451
AMBN	100%	100%	100%	100%	99.9%	Amelogenesis imperfecta, type IF, 616270
AMELX	100%	100%	98.3%	89.3%	72.6%	Amelogenesis imperfecta, type 1E, 301200
AMER1	100%	100%	98.9%	90.5%	71.9%	Osteopathia striata with cranial sclerosis, 300373
AMTN	100%	100%	100%	100%	99.8%	?Amelogenesis imperfecta, type IIIB, 617607
ANKRD11	100%	100%	100%	100%	99.2%	KBG syndrome, 148050

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ARHGAP29	100%	100%	100%	100%	99.8%	
AXIN2	100%	100%	100%	100%	99.2%	Colorectal cancer, somatic, 114500;Oligodontia-colorectal cancer syndrome, 608615
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
BMP4	100%	100%	100%	100%	99.5%	Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932
BPNT2	94.9%	94.9%	100%	100%	99.5%	Chondrodysplasia with joint dislocations, GPAPP type, 614078
CCBE1	100%	100%	100%	100%	99.7%	Hennekam lymphangiectasia-lymphedema syndrome 1, 235510
CDC45	100%	100%	100%	100%	99.5%	Meier-Gorlin syndrome 7, 617063
CDON	100%	100%	100%	100%	99.9%	Holoprosencephaly 11, 614226
CDSN	100%	100%	100%	100%	99.5%	Hypotrichosis 2, 146520;Peeling skin syndrome 1, 270300
CHD7	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

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

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

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
COL9A2	100%	100%	100%	100%	99.3%	Epiphyseal dysplasia, multiple, 2, 600204;?Stickler syndrome, type V, 614284

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

COL9A3	100%	100%	100%	100%	99.4%	{Intervertebral disc disease, susceptibility to}, 603932;Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969;Stickler syndrome, type VI, 620022
COLEC11	100%	100%	100%	100%	99.6%	3MC syndrome 2, 265050
CTSK	100%	99.8%	100%	100%	99.7%	Pycnodysostosis, 265800
CYP26B1	100%	100%	100%	100%	99.3%	Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416
DHODH	89%	86.1%	100%	100%	99.5%	Miller syndrome, 263750
DHRS3	100%	100%	100%	100%	99.5%	Craniosynostosis-scoliosis syndrome, 621499
DISP1	100%	100%	100%	100%	99.8%	Holoprosencephaly 10, 621143
DLX3	100%	100%	100%	99.9%	99.4%	Trichodontoosseous syndrome, 190320;Amelogenesis imperfecta, type IV, 104510
DLX4	100%	100%	100%	100%	99.3%	?Orofacial cleft 15, 616788
DSPP	100%	100%	99.1%	98.3%	97%	Dentinogenesis imperfecta, Shields type III, 125500;Dentinogenesis imperfecta, Shields type II, 125490;Dentin dysplasia, type II, 125420;Deafness, autosomal dominant 39, with dentinogenesis, 605594

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

EDA	100%	100%	98.6%	87.3%	67.6%	Tooth agenesis, selective, X-linked 1, 313500;Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100
EDAR	100%	100%	100%	100%	99.7%	[Hair morphology 1, hair thickness], 612630;Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490;Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900
EDARADD	100%	100%	100%	100%	99.7%	Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941;Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940
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
EFNA4	100%	100%	100%	100%	99.4%	
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
EIF4A3	100%	100%	100%	100%	99.4%	Robin sequence with cleft mandible and limb anomalies, 268305

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

ENAM	100%	100%	100%	100%	99.8%	Amelogenesis imperfecta, type IC, 204650;Amelogenesis imperfecta, type IB, 104500
ERF	100%	100%	100%	100%	97.9%	Craniosynostosis 4, 600775;Chitayat syndrome, 617180
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
EZH2	100%	100%	100%	100%	99.7%	Weaver syndrome, 277590
FAM20A	100%	99.9%	100%	99.8%	99%	Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690
FAM83H	100%	100%	100%	100%	99.3%	
FGD1	100%	100%	98.4%	88%	69.3%	Aarskog-Scott syndrome, 305400
FGF10	100%	100%	100%	100%	99.9%	LADD syndrome 3, 620193;Aplasia of lacrimal and salivary glands, 180920
FGF3	100%	100%	100%	99.8%	97.8%	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF8	100%	100%	100%	99.8%	98.3%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FGF9	100%	100%	100%	100%	99.8%	Multiple synostoses syndrome 3, 612961
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

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

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

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

FGFR3	100%	100%	100%	99.9%	99.1%	Muenke syndrome, 602849;SADDAN, 616482;Hypochondroplasia, 146000;Thanatophoric dysplasia, type II, 187601;Nevus, epidermal, somatic, 162900;CATSHL syndrome, 610474;Thanatophoric dysplasia, type I, 187600;Spermatocytic seminoma, somatic, 273300;Bladder cancer, somatic, 109800;LADD syndrome 2, 620192;Achondroplasia, 100800;Cervical cancer, somatic, 603956;Colorectal cancer, somatic, 114500;Crouzon syndrome with acanthosis nigricans, 612247
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

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

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
FOXC1	100%	100%	100%	99.4%	94.5%	Axenfeld-Rieger syndrome, type 3, 602482;Anterior segment dysgenesis 3, multiple subtypes, 601631
FOXE1	100%	100%	100%	99.9%	97.1%	Bamforth-Lazarus syndrome, 241850;{Thyroid cancer, nonmedullary, 4}, 616534
GDF3	100%	100%	100%	100%	99.6%	Klippel-Feil syndrome 3, autosomal dominant, 613702;Microphthalmia, isolated, with coloboma 6, 613703;Microphthalmia, isolated 7, 613704
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

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

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
GJB6	100%	100%	100%	99.9%	99.8%	Ectodermal dysplasia 2, Clouston type, 129500;Deafness, autosomal dominant 3B, 612643;Deafness, autosomal recessive 1B, 612645;Deafness, digenic GJB2/GJB6, 220290
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
GNPTAB	100%	100%	100%	100%	99.8%	Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500
GPR68	100%	100%	100%	99.9%	99%	Amelogenesis imperfecta, hypomaturation type, IIA6, 617217

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

GRHL3	100%	100%	100%	100%	99.7%	van der Woude syndrome 2, 606713
GSC	100%	100%	100%	99.9%	98.6%	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
HOXA2	100%	100%	100%	99.6%	98%	Microtia with or without hearing impairment (AD), 612290;?Microtia, hearing impairment, and cleft palate (AR), 612290
HUWE1	100%	100%	98.9%	90.1%	71.6%	Intellectual developmental disorder, X-linked syndromic, Turner type, 309590
HYAL2	100%	100%	100%	100%	99.5%	Muggenthaler-Chowdhury-Chioza syndrome, 621063
IFT122	100%	100%	100%	100%	99.6%	Cranioectodermal dysplasia 1, 218330
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
IFT43	93.5%	93.5%	100%	100%	99.7%	?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT88	100%	100%	100%	100%	99.8%	

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

IKBKG	94.1%	93.5%	99%	90.5%	73.6%	Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081
IL11RA	92.1%	92.1%	100%	100%	99.3%	Craniosynostosis and dental anomalies, 614188
IL6ST	96.7%	96.7%	100%	100%	99.6%	Hyper-IgE syndrome 4A, autosomal dominant, with recurrent infections, 619752;Stuve-Wiedemann syndrome 2, 619751;Hyper-IgE syndrome 4B, autosomal recessive, with recurrent infections, 618523;?Immunodeficiency 94 with autoinflammation and dysmorphic facies, 619750
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
ITGB6	96.2%	96.2%	100%	100%	99.9%	Amelogenesis imperfecta, type IH, 616221
KAT6B	100%	100%	100%	100%	99.5%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

KDF1	100%	100%	100%	100%	98.8%	?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/ nail type, 617337
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
KLK4	100%	100%	100%	99.9%	99.2%	Amelogenesis imperfecta, type IIA1, 204700
KMT2D	100%	100%	100%	100%	99.2%	Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920
KREMEN1	85.3%	83.4%	100%	100%	99.2%	Ectodermal dysplasia 13, hair/tooth type, 617392
LAMB3	100%	100%	100%	99.9%	99.6%	Epidermolysis bullosa, junctional 1B, severe, 226700;Epidermolysis bullosa, junctional 1A, intermediate, 226650;Amelogenesis imperfecta, type IA, 104530
LRP2	100%	100%	100%	100%	99.8%	Donnai-Barrow syndrome, 222448

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

LRP6	98%	98%	100%	100%	99.6%	Osteopetrosis, autosomal dominant 4, 621449;{Coronary artery disease, autosomal dominant, 2}, 610947;Tooth agenesis, selective, 7, 616724;Exudative vitreoretinopathy 8, 621268
LTBP3	100%	100%	100%	99.9%	98.9%	Dental anomalies and short stature, 601216;Geleophysic dysplasia 3, 617809
MASP1	100%	100%	100%	100%	99.6%	3MC syndrome 1, 257920
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
MEGF8	100%	100%	100%	99.8%	98.9%	Carpenter syndrome 2, 614976
MEIS2	89.6%	88.5%	100%	100%	99.6%	Cleft palate, cardiac defects, and impaired intellectual development, 600987
MEOX1	100%	100%	100%	100%	98.8%	Klippel-Feil syndrome 2, 214300
MID1	100%	100%	98.8%	88.7%	69.8%	Opitz GBBB syndrome, 300000
MITF	94.7%	94.7%	100%	100%	99.9%	Waardenburg syndrome, type 2A, 193510;{Melanoma, cutaneous malignant, susceptibility to, 8}, 614456;Tietz albinism-deafness syndrome, 103500;COMMAD syndrome, 617306
MMP20	100%	100%	100%	100%	99.8%	Amelogenesis imperfecta, type IIA2, 612529

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

MN1	100%	100%	100%	99.9%	98.5%	CEBALID syndrome, 618774;Meningioma, 607174
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
MSX2	100%	100%	100%	99.9%	99%	Parietal foramina with cleidocranial dysplasia, 168550;Craniosynostosis 2, 604757;Parietal foramina 1, 168500
NAA10	100%	100%	99%	89%	68.4%	Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855
NECTIN1	93.4%	93.4%	100%	99.9%	99.1%	Cleft lip/palate-ectodermal dysplasia syndrome, 225060;Orofacial cleft 7, 225060
NFKBIA	100%	100%	100%	100%	99.3%	Ectodermal dysplasia and immunodeficiency 2, 612132
NIPBL	100%	100%	100%	100%	99.8%	Cornelia de Lange syndrome 1, 122470
NOG	100%	100%	100%	99.7%	98.3%	Symphalangism, proximal, 1A, 185800;Brachydactyly, type B2, 611377;Stapes ankylosis with broad thumbs and toes, 184460;Tarsal-carpal coalition syndrome, 186570;Multiple synostoses syndrome 1, 186500
NSD1	100%	100%	100%	100%	99.5%	Sotos syndrome, 117550
ODAPH	100%	100%	100%	100%	99.8%	Amelogenesis imperfecta, type IIA4, 614832

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

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
OTX2	100%	100%	100%	100%	99.9%	Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125
P4HB	90.9%	89.4%	100%	99.9%	99%	Cole-Carpenter syndrome 1, 112240
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
PAX6	100%	100%	100%	100%	99.5%	Optic nerve hypoplasia, 165550;Cataract with late-onset corneal dystrophy, 106210;Microphthalmia/coloboma 12, 120200;?Coloboma of optic nerve, 120430;Aniridia, 106210;Anterior segment dysgenesis 5, multiple subtypes, 604229;?Morning glory disc anomaly, 120430;Foveal hypoplasia 1, 136520;Keratitis, 148190

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PAX7	94.2%	94.2%	100%	99.9%	99.1%	Congenital myopathy 19, 618578;Rhabdomyosarcoma 2, alveolar, 268220
PAX9	100%	100%	100%	100%	99.4%	Tooth agenesis, selective, 3, 604625
PGM1	87.5%	87.5%	100%	100%	99.4%	Congenital disorder of glycosylation, type It, 614921
PITX2	100%	100%	100%	100%	99.1%	Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600
PLCB4	99%	99%	100%	100%	99.8%	Auriculocondylar syndrome 2B, 620458;Auriculocondylar syndrome 2A, 614669
POLR1B	100%	100%	100%	100%	99.7%	Treacher-Collins syndrome 4, 618939
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
POR	98%	94.9%	100%	100%	99.6%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PORCN	100%	100%	98.5%	89.4%	70.4%	Focal dermal hypoplasia, 305600
PRRX1	100%	100%	100%	100%	99.5%	Agnathia-otocephaly complex, 202650

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

PTCH1	100%	100%	100%	100%	99.2%	Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828
PTH1R	100%	100%	100%	100%	99%	Metaphyseal chondrodysplasia, Murk Jansen type, 156400;Eiken syndrome, 600002;Failure of tooth eruption, primary, 125350;Chondrodysplasia, Blomstrand type, 215045
RAB23	100%	100%	100%	100%	99.4%	Carpenter syndrome 1, 201000
RAD21	100%	100%	100%	99.9%	99.6%	Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376
RBM10	95.6%	95.6%	98.7%	88.7%	70.4%	TARP syndrome, 311900
RECQL4	100%	100%	100%	100%	99.4%	Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280
RIPK4	100%	100%	100%	100%	99.7%	CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RNU12						CDAGS syndrome, 603116;?Spinocerebellar ataxia, autosomal recessive 33, 620208

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

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
SALL1	100%	100%	100%	100%	99.2%	Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480
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
SEC24D	100%	100%	100%	100%	99.6%	Cole-Carpenter syndrome 2, 616294
SEMA3E	100%	100%	100%	100%	99.9%	
SF3B2	100%	100%	100%	100%	99.4%	Craniofacial microsomia, 164210
SF3B4	100%	100%	100%	100%	99.1%	Acrofacial dysostosis 1, Nager type, 154400
SH3BP2	100%	99.7%	100%	100%	99.5%	Cherubism, 118400
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

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

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
SLC24A4	100%	100%	100%	100%	99.3%	[Skin/hair/eye pigmentation 6, blond/brown hair], 210750;Amelogenesis imperfecta, type IIA5, 615887;[Skin/hair/eye pigmentation 6, blue/green eyes], 210750
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
SMAD6	100%	100%	100%	99.8%	98.1%	Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439
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

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

SMO	100%	100%	100%	100%	99.7%	Pallister-Hall-like syndrome, 241800;Basal cell carcinoma, somatic, 605462;Curry-Jones syndrome, somatic mosaic, 601707
SMOC2	100%	100%	100%	100%	99.6%	Dentin dysplasia, type IA, 125400
SNAI2	100%	100%	100%	100%	99.7%	
SOX10	97.8%	97.8%	100%	100%	99.2%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX6	100%	100%	100%	100%	99.6%	Tolchin-Le Caignec syndrome, 618971
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
STAG2	100%	100%	99.3%	91.5%	74%	Holoprosencephaly 13, X-linked, 301043;Mullegama-Klein-Martinez syndrome, 301022
SUMO1	71%	71%	100%	100%	99.9%	?Orofacial cleft 10, 613705

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

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
TBX22	100%	100%	99%	91.2%	72.3%	Cleft palate with ankyloglossia, 303400;?Abruzzo-Erickson syndrome, 302905
TCF12	96.1%	96.1%	100%	100%	99.7%	Craniosynostosis 3, 615314;Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TCOF1	97.9%	97.9%	100%	100%	99.6%	Treacher Collins syndrome 1, 154500
TFAP2A	100%	100%	100%	100%	98.8%	Branchiooculofacial syndrome, 113620
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
TLK2	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal dominant 57, 618050

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

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
TRAF6	100%	100%	100%	100%	99.8%	
TSHZ1	100%	100%	100%	99.9%	99.4%	Aural atresia, congenital, 607842
TSPEAR	93.9%	93.9%	100%	100%	99.1%	Tooth agenesis, selective, 10, 620173;?Deafness, autosomal recessive 98, 614861;Ectodermal dysplasia 14, hypohidrotic/hair/tooth/ nail type, 618180
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
UBB	100%	100%	100%	99.9%	97.7%	
VAX1	100%	100%	100%	100%	98.5%	?Microphthalmia, syndromic 11, 614402

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

WDR19	100%	100%	100%	100%	99.8%	Nephronophthisis 13, 614377;Cranioectodermal dysplasia 4, 614378;Senior-Loken syndrome 8, 616307;Short-rib thoracic dysplasia 5 with or without polydactyly, 614376;?Spermatogenic failure 72, 619867
WDR35	100%	100%	100%	100%	99.7%	Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610
WDR72	100%	100%	100%	100%	99.6%	Amelogenesis imperfecta, type IIA3, 613211
WNT10A	79.3%	79.3%	100%	100%	99.7%	Schopf-Schulz-Passarge syndrome, 224750;Tooth agenesis, selective, 4, 150400;Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980
WNT10B	100%	100%	100%	99.7%	98.8%	Tooth agenesis, selective, 8, 617073;Split-hand/foot malformation 6, 225300
ZEB2	100%	100%	100%	99.9%	99.4%	Mowat-Wilson syndrome, 235730
ZIC1	100%	100%	100%	99.9%	98.6%	?Craniosynostosis 6, 616602;Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736
ZIC2	100%	100%	99.9%	99.6%	97%	Holoprosencephaly 5, 609637

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

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

Evaluation Only. Created with Aspose.PDF. Copyright 2002-2025 Aspose Pty Ltd.

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 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors