# CRANIOFACIAL ANOMALIES GENE DG 2.17 ( 171 genes)

Releasedate: 06-12-2019

| *Gene* | *Median Coverage* | *% covered > 10x* | *% covered > 20x* | *Associated Phenotype description and OMIM disease ID* |
|---|---|---|---|---|
| ACP4 | 102.5 | 98.0% | 90.4% | Amelogenesis imperfecta, type IJ, 617297 |
| ADAMTSL4 | 137.7 | 100.0% | 99.8% | Ectopia lentis et pupillae, 225200<br>Ectopia lentis, isolated, autosomal recessive, 225100 |
| ALX1 | 139.5 | 100.0% | 99.2% | ?Frontonasal dysplasia 3, 613456 |
| ALX3 | 148.9 | 91.7% | 80.3% | Frontonasal dysplasia 1, 136760 |
| ALX4 | 175.4 | 100.0% | 100.0% | Frontonasal dysplasia 2, 613451<br>Parietal foramina 2, 609597 |
| AMBN | 181.1 | 99.3% | 95.8% | Amelogenesis imperfecta, type IF, 616270 |
| AMELX | 90.7 | 99.0% | 93.8% | Amelogenesis imperfecta, type 1E, 301200 |
| AMER1 | 106.2 | 99.8% | 99.1% | Osteopathia striata with cranial sclerosis, 300373 |
| AMTN | 120.4 | 99.7% | 98.1% | ?Amelogenesis imperfecta, type IIIB, 617607 |
| ANKRD11 | 131.8 | 99.6% | 97.6% | KBG syndrome, 148050 |
| ARHGAP29 | 137.9 | 99.9% | 98.5% | No OMIM Disease ID |
| AXIN2 | 134.4 | 100.0% | 99.9% | Colorectal cancer, somatic, 114500<br>Oligodontia-colorectal cancer syndrome, 608615 |
| BCOR | 109.0 | 99.2% | 96.2% | Microphthalmia, syndromic 2, 300166 |
| BMP2 | 180.6 | 100.0% | 100.0% | Brachydactyly, type A2, 112600<br>Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 |
| BMP4 | 192.0 | 100.0% | 100.0% | Microphthalmia, syndromic 6, 607932<br>Orofacial cleft 11, 600625 |
| C4orf26 | 207.0 | 100.0% | 100.0% | Amelogenesis imperfecta, type IIA4, 614832 |
| CDC45 | 148.0 | 99.6% | 98.4% | Meier-Gorlin syndrome 7, 617063 |
| CDON | 110.7 | 99.9% | 99.0% | Holoprosencephaly 11, 614226 |
| CDSN | 141.6 | 100.0% | 100.0% | Hypotrichosis 2, 146520<br>Peeling skin syndrome 1, 270300 |
| CHD7 | 143.6 | 100.0% | 99.5% | CHARGE syndrome, 214800<br>Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 |
| COL11A1 | 94.6 | 98.0% | 93.6% | Stickler syndrome, type II, 604841<br>Marshall syndrome, 154780 |

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| | | | | ?Deafness, autosomal dominant 37, 618533<br>Fibrochondrogenesis 1, 228520 |
| COL11A2 | 122.3 | 100.0% | 99.5% | Deafness, autosomal dominant 13, 601868<br>Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150<br>Fibrochondrogenesis 2, 614524<br>Deafness, autosomal recessive 53, 609706<br>Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 |
| COL2A1 | 121.1 | 100.0% | 99.8% | Achondrogenesis, type II or hypochondrogenesis, 200610<br>Spondyloperipheral dysplasia, 271700<br>Kniest dysplasia, 156550<br>Stickler syndrome, type I, 108300<br>Epiphyseal dysplasia, multiple, with myopia and deafness, 132450<br>Osteoarthritis with mild chondrodysplasia, 604864<br>Platyspondylic skeletal dysplasia, Torrance type, 151210<br>Avascular necrosis of the femoral head, 608805<br>SED congenita, 183900<br>Legg-Calve-Perthes disease, 150600<br>SMED Strudwick type, 184250<br>Czech dysplasia, 609162<br>Stickler sydrome, type I, nonsyndromic ocular, 609508<br>Spondyloepiphyseal dysplasia, Stanescu type, 616583<br>Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0 |
| COL9A1 | 132.9 | 100.0% | 99.5% | ?Epiphyseal dysplasia, multiple, 6, 614135<br>Stickler syndrome, type IV, 614134 |
| COL9A2 | 104.9 | 100.0% | 99.6% | ?Stickler syndrome, type V, 614284<br>Epiphyseal dysplasia, multiple, 2, 600204 |
| COL9A3 | 120.1 | 99.9% | 98.3% | Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 |
| CTSK | 90.0 | 100.0% | 100.0% | Pycnodysostosis, 265800 |
| DHODH | 107.2 | 100.0% | 100.0% | Miller syndrome, 263750 |
| DISP1 | 170.7 | 100.0% | 99.7% | No OMIM Disease ID |
| DLX3 | 165.9 | 100.0% | 99.8% | Trichodontoosseous syndrome, 190320<br>Amelogenesis imperfecta, type IV, 104510 |
| DLX4 | 253.0 | 100.0% | 100.0% | ?Orofacial cleft 15, 616788 |
| DSPP | 83.9 | 98.8% | 95.5% | Dentin dysplasia, type II, 125420<br>Deafness, autosomal dominant 39, with dentinogenesis, 605594<br>Dentinogenesis imperfecta, Shields type II, 125490<br>Dentinogenesis imperfecta, Shields type III, 125500 |

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| EDA | 108.8 | 96.6% | 88.1% | Tooth agenesis, selective, X-linked 1, 313500<br>Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 |
| EDAR | 135.7 | 100.0% | 100.0% | Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490<br>Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 |
| EDARADD | 93.4 | 99.8% | 97.5% | Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941<br>Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 |
| EDN1 | 165.0 | 100.0% | 99.9% | Auriculocondylar syndrome 3, 615706<br>Question mark ears, isolated, 612798 |
| EDNRA | 153.6 | 100.0% | 99.9% | Mandibulofacial dysostosis with alopecia, 616367 |
| EFNA4 | 166.5 | 100.0% | 100.0% | No OMIM disease ID |
| EFNB1 | 122.4 | 100.0% | 100.0% | Craniofrontonasal dysplasia, 304110 |
| EFTUD2 | 107.1 | 100.0% | 99.5% | Mandibulofacial dysostosis, Guion-Almeida type, 610536 |
| EIF4A3 | 89.6 | 100.0% | 98.9% | Robin sequence with cleft mandible and limb anomalies, 268305 |
| ENAM | 144.6 | 100.0% | 100.0% | Amelogenesis imperfecta, type IC, 204650<br>Amelogenesis imperfecta, type IB, 104500 |
| ERF | 166.3 | 100.0% | 99.6% | Craniosynostosis 4, 600775<br>Chitayat syndrome, 617180 |
| EYA1 | 121.6 | 100.0% | 99.9% | ?Otofaciocervical syndrome, 166780<br>Anterior segment anomalies with or without cataract, 602588<br>Branchiootorenal syndrome 1, with or without cataracts, 113650<br>Branchiootic syndrome 1, 602588 |
| EZH2 | 130.3 | 99.5% | 98.0% | Weaver syndrome, 277590 |
| FAM20A | 122.6 | 99.9% | 99.2% | Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 |
| FAM83H | 139.0 | 100.0% | 100.0% | Amelogenesis imperfecta, type IIIA, 130900 |
| FGD1 | 93.2 | 98.7% | 94.4% | Mental retardation, X-linked syndromic 16, 305400<br>Aarskog-Scott syndrome, 305400 |
| FGF10 | 118.7 | 100.0% | 99.7% | Aplasia of lacrimal and salivary glands, 180920<br>LADD syndrome, 149730 |
| FGF3 | 160.1 | 100.0% | 100.0% | Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 |
| FGF8 | 141.1 | 98.8% | 90.5% | Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 |
| FGF9 | 156.9 | 100.0% | 100.0% | Multiple synostoses syndrome 3, 612961 |
| FGFR1 | 131.6 | 100.0% | 99.7% | Pfeiffer syndrome, 101600<br>Jackson-Weiss syndrome, 123150<br>Trigonocephaly 1, 190440<br>Hypogonadotropic hypogonadism 2 with or without anosmia, 147950<br>Hartsfield syndrome, 615465<br>Osteoglophonic dysplasia, 166250<br>Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 |

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| FGFR2 | 118.0 | 97.7% | 97.1% | Apert syndrome, 101200<br>Jackson-Weiss syndrome, 123150<br>Saethre-Chotzen syndrome, 101400<br>Gastric cancer, somatic, 613659<br>Scaphocephaly, maxillary retrusion, and mental retardation, 609579<br>Bent bone dysplasia syndrome, 614592<br>LADD syndrome, 149730<br>Craniofacial-skeletal-dermatologic dysplasia, 101600<br>Pfeiffer syndrome, 101600<br>Crouzon syndrome, 123500<br>Beare-Stevenson cutis gyrata syndrome, 123790<br>Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410<br>Craniosynostosis, nonspecific, 0<br>Scaphocephaly and Axenfeld-Rieger anomaly, 0 |
| FGFR3 | 157.1 | 100.0% | 99.9% | Muenke syndrome, 602849<br>Nevus, epidermal, somatic, 162900<br>Thanatophoric dysplasia, type II, 187601<br>Bladder cancer, somatic, 109800<br>CATSHL syndrome, 610474<br>Crouzon syndrome with acanthosis nigricans, 612247<br>Hypochondroplasia, 146000<br>LADD syndrome, 149730<br>Achondroplasia, 100800<br>Thanatophoric dysplasia, type I, 187600<br>Colorectal cancer, somatic, 114500<br>Spermatocytic seminoma, somatic, 273300<br>Cervical cancer, somatic, 603956<br>SADDAN, 616482 |
| FOXC1 | 96.1 | 99.9% | 99.2% | Axenfeld-Rieger syndrome, type 3, 602482<br>Anterior segment dysgenesis 3, multiple subtypes, 601631 |
| FOXE1 | 110.9 | 100.0% | 100.0% | Bamforth-Lazarus syndrome, 241850 |
| GDF3 | 132.5 | 100.0% | 100.0% | Microphthalmia, isolated 7, 613704<br>Microphthalmia with coloboma 6, 613703<br>Klippel-Feil syndrome 3, autosomal dominant, 613702 |
| GDF6 | 180.6 | 100.0% | 100.0% | Leber congenital amaurosis 17, 615360<br>Klippel-Feil syndrome 1, autosomal dominant, 118100<br>Multiple synostoses syndrome 4, 617898 |

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| | | | | Microphthalmia with coloboma 6, digenic, 613703<br>Microphthalmia, isolated 4, 613094 |
| GJA1 | 162.4 | 100.0% | 100.0% | Erythrokeratodermia variabilis et progressiva 3, 617525<br>Craniometaphyseal dysplasia, autosomal recessive, 218400<br>Atrioventricular septal defect 3, 600309<br>Oculodentodigital dysplasia, 164200<br>Syndactyly, type III, 186100<br>Oculodentodigital dysplasia, autosomal recessive, 257850<br>Hypoplastic left heart syndrome 1, 241550<br>Palmoplantar keratoderma with congenital alopecia, 104100 |
| GJB6 | 146.2 | 100.0% | 100.0% | Deafness, autosomal dominant 3B, 612643<br>Deafness, autosomal recessive 1B, 612645<br>Deafness, digenic GJB2/GJB6, 220290<br>Ectodermal dysplasia 2, Clouston type, 129500 |
| GLI2 | 177.4 | 100.0% | 100.0% | Culler-Jones syndrome, 615849<br>Holoprosencephaly 9, 610829 |
| GLI3 | 151.8 | 100.0% | 99.5% | Polydactyly, postaxial, types A1 and B, 174200<br>Greig cephalopolysyndactyly syndrome, 175700<br>Polydactyly, preaxial, type IV, 174700<br>Pallister-Hall syndrome, 146510 |
| GNAI3 | 88.5 | 98.9% | 93.5% | Auriculocondylar syndrome 1, 602483 |
| GPR68 | 184.6 | 100.0% | 99.6% | Amelogenesis imperfecta, hypomaturation type, IIA6, 617217 |
| GRHL3 | 141.9 | 100.0% | 99.9% | Van der Woude syndrome 2, 606713 |
| GSC | 149.2 | 100.0% | 99.6% | Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471 |
| HOXA2 | 90.9 | 100.0% | 100.0% | Microtia with or without hearing impairment (AD), 612290<br>?Microtia, hearing impairment, and cleft palate (AR), 612290 |
| HUWE1 | 82.1 | 99.1% | 95.0% | Mental retardation, X-linked syndromic, Turner type, 309590 |
| HYAL2 | 192.4 | 100.0% | 99.9% | No OMIM Disease ID |
| IFT122 | 126.6 | 100.0% | 99.6% | Cranioectodermal dysplasia 1, 218330 |
| IFT43 | 119.5 | 100.0% | 100.0% | ?Cranioectodermal dysplasia 3, 614099<br>Short-rib thoracic dysplasia 18 with polydactyly, 617866<br>?Retinitis pigmentosa 81, 617871 |
| IFT88 | 91.0 | 99.9% | 97.0% | No OMIM Disease ID |
| IKBKG | 64.7 | 90.1% | 80.2% | Immunodeficiency 33, 300636<br>Incontinentia pigmenti, 308300<br>Immunodeficiency, isolated, 300584<br>Ectodermal dysplasia and immunodeficiency 1, 300291 |

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| | | | | Invasive pneumococcal disease, recurrent isolated, 2, 300640<br>Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 |
| IL11RA | 141.0 | 100.0% | 99.8% | Craniosynostosis and dental anomalies, 614188 |
| INTU | 112.9 | 99.8% | 98.0% | ?Short-rib thoracic dysplasia 20 with polydactyly, 617925<br>?Orofaciodigital syndrome XVII, 617926 |
| IRF6 | 94.5 | 99.4% | 95.8% | Popliteal pterygium syndrome 1, 119500<br>van der Woude syndrome, 119300 |
| ITGB6 | 129.5 | 96.3% | 95.1% | Amelogenesis imperfecta, type IH, 616221 |
| KAT6B | 162.3 | 99.8% | 99.2% | SBBYSS syndrome, 603736<br>Genitopatellar syndrome, 606170 |
| KDF1 | 120.4 | 100.0% | 100.0% | ?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337 |
| KDM1A | 134.2 | 100.0% | 99.4% | Cleft palate, psychomotor retardation, and distinctive facial features, 616728 |
| KDM6A | 98.1 | 95.6% | 87.7% | Kabuki syndrome 2, 300867 |
| KLK4 | 176.5 | 100.0% | 99.8% | Amelogenesis imperfecta, type IIA1, 204700 |
| KMT2D | 150.7 | 100.0% | 99.9% | Kabuki syndrome 1, 147920 |
| KREMEN1 | 152.0 | 99.8% | 98.7% | Ectodermal dysplasia 13, hair/tooth type, 617392 |
| LAMB3 | 125.6 | 100.0% | 99.5% | Amelogenesis imperfecta, type IA, 104530<br>Epidermolysis bullosa, junctional, Herlitz type, 226700<br>Epidermolysis bullosa, junctional, non-Herlitz type, 226650 |
| LRP2 | 140.5 | 100.0% | 99.9% | Donnai-Barrow syndrome, 222448 |
| LRP6 | 137.9 | 99.9% | 99.3% | Tooth agenesis, selective, 7, 616724 |
| LTBP3 | 166.1 | 100.0% | 100.0% | Dental anomalies and short stature, 601216<br>Geleophysic dysplasia 3, 617809 |
| MASP1 | 140.7 | 100.0% | 99.5% | 3MC syndrome 1, 257920 |
| MED12 | 89.4 | 99.6% | 96.5% | Ohdo syndrome, X-linked, 300895<br>Lujan-Fryns syndrome, 309520<br>Opitz-Kaveggia syndrome, 305450 |
| MEGF8 | 158.7 | 100.0% | 99.8% | Carpenter syndrome 2, 614976 |
| MEIS2 | 128.4 | 100.0% | 99.9% | Cleft palate, cardiac defects, and mental retardation, 600987 |
| MEOX1 | 114.4 | 100.0% | 98.8% | Klippel-Feil syndrome 2, 214300 |
| MID1 | 131.5 | 99.9% | 98.4% | Opitz GBBB syndrome, type I, 300000 |
| MITF | 145.6 | 100.0% | 100.0% | COMMAD syndrome, 617306<br>Waardenburg syndrome, type 2A, 193510<br>Waardenburg syndrome/ocular albinism, digenic, 103470<br>Tietz albinism-deafness syndrome, 103500 |
| MMP20 | 91.0 | 99.9% | 97.9% | Amelogenesis imperfecta, type IIA2, 612529 |

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| MSX1 | 164.1 | 100.0% | 99.4% | Orofacial cleft 5, 608874<br>Ectodermal dysplasia 3, Witkop type, 189500<br>Tooth agenesis, selective, 1, with or without orofacial cleft, 106600 |
| MSX2 | 111.9 | 100.0% | 100.0% | Parietal foramina 1, 168500<br>Craniosynostosis 2, 604757<br>Parietal foramina with cleidocranial dysplasia, 168550 |
| NAA10 | 112.8 | 100.0% | 99.4% | Ogden syndrome, 300855<br>?Microphthalmia, syndromic 1, 309800 |
| NECTIN1 | 146.4 | 100.0% | 100.0% | Orofacial cleft 7, 225060<br>Cleft lip/palate-ectodermal dysplasia syndrome, 225060 |
| NFKBIA | 148.5 | 96.7% | 90.7% | Ectodermal dysplasia and immunodeficiency 2, 612132 |
| NIPBL | 123.0 | 98.9% | 96.7% | Cornelia de Lange syndrome 1, 122470 |
| NOG | 270.7 | 100.0% | 100.0% | Tarsal-carpal coalition syndrome, 186570<br>Symphalangism, proximal, 1A, 185800<br>Stapes ankylosis with broad thumbs and toes, 184460<br>Multiple synostoses syndrome 1, 186500<br>Brachydactyly, type B2, 611377 |
| NSD1 | 152.6 | 100.0% | 99.8% | Sotos syndrome 1, 117550 |
| OFD1 | 52.3 | 85.5% | 70.0% | Orofaciodigital syndrome I, 311200<br>?Retinitis pigmentosa 23, 300424<br>Joubert syndrome 10, 300804<br>Simpson-Golabi-Behmel syndrome, type 2, 300209 |
| OTX2 | 135.4 | 100.0% | 99.6% | Microphthalmia, syndromic 5, 610125<br>Pituitary hormone deficiency, combined, 6, 613986<br>Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125 |
| PAX3 | 116.1 | 100.0% | 99.9% | Craniofacial-deafness-hand syndrome, 122880<br>Rhabdomyosarcoma 2, alveolar, 268220<br>Waardenburg syndrome, type 1, 193500<br>Waardenburg syndrome, type 3, 148820 |
| PAX6 | 122.8 | 100.0% | 99.9% | Optic nerve hypoplasia, 165550<br>?Coloboma, ocular, 120200<br>Foveal hypoplasia 1, 136520<br>Aniridia, 106210<br>Keratitis, 148190<br>Cataract with late-onset corneal dystrophy, 106210<br>?Coloboma of optic nerve, 120430<br>?Morning glory disc anomaly, 120430<br>Anterior segment dysgenesis 5, multiple subtypes, 604229 |

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| PAX7 | 147.9 | 100.0% | 100.0% | Myopathy, congenital, progressive, with scoliosis, 618578<br>Rhabdomyosarcoma 2, alveolar, 268220 |
| PAX9 | 262.1 | 99.8% | 99.6% | Tooth agenesis, selective, 3, 604625 |
| PGM1 | 134.8 | 100.0% | 99.8% | Congenital disorder of glycosylation, type It, 614921 |
| PITX2 | 186.2 | 100.0% | 99.6% | Axenfeld-Rieger syndrome, type 1, 180500<br>Ring dermoid of cornea, 180550<br>Anterior segment dysgenesis 4, 137600 |
| PLCB4 | 102.7 | 99.8% | 97.9% | Auriculocondylar syndrome 2, 614669 |
| POLR1C | 103.3 | 99.3% | 95.4% | Treacher Collins syndrome 3, 248390<br>Leukodystrophy, hypomyelinating, 11, 616494 |
| POLR1D | 186.7 | 91.6% | 91.6% | Treacher Collins syndrome 2, 613717 |
| PORCN | 119.6 | 99.9% | 98.9% | Focal dermal hypoplasia, 305600 |
| PTCH1 | 117.3 | 100.0% | 99.2% | Basal cell carcinoma, somatic, 605462<br>Basal cell nevus syndrome, 109400<br>Holoprosencephaly 7, 610828 |
| PTH1R | 121.2 | 100.0% | 99.5% | Metaphyseal chondrodysplasia, Murk Jansen type, 156400<br>Failure of tooth eruption, primary, 125350<br>Eiken syndrome, 600002<br>Chondrodysplasia, Blomstrand type, 215045 |
| RAB23 | 102.4 | 100.0% | 99.8% | Carpenter syndrome, 201000 |
| RAD21 | 80.8 | 98.0% | 93.5% | ?Mungan syndrome, 611376<br>Cornelia de Lange syndrome 4, 614701 |
| RBM10 | 122.0 | 99.9% | 98.5% | TARP syndrome, 311900 |
| RECQL4 | 181.4 | 100.0% | 100.0% | RAPADILINO syndrome, 266280<br>Baller-Gerold syndrome, 218600<br>Rothmund-Thomson syndrome, type 2,, 268400 |
| RIPK4 | 189.1 | 100.0% | 100.0% | Popliteal pterygium syndrome, Bartsocas-Papas type, 263650<br>CHAND syndrome, 214350 |
| RUNX2 | 109.5 | 73.6% | 72.3% | Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600<br>Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510<br>Cleidocranial dysplasia, 119600<br>Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 |
| SALL1 | 127.8 | 99.9% | 99.3% | Townes-Brocks syndrome 1, 107480<br>Townes-Brocks branchiootorenal-like syndrome, 107480 |
| SALL4 | 147.5 | 100.0% | 98.7% | Duane-radial ray syndrome, 607323<br>IVIC syndrome, 147750 |
| SATB2 | 115.6 | 99.9% | 98.3% | Glass syndrome, 612313 |
| SEMA3E | 131.0 | 100.0% | 99.7% | ?CHARGE syndrome, 214800 |

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| SF3B4 | 82.6 | 99.9% | 98.5% | Acrofacial dysostosis 1, Nager type, 154400 |
| SH3BP2 | 154.6 | 92.4% | 91.4% | Cherubism, 118400 |
| SHH | 165.7 | 100.0% | 100.0% | Schizencephaly, 269160<br>Microphthalmia with coloboma 5, 611638<br>Single median maxillary central incisor, 147250<br>Holoprosencephaly 3, 142945 |
| SIX1 | 150.4 | 99.9% | 99.2% | Deafness, autosomal dominant 23, 605192<br>Branchiootic syndrome 3, 608389 |
| SIX3 | 240.1 | 100.0% | 100.0% | Holoprosencephaly 2, 157170<br>Schizencephaly, 269160 |
| SKI | 149.5 | 100.0% | 99.7% | Shprintzen-Goldberg syndrome, 182212 |
| SLC24A4 | 109.6 | 100.0% | 99.9% | Amelogenesis imperfecta, type IIA5, 615887 |
| SLC26A2 | 203.7 | 100.0% | 99.9% | De la Chapelle dysplasia, 256050<br>Atelosteogenesis, type II, 256050<br>Diastrophic dysplasia, broad bone-platyspondylic variant, 222600<br>Diastrophic dysplasia, 222600<br>Achondrogenesis Ib, 600972<br>Epiphyseal dysplasia, multiple, 4, 226900 |
| SMAD6 | 209.5 | 99.9% | 95.6% | Aortic valve disease 2, 614823 |
| SMC1A | 93.2 | 99.9% | 98.3% | Cornelia de Lange syndrome 2, 300590 |
| SMC3 | 82.8 | 96.4% | 89.7% | Cornelia de Lange syndrome 3, 610759 |
| SMO | 154.2 | 100.0% | 99.5% | Curry-Jones syndrome, somatic mosaic, 601707<br>Basal cell carcinoma, somatic, 605462 |
| SMOC2 | 93.7 | 76.9% | 75.9% | Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400 |
| SNAI2 | 106.3 | 100.0% | 99.1% | Waardenburg syndrome, type 2D, 608890<br>Piebaldism, 172800 |
| SOX10 | 101.7 | 100.0% | 99.8% | Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584<br>PCWH syndrome, 609136<br>Waardenburg syndrome, type 4C, 613266 |
| SOX6 | 95.3 | 99.8% | 98.3% | No OMIM Disease ID |
| SOX9 | 181.5 | 100.0% | 100.0% | Acampomelic campomelic dysplasia, 114290<br>Campomelic dysplasia, 114290<br>Campomelic dysplasia with autosomal sex reversal, 114290 |
| SPECC1L | 133.2 | 100.0% | 99.7% | Hypertelorism, Teebi type, 145420<br>?Facial clefting, oblique, 1, 600251<br>Opitz GBBB syndrome, type II, 145410 |
| SUMO1 | 18.5 | 61.9% | 40.9% | ?Orofacial cleft 10, 613705 |

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| TBX1 | 114.2 | 93.7% | 88.3% | Velocardiofacial syndrome, 192430<br>DiGeorge syndrome, 188400<br>Tetralogy of Fallot, 187500<br>Conotruncal anomaly face syndrome, 217095 |
| TBX22 | 104.3 | 99.0% | 94.5% | ?Abruzzo-Erickson syndrome, 302905<br>Cleft palate with ankyloglossia, 303400 |
| TCF12 | 137.8 | 100.0% | 99.9% | Craniosynostosis 3, 615314 |
| TCOF1 | 119.1 | 99.9% | 99.3% | Treacher Collins syndrome 1, 154500 |
| TFAP2A | 124.1 | 100.0% | 99.3% | Branchiooculofacial syndrome, 113620 |
| TGFBR1 | 156.6 | 97.3% | 94.3% | Loeys-Dietz syndrome 1, 609192 |
| TGFBR2 | 169.1 | 100.0% | 100.0% | Esophageal cancer, somatic, 133239<br>Colorectal cancer, hereditary nonpolyposis, type 6, 614331<br>Loeys-Dietz syndrome 2, 610168 |
| TGIF1 | 150.3 | 100.0% | 100.0% | Holoprosencephaly 4, 142946 |
| TP63 | 169.9 | 100.0% | 100.0% | Limb-mammary syndrome, 603543<br>Orofacial cleft 8, 618149<br>Split-hand/foot malformation 4, 605289<br>Hay-Wells syndrome, 106260<br>Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292<br>Rapp-Hodgkin syndrome, 129400<br>ADULT syndrome, 103285 |
| TRAF6 | 77.9 | 95.4% | 85.4% | No OMIM Disease ID |
| TSHZ1 | 160.9 | 98.8% | 98.7% | Aural atresia, congenital, 607842 |
| TSPEAR | 151.2 | 100.0% | 99.9% | Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180<br>?Deafness, autosomal recessive 98, 614861 |
| TWIST1 | 185.6 | 100.0% | 100.0% | Robinow-Sorauf syndrome, 180750<br>Craniosynostosis 1, 123100<br>Sweeney-Cox syndrome, 617746<br>Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 |
| UBB | 41.7 | 100.0% | 97.1% | No OMIM Disease ID |
| VAX1 | 108.2 | 100.0% | 99.4% | ?Microphthalmia, syndromic 11, 614402 |
| WDR19 | 125.3 | 100.0% | 99.4% | Nephronophthisis 13, 614377<br>Senior-Loken syndrome 8, 616307<br>?Cranioectodermal dysplasia 4, 614378<br>?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 |
| WDR35 | 137.8 | 99.5% | 98.3% | Short-rib thoracic dysplasia 7 with or without polydactyly, 614091<br>Cranioectodermal dysplasia 2, 613610 |
| WDR72 | 123.2 | 96.9% | 96.1% | Amelogenesis imperfecta, type IIA3, 613211 |

| | | | | |
|---|---|---|---|---|
| WNT10A | 159.7 | 100.0% | 100.0% | Schopf-Schulz-Passarge syndrome, 224750<br>Tooth agenesis, selective, 4, 150400<br>Odontoonychodermal dysplasia, 257980 |
| WNT10B | 176.5 | 100.0% | 100.0% | Split-hand/foot malformation 6, 225300<br>Tooth agenesis, selective, 8, 617073 |
| ZEB2 | 145.0 | 99.7% | 98.6% | Mowat-Wilson syndrome, 235730 |
| ZIC1 | 319.7 | 100.0% | 100.0% | Craniosynostosis 6, 616602 |
| ZIC2 | 190.6 | 98.4% | 96.3% | 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.*
*Median Coverage describes the average number of reads seen across 50 exomes.*
*% 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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.*
*OMIM release used for OMIM disease identifiers and descriptions : December 11$^{th}$ , 2019.*
*This list is accurate for panel version DG 2.17*
*Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*