# CRANIOFACIAL ANOMALIES PANEL WITH GENOME WIDE CNV ANALYSIS DG-4.2.0 (192 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% | 100% | 99.1% | Amelogenesis imperfecta, type IJ, 617297 |
| ACTG1 | 100% | 100% | 100% | 99.9% | 98.5% | Deafness, autosomal dominant 20/26, 604717;Baraitser-Winter syndrome 2, 614583 |
| ADAMTSL4 | 100% | 100% | 100% | 99.9% | 98.8% | Ectopia lentis et pupillae, 225200;Ectopia lentis, isolated, autosomal recessive, 225100 |
| ALX1 | 100% | 100% | 100% | 100% | 99.4% | Frontonasal dysplasia 3, 613456 |
| ALX3 | 100% | 100% | 100% | 99.8% | 97.8% | Frontonasal dysplasia 1, 136760 |

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| 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% | 99.5% | Amelogenesis imperfecta, type IF, 616270 |
| AMELX | 100% | 100% | 99% | 90.1% | 67.2% | Amelogenesis imperfecta, type 1E, 301200 |
| AMER1 | 100% | 99.9% | 98.5% | 85.6% | 65% | Osteopathia striata with cranial sclerosis, 300373 |
| AMTN | 100% | 100% | 100% | 100% | 99.7% | ?Amelogenesis imperfecta, type IIIB, 617607 |
| ANKRD11 | 100% | 100% | 100% | 99.9% | 98.7% | KBG syndrome, 148050 |
| ARHGAP29 | 100% | 100% | 100% | 100% | 99.6% | |
| AXIN2 | 100% | 100% | 100% | 99.9% | 98.4% | Colorectal cancer, somatic, 114500;Oligodontia-colorectal cancer syndrome, 608615 |
| BCOR | 100% | 99.4% | 98.6% | 87.3% | 66.6% | Microphthalmia, syndromic 2, 300166 |

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| BMP2 | 100% | 100% | 100% | 100% | 98.3% | 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% | 99.3% | Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932 |
| BPNT2 | 100% | 100% | 100% | 100% | 99.6% | Chondrodysplasia with joint dislocations, GPAPP type, 614078 |
| CCBE1 | 100% | 100% | 100% | 100% | 99.3% | Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 |
| CDC45 | 100% | 100% | 100% | 100% | 99.3% | Meier-Gorlin syndrome 7, 617063 |
| CDON | 100% | 100% | 100% | 100% | 99.6% | Holoprosencephaly 11, 614226 |
| CDSN | 100% | 100% | 100% | 100% | 99.4% | Hypotrichosis 2, 146520;Peeling skin syndrome 1, 270300 |
| CHD7 | 100% | 100% | 100% | 100% | 99.4% | Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800 |

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| COL11A1 | 100% | 100% | 100% | 100% | 99.3% | 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% | 99.9% | 98.7% | Deafness, autosomal dominant 13, 601868;Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150;Fibrochondrogenesis 2, 614524;Deafness, autosomal recessive 53, 609706;Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 |

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| COL2A1 | 100% | 100% | 100% | 99.9% | 99.2% | ?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, |

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| COL9A1 | 100% | 100% | 100% | 100% | 99.6% | Stickler syndrome, type IV, 614134;?Epiphyseal dysplasia, multiple, 6, 614135 |
| COL9A2 | 100% | 100% | 100% | 100% | 98.8% | Epiphyseal dysplasia, multiple, 2, 600204;?Stickler syndrome, type V, 614284 |
| COL9A3 | 100% | 99.9% | 100% | 99.9% | 98.2% | {Intervertebral disc disease, susceptibility to}, 603932;Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969;Stickler syndrome, type VI, 620022 |
| COLEC11 | 100% | 100% | 100% | 99.8% | 99% | 3MC syndrome 2, 265050 |
| CTSK | 100% | 100% | 100% | 100% | 99.6% | Pycnodysostosis, 265800 |
| CYP26B1 | 100% | 100% | 100% | 99.9% | 98.9% | Craniosynostosis with radiohumeral fusions and other skeletal and craniofacial anomalies, 614416 |
| DHODH | 100% | 100% | 100% | 99.7% | 98.6% | Miller syndrome, 263750 |
| DISP1 | 100% | 100% | 100% | 100% | 99.2% | |

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| DLX3 | 100% | 100% | 100% | 100% | 98.8% | Trichodontoosseous syndrome, 190320;Amelogenesis imperfecta, type IV, 104510 |
| DLX4 | 100% | 100% | 100% | 99.9% | 98.9% | ?Orofacial cleft 15, 616788 |
| DSPP | 100% | 100% | 99% | 98% | 96.1% | Dentinogenesis imperfecta, Shields type III, 125500;Dentinogenesis imperfecta, Shields type II, 125490;Dentin dysplasia, type II, 125420;Deafness, autosomal dominant 39, with dentinogenesis, 605594 |
| EDA | 100% | 99.7% | 98.3% | 85.7% | 64.2% | Tooth agenesis, selective, X-linked 1, 313500;Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 |

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| EDAR | 100% | 100% | 100% | 100% | 99.1% | [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% | 99.9% | 98.2% | Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941;Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 |
| EDN1 | 100% | 100% | 100% | 99.9% | 98.9% | Question mark ears, isolated, 612798;Auriculocondylar syndrome 3, 615706 |
| EDNRA | 100% | 100% | 100% | 100% | 99.5% | {Migraine, resistance to}, 157300;Mandibulofacial dysostosis with alopecia, 616367 |
| EFNA4 | 100% | 100% | 100% | 100% | 99.1% | |

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| EFNB1 | 100% | 100% | 98.7% | 87.4% | 67.5% | Craniofrontonasal dysplasia, 304110 |
| EFTUD2 | 100% | 100% | 100% | 100% | 99% | Mandibulofacial dysostosis, Guion-Almeida type, 610536 |
| EIF4A3 | 100% | 100% | 100% | 100% | 99.2% | Robin sequence with cleft mandible and limb anomalies, 268305 |
| ENAM | 100% | 100% | 100% | 100% | 99.7% | Amelogenesis imperfecta, type IC, 204650;Amelogenesis imperfecta, type IB, 104500 |
| ERF | 100% | 100% | 100% | 99.6% | 96% | Craniosynostosis 4, 600775;Chitayat syndrome, 617180 |
| ESCO2 | 100% | 100% | 100% | 100% | 99.3% | Juberg-Hayward syndrome, 216100;Roberts-SC phocomelia syndrome, 268300 |
| EYA1 | 100% | 100% | 100% | 100% | 99.2% | Branchiootic syndrome 1, 602588;Branchiootorenal syndrome 1, with or without cataracts, 113650;Anterior segment anomalies with or without cataract, 602588;?Otofaciocervical syndrome, 166780 |

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| EZH2 | 100% | 100% | 100% | 100% | 99.5% | Weaver syndrome, 277590 |
| FAM20A | 100% | 100% | 100% | 99.9% | 99% | Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 |
| FAM83H | 100% | 100% | 100% | 100% | 99.2% | Amelogenesis imperfecta, type IIIA, 130900 |
| FGD1 | 100% | 99% | 98.8% | 87.6% | 67.6% | Intellectual developmental disorder, X-linked syndromic 16, 305400;Aarskog-Scott syndrome, 305400 |
| FGF10 | 100% | 100% | 100% | 100% | 99.8% | LADD syndrome 3, 620193;Aplasia of lacrimal and salivary glands, 180920 |
| FGF3 | 100% | 100% | 100% | 99.4% | 95.9% | Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 |
| FGF8 | 100% | 100% | 100% | 99.7% | 97.5% | Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 |
| FGF9 | 100% | 100% | 100% | 100% | 99.5% | Multiple synostoses syndrome 3, 612961 |

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| FGFR1 | 99.9% | 98.9% | 100% | 100% | 99.2% | 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 |

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| FGFR2 | 100% | 100% | 100% | 99.9% | 99% | 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, |

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| FGFR3 | 100% | 100% | 100% | 99.9% | 98.4% | 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 |

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| FLNA | 100% | 99.7% | 98.4% | 86.7% | 66.4% | Otopalatodigital syndrome, type II, 304120;Intestinal pseudoobstruction, neuronal, 300048;Cardiac valvular dysplasia, X-linked, 314400;?FG syndrome 2, 300321;Melnick-Needles syndrome, 309350;Terminal osseous dysplasia, 300244;Congenital short bowel syndrome, 300048;Otopalatodigital syndrome, type I, 311300;Heterotopia, periventricular, 1, 300049;Frontometaphyseal dysplasia 1, 305620 |
| FLNB | 100% | 100% | 100% | 100% | 99.2% | Larsen syndrome, 150250;Atelosteogenesis, type I, 108720;Atelosteogenesis, type III, 108721;Spondylocarpotarsal synostosis syndrome, 272460;Boomerang dysplasia, 112310 |

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| FOXC1 | 100% | 99.7% | 100% | 98.9% | 92.7% | Axenfeld-Rieger syndrome, type 3, 602482;Anterior segment dysgenesis 3, multiple subtypes, 601631 |
| FOXE1 | 100% | 100% | 100% | 99.5% | 95% | Bamforth-Lazarus syndrome, 241850;{Thyroid cancer, nonmedullary, 4}, 616534 |
| GDF3 | 100% | 100% | 100% | 100% | 99.4% | Klippel-Feil syndrome 3, autosomal dominant, 613702;Microphthalmia, isolated, with coloboma 6, 613703;Microphthalmia, isolated 7, 613704 |
| GDF6 | 100% | 100% | 100% | 99.8% | 97.7% | 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 |

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| GJA1 | 100% | 100% | 100% | 100% | 98.9% | 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% | 99.5% | 97.5% | 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% | 98.7% | Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829 |

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| 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% | 99.9% | 98.6% | Auriculocondylar syndrome 1, 602483 |
| GNPTAB | 100% | 100% | 100% | 100% | 99.7% | Mucolipidosis III alpha/beta, 252600;Mucolipidosis II alpha/beta, 252500 |
| GPR68 | 100% | 100% | 100% | 100% | 99.3% | Amelogenesis imperfecta, hypomaturation type, IIA6, 617217 |
| GRHL3 | 100% | 99.9% | 100% | 99.9% | 98.8% | van der Woude syndrome 2, 606713 |
| GSC | 100% | 100% | 100% | 99.9% | 97% | Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471 |
| HOXA2 | 100% | 100% | 100% | 99.9% | 98.5% | Microtia with or without hearing impairment (AD), 612290;?Microtia, hearing impairment, and cleft palate (AR), 612290 |

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| HUWE1 | 100% | 99.6% | 98.6% | 87.6% | 67.5% | Intellectual developmental disorder, X-linked syndromic, Turner type, 309590 |
| HYAL2 | 100% | 100% | 100% | 99.9% | 99.3% | |
| IFT122 | 100% | 100% | 100% | 100% | 99.3% | Cranioectodermal dysplasia 1, 218330 |
| IFT43 | 100% | 100% | 100% | 100% | 99.5% | ?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866 |
| IFT88 | 100% | 100% | 100% | 100% | 99.4% | |
| IKBKG | 96.4% | 94.2% | 98.8% | 90.3% | 72.8% | Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081 |
| IL11RA | 100% | 100% | 100% | 99.9% | 98.3% | Craniosynostosis and dental anomalies, 614188 |

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| IL6ST | 100% | 100% | 100% | 100% | 99.5% | 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% | 99.9% | 99.3% | ?Orofaciodigital syndrome XVII, 617926;?Short-rib thoracic dysplasia 20 with polydactyly, 617925 |
| IRF6 | 100% | 100% | 100% | 99.9% | 99% | {Orofacial cleft 6}, 608864;Popliteal pterygium syndrome 1, 119500;van der Woude syndrome 1, 119300 |
| ITGB6 | 100% | 100% | 100% | 99.9% | 99.4% | Amelogenesis imperfecta, type IH, 616221 |
| KAT6B | 100% | 100% | 100% | 100% | 99.2% | SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170 |

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| KDF1 | 100% | 100% | 100% | 99.9% | 98.6% | ?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/ nail type, 617337 |
| KDM1A | 96.9% | 96.9% | 100% | 99.9% | 99.2% | Cleft palate, psychomotor retardation, and distinctive facial features, 616728;{ACTH-independent macronodular adrenal hyperplasia 3}, 620990 |
| KDM6A | 100% | 100% | 99.1% | 89.6% | 70.8% | Kabuki syndrome 2, 300867 |
| KLK4 | 100% | 100% | 100% | 100% | 98% | Amelogenesis imperfecta, type IIA1, 204700 |
| KMT2D | 100% | 100% | 100% | 99.9% | 98.3% | Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920 |
| KREMEN1 | 100% | 99.7% | 100% | 100% | 99.1% | Ectodermal dysplasia 13, hair/tooth type, 617392 |

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| LAMB3 | 100% | 100% | 100% | 99.9% | 98.6% | Epidermolysis bullosa, junctional 1B, severe, 226700;Epidermolysis bullosa, junctional 1A, intermediate, 226650;Amelogenesis imperfecta, type IA, 104530 |
| LRP2 | 100% | 100% | 100% | 100% | 99.4% | Donnai-Barrow syndrome, 222448 |
| LRP6 | 100% | 100% | 100% | 100% | 99.2% | {Coronary artery disease, autosomal dominant, 2}, 610947;Tooth agenesis, selective, 7, 616724 |
| LTBP3 | 100% | 100% | 100% | 99.8% | 97.5% | Dental anomalies and short stature, 601216;Geleophysic dysplasia 3, 617809 |
| MASP1 | 100% | 100% | 100% | 99.9% | 99.1% | 3MC syndrome 1, 257920 |
| MED12 | 100% | 99.7% | 98.4% | 86% | 64.8% | Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450 |
| MEGF8 | 100% | 100% | 99.9% | 99.5% | 97.6% | Carpenter syndrome 2, 614976 |

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| MEIS2 | 91.5% | 91.5% | 100% | 100% | 98.6% | Cleft palate, cardiac defects, and impaired intellectual development, 600987 |
| MEOX1 | 100% | 99.9% | 100% | 99.9% | 98.6% | Klippel-Feil syndrome 2, 214300 |
| MID1 | 100% | 99.9% | 98.7% | 88.6% | 67.1% | Opitz GBBB syndrome, 300000 |
| MITF | 100% | 100% | 100% | 100% | 99.3% | 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% | 99.7% | Amelogenesis imperfecta, type IIA2, 612529 |
| MN1 | 100% | 100% | 100% | 99.9% | 98.8% | CEBALID syndrome, 618774;Meningioma, 607174 |
| MSX1 | 100% | 100% | 100% | 100% | 99.6% | Tooth agenesis, selective, 1, with or without orofacial cleft, 106600;Ectodermal dysplasia 3, Witkop type, 189500;Orofacial cleft 5, 608874 |

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| MSX2 | 100% | 100% | 100% | 99.8% | 98.8% | Parietal foramina with cleidocranial dysplasia, 168550;Craniosynostosis 2, 604757;Parietal foramina 1, 168500 |
| NAA10 | 100% | 99.7% | 98.4% | 88.4% | 66.6% | Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855 |
| NECTIN1 | 93.4% | 93.4% | 100% | 99.8% | 98% | Cleft lip/palate-ectodermal dysplasia syndrome, 225060;Orofacial cleft 7, 225060 |
| NFKBIA | 100% | 100% | 100% | 100% | 99.1% | Ectodermal dysplasia and immunodeficiency 2, 612132 |
| NIPBL | 100% | 100% | 100% | 99.9% | 99.3% | Cornelia de Lange syndrome 1, 122470 |
| NOG | 100% | 99.9% | 100% | 99.8% | 98.6% | 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 |

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| ODAPH | 100% | 100% | 100% | 100% | 99.8% | Amelogenesis imperfecta, type IIA4, 614832 |
| OFD1 | 100% | 99.9% | 98.8% | 90.2% | 70.7% | Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804 |
| OTX2 | 100% | 100% | 100% | 100% | 99.3% | Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125;Pituitary hormone deficiency, combined, 6, 613986;Microphthalmia, syndromic 5, 610125 |
| P4HB | 96% | 94.7% | 100% | 100% | 99.2% | Cole-Carpenter syndrome 1, 112240 |
| PAX3 | 100% | 100% | 100% | 99.9% | 98.5% | Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220 |

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| PAX6 | 100% | 100% | 100% | 100% | 99% | 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 |
| PAX7 | 100% | 100% | 100% | 99.9% | 98.4% | Congenital myopathy 19, 618578;Rhabdomyosarcoma 2, alveolar, 268220 |
| PAX9 | 100% | 100% | 100% | 100% | 98.9% | Tooth agenesis, selective, 3, 604625 |
| PGM1 | 94% | 94% | 100% | 100% | 99.2% | Congenital disorder of glycosylation, type It, 614921 |

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| PITX2 | 100% | 99.8% | 100% | 99.9% | 98.7% | Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600 |
| PLCB4 | 99% | 99% | 100% | 100% | 99.6% | Auriculocondylar syndrome 2B, 620458;Auriculocondylar syndrome 2A, 614669 |
| POLR1B | 100% | 100% | 100% | 100% | 99.4% | Treacher-Collins syndrome 4, 618939 |
| POLR1C | 83.4% | 83.2% | 100% | 99.9% | 98.6% | Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390 |
| POLR1D | 100% | 100% | 100% | 100% | 99.6% | Treacher Collins syndrome 2, 613717 |
| POR | 100% | 100% | 100% | 100% | 99.2% | Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 |
| PORCN | 100% | 99.7% | 98.5% | 86.5% | 66% | Focal dermal hypoplasia, 305600 |

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| PRRX1 | 100% | 100% | 100% | 99.7% | 99% | Agnathia-otocephaly complex, 202650 |
| PTCH1 | 100% | 100% | 100% | 99.9% | 98.7% | Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828 |
| PTH1R | 100% | 100% | 100% | 100% | 98.7% | Metaphyseal chondrodysplasia, Murk Jansen type, 156400;Eiken syndrome, 600002;Failure of tooth eruption, primary, 125350;Chondrodysplasia, Blomstrand type, 215045 |
| RAB23 | 100% | 100% | 100% | 100% | 99.3% | Carpenter syndrome, 201000 |
| RAD21 | 100% | 100% | 100% | 100% | 99.6% | Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376 |
| RBM10 | 100% | 99.6% | 98% | 86.5% | 66.1% | TARP syndrome, 311900 |
| RECQL4 | 100% | 100% | 100% | 99.9% | 99.1% | Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280 |

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|---|---|---|---|---|---|---|
| RIPK4 | 100% | 99.9% | 100% | 99.9% | 99.3% | CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650 |
| RNU12 | | | | | | CDAGS syndrome, 603116;?Spinocerebellar ataxia, autosomal recessive 33, 620208 |
| RUNX2 | 100% | 100% | 100% | 100% | 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% | 99.3% | Townes-Brocks syndrome 1, 107480;Townes-Brocks branchiootorenal-like syndrome, 107480 |
| SALL4 | 100% | 100% | 100% | 99.9% | 99.3% | ?IVIC syndrome, 147750;Duane-radial ray syndrome, 607323 |
| SATB2 | 100% | 100% | 100% | 100% | 99% | Glass syndrome, 612313 |
| SCARF2 | 100% | 99.8% | 100% | 99.6% | 95.9% | Van den Ende-Gupta syndrome, 600920 |

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|---|---|---|---|---|---|---|
| SEC24D | 100% | 100% | 100% | 100% | 99.4% | Cole-Carpenter syndrome 2, 616294 |
| SEMA3E | 100% | 100% | 100% | 100% | 99.3% | |
| SF3B2 | 100% | 100% | 100% | 99.9% | 98.9% | Craniofacial microsomia, 164210 |
| SF3B4 | 100% | 100% | 100% | 99.9% | 97.8% | Acrofacial dysostosis 1, Nager type, 154400 |
| SH3BP2 | 99% | 97.6% | 100% | 100% | 98.7% | Cherubism, 118400 |
| SHH | 100% | 100% | 100% | 100% | 97.8% | Single median maxillary central incisor, 147250;Holoprosencephaly 3, 142945;Microphthalmia/coloboma 5, 611638 |
| SIX1 | 100% | 100% | 100% | 100% | 99.4% | Deafness, autosomal dominant 23, 605192;Branchiootic syndrome 3, 608389 |
| SIX3 | 100% | 100% | 100% | 99.8% | 96.5% | Schizencephaly, 269160;Holoprosencephaly 2, 157170 |
| SIX5 | 100% | 100% | 100% | 99.8% | 97.9% | Branchiootorenal syndrome 2, 610896 |
| SKI | 100% | 99.7% | 100% | 99.8% | 97.8% | Shprintzen-Goldberg syndrome, 182212 |

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|---|---|---|---|---|---|---|
| SLC24A4 | 100% | 100% | 100% | 100% | 99.5% | [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% | 96.7% | Aortic valve disease 2, 614823;{Radioulnar synostosis, nonsyndromic}, 179300;{Craniosynostosis 7, susceptibility to}, 617439 |

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|---|---|---|---|---|---|---|
| SMC1A | 100% | 99.7% | 98.5% | 86.5% | 65% | Cornelia de Lange syndrome 2, 300590;Developmental and epileptic encephalopathy 85, with or without midline brain defects, 301044 |
| SMC3 | 100% | 100% | 100% | 99.9% | 99.3% | Cornelia de Lange syndrome 3, 610759 |
| SMO | 100% | 100% | 100% | 100% | 99.1% | Pallister-Hall-like syndrome, 241800;Basal cell carcinoma, somatic, 605462;Curry-Jones syndrome, somatic mosaic, 601707 |
| SMOC2 | 100% | 100% | 100% | 100% | 98.9% | Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400 |
| SNAI2 | 100% | 100% | 100% | 100% | 99.6% | |
| SOX10 | 97.8% | 97.8% | 100% | 100% | 98.2% | Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 |
| SOX6 | 100% | 100% | 100% | 99.9% | 99.4% | Tolchin-Le Caignec syndrome, 618971 |

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| SOX9 | 100% | 100% | 100% | 99.5% | 95.8% | Campomelic dysplasia with autosomal sex reversal, 114290;Acampomelic campomelic dysplasia, 114290;Campomelic dysplasia, 114290 |
| SPECC1L | 100% | 99.9% | 100% | 100% | 99.5% | Teebi hypertelorism syndrome 1, 145420;?Facial clefting, oblique, 1, 600251 |
| SUMO1 | 71% | 71% | 100% | 100% | 99.9% | ?Orofacial cleft 10, 613705 |
| TBX1 | 96% | 91.6% | 99.8% | 98.8% | 93.2% | Tetralogy of Fallot, 187500;DiGeorge syndrome, 188400;Conotruncal anomaly face syndrome, 217095;Velocardiofacial syndrome, 192430 |
| TBX22 | 100% | 99.9% | 99% | 90.3% | 68.1% | Cleft palate with ankyloglossia, 303400;?Abruzzo-Erickson syndrome, 302905 |
| TCF12 | 100% | 100% | 100% | 100% | 99.4% | Craniosynostosis 3, 615314;Hypogonadotropic hypogonadism 26 with or without anosmia, 619718 |
| TCOF1 | 100% | 100% | 100% | 99.9% | 98.9% | Treacher Collins syndrome 1, 154500 |

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|---|---|---|---|---|---|---|
| TFAP2A | 100% | 100% | 100% | 99.9% | 97.7% | Branchiooculofacial syndrome, 113620 |
| TGFBR1 | 99.8% | 99.3% | 100% | 100% | 99.4% | {Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192 |
| TGFBR2 | 100% | 100% | 100% | 99.9% | 98.8% | Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239 |
| TGIF1 | 100% | 100% | 100% | 100% | 99.5% | Holoprosencephaly 4, 142946 |
| TLK2 | 100% | 100% | 100% | 99.9% | 99.4% | Intellectual developmental disorder, autosomal dominant 57, 618050 |

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|---|---|---|---|---|---|---|
| TP63 | 100% | 100% | 100% | 99.9% | 99% | 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% | 99.5% | |
| TSHZ1 | 100% | 100% | 100% | 99.9% | 98.8% | Aural atresia, congenital, 607842 |
| TSPEAR | 100% | 100% | 100% | 99.9% | 98.4% | Tooth agenesis, selective, 10, 620173;?Deafness, autosomal recessive 98, 614861;Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 |

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|---|---|---|---|---|---|---|
| TWIST1 | 99.9% | 98.9% | 100% | 100% | 98.8% | Craniosynostosis 1, 123100;Robinow-Sorauf syndrome, 180750;Sweeney-Cox syndrome, 617746;Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 |
| UBB | 100% | 100% | 100% | 100% | 99.3% | |
| VAX1 | 99.4% | 96.4% | 100% | 100% | 98.5% | ?Microphthalmia, syndromic 11, 614402 |
| WDR19 | 100% | 100% | 100% | 100% | 99.6% | 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% | 99.4% | Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610 |
| WDR72 | 96.8% | 96.8% | 100% | 100% | 99.5% | Amelogenesis imperfecta, type IIA3, 613211 |

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|---|---|---|---|---|---|---|
| WNT10A | 100% | 100% | 100% | 100% | 98.8% | Schopf-Schulz-Passarge syndrome, 224750;Tooth agenesis, selective, 4, 150400;Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980 |
| WNT10B | 100% | 100% | 100% | 100% | 99.2% | Tooth agenesis, selective, 8, 617073;Split-hand/foot malformation 6, 225300 |
| ZEB2 | 100% | 100% | 100% | 99.8% | 98.5% | Mowat-Wilson syndrome, 235730 |
| ZIC1 | 100% | 100% | 100% | 100% | 98.9% | ?Craniosynostosis 6, 616602;Structural brain anomalies with impaired intellectual development and craniosynostosis, 618736 |
| ZIC2 | 100% | 98.1% | 100% | 99.3% | 95.3% | 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 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 4.2.0*

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