CRANIOFACIAL ANOMALIES GENE PANEL DG 2.14 (151 genes)

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ADAMTSL4	90.6	99.9	98.8	Ectopia lentis et pupillae, 225200 Ectopia lentis, isolated, autosomal recessive, 225100
ALX1	153.2	99.9	98.4	?Frontonasal dysplasia 3, 613456
ALX3	102.7	73.3	70.9	Frontonasal dysplasia 1, 136760
ALX4	132.7	98.4	92.5	Frontonasal dysplasia 2, 613451 Parietal foramina 2, 609597 {Craniosynostosis 5, susceptibility to}, 615529
AMELX	98.2	99	95	Amelogenesis imperfecta, type 1E, 301200
ANKRD11	96.3	97.4	94.1	KBG syndrome, 148050
ARHGAP29	136.4	98.9	95	No OMIM phenotype Cleft lip with or without cleft palate (Leslie (2015) Am J Hum Genet 96,397)
AXIN2	114.5	99.7	98.9	Colorectal cancer, somatic, 114500 Oligodontia-colorectal cancer syndrome, 608615
BCOR	109.7	99.3	96.8	Microphthalmia, syndromic 2, 300166
BMP2	173.4	100	99.9	Brachydactyly, type A2, 112600 Short stature, facial dysmorphism, and skeletal anomalies with or without cardiac anomalies, 617877 {HFE hemochromatosis, modifier of}, 235200
BMP4	151.7	100	99.9	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
C4orf26	197.6	100	100	Amelogenesis imperfecta, type IIA4, 614832
CDC45	160.7	99.4	97.5	Meier-Gorlin syndrome 7, 617063
CDON	143.8	100	99.6	Holoprosencephaly 11, 614226
CDSN	119.3	100	99.5	Hypotrichosis 2, 146520 Peeling skin syndrome 1, 270300
CHD7	150.7	99.9	98.9	CHARGE syndrome, 214800 Hypogonadotropic hypogonadism 5 with or without anosmia, 612370
COL11A1	90.8	94.9	89.6	Fibrochondrogenesis 1, 228520 Marshall syndrome, 154780

				Stickler syndrome, type II, 604841 {Lumbar disc herniation, susceptibility to}, 603932
COL11A2	92.2	99.9	98.3	Deafness, autosomal dominant 13, 601868 Deafness, autosomal recessive 53, 609706 Fibrochondrogenesis 2, 614524 Otospondylomegaepiphyseal dysplasia, autosomal dominant, 184840 Otospondylomegaepiphyseal dysplasia, autosomal recessive, 215150
COL2A1	103.4	99.9	99	Achondrogenesis, type II or hypochondrogenesis, 200610 Avascular necrosis of the femoral head, 608805 Czech dysplasia, 609162 Epiphyseal dysplasia, multiple, with myopia and deafness, 132450 Kniest dysplasia, 156550 Legg-Calve-Perthes disease, 150600 Osteoarthritis with mild chondrodysplasia, 604864 Platyspondylic skeletal dysplasia, Torrance type, 151210 SED congenita, 183900 SMED Strudwick type, 184250 Spondyloepiphyseal dysplasia, Stanescu type, 616583 Spondyloperipheral dysplasia, 271700 Stickler sydrome, type I, nonsyndromic ocular, 609508 Stickler syndrome, type I, 108300 Vitreoretinopathy with phalangeal epiphyseal dysplasia, 0
COL9A1	121.2	99.5	96.9	?Epiphyseal dysplasia, multiple, 6, 614135 Stickler syndrome, type IV, 614134
COL9A2	65.1	98.3	88.8	?Stickler syndrome, type V, 614284 Epiphyseal dysplasia, multiple, 2, 600204
COL9A3	65.5	95.6	86.7	Epiphyseal dysplasia, multiple, 3, with or without myopathy, 600969 {Intervertebral disc disease, susceptibility to}, 603932
CTSK	105	100	99.9	Pycnodysostosis, 265800
DHODH	92.2	100	99.9	Miller syndrome, 263750
DISP1	211.4	99.9	99.1	No OMIM phenotype Craniofacial and neuro-developmental abnormalities (Roessler (2009) Hum Genet 125,393) Diaphragmatic hernia, congenital (Kantarci (2010) Am J Med Genet A 152A,2493) Tetralogy of Fallot (Silversides (2012) PLoS Genet 8, e1002843)

DLX3	109.8	100	99.1	Amelogenesis imperfecta, type IV, 104510 Trichodontoosseous syndrome, 190320
DLX4	162.4	100	100	?Orofacial cleft 15, 616788
DSPP	155.7	99.9	99.3	Deafness, autosomal dominant 39, with dentinogenesis, 605594 Dentin dysplasia, type II, 125420 Dentinogenesis imperfecta, Shields type II, 125490 Dentinogenesis imperfecta, Shields type III, 125500
EDA	88.5	85.7	77.3	Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 Tooth agenesis, selective, X-linked 1, 313500
EDAR	138.6	100	99.6	Ectodermal dysplasia 10A, hypohidrotic/hair/nail type, autosomal dominant, 129490 Ectodermal dysplasia 10B, hypohidrotic/hair/tooth type, autosomal recessive, 224900 [Hair morphology 1, hair thickness], 612630
EDARADD	99.1	99.3	93.3	Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941
EDN1	145.5	100	100	Auriculocondylar syndrome 3, 615706 Question mark ears, isolated, 612798 {High density lipoprotein cholesterol level QTL 7}, 0
EDNRA	218.9	100	99.7	Mandibulofacial dysostosis with alopecia, 616367 {Migraine, resistance to}, 157300
EFNA4	106.6	100	100	No OMIM phenotype Craniosynostosis 1 (Merrill et al. (2006) Hum Molec Genet 15)
EFNB1	118.5	100	99.9	Craniofrontonasal dysplasia, 304110
EFTUD2	124.2	100	99.4	Mandibulofacial dysostosis, Guion-Almeida type, 610536
EIF4A3	106.8	100	99.9	Robin sequence with cleft mandible and limb anomalies, 268305
ENAM	148.9	100	99.9	Amelogenesis imperfecta, type IB, 104500 Amelogenesis imperfecta, type IC, 204650
ERF	107.9	99.9	97.7	Chitayat syndrome, 617180 Craniosynostosis 4, 600775
EYA1	144.2	100	99.7	?Otofaciocervical syndrome, 166780 Anterior segment anomalies with or without cataract, 602588 Branchiootic syndrome 1, 602588 Branchiootorenal syndrome 1, with or without cataracts, 113650
EZH2	139.5	99.8	97.6	Weaver syndrome, 277590
FAM83H	76.7	94.9	87.7	Amelogenesis imperfecta, type IIIA, 130900

FGD1	85.7	92.7	86.5	Aarskog-Scott syndrome, 305400 Mental retardation, X-linked syndromic 16, 305400
FGF10	142.2	100	100	Aplasia of lacrimal and salivary glands, 180920 LADD syndrome, 149730
FGF3	73.9	92	75.7	Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706
FGF8	111.4	90.2	79.7	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	148	99.7	98.3	Encephalocraniocutaneous lipomatosis, 613001 Hartsfield syndrome, 615465 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Jackson-Weiss syndrome, 123150 Osteoglophonic dysplasia, 166250 Pfeiffer syndrome, 101600 Trigonocephaly 1, 190440
FGFR2	140.1	97.4	96.4	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific, 0 Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, 0 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FGFR3	110.2	99.6	97	Achondroplasia, 100800 Bladder cancer, somatic, 109800 CATSHL syndrome, 610474 Cervical cancer, somatic, 603956 Colorectal cancer, somatic, 114500 Crouzon syndrome with acanthosis nigricans, 612247 Hypochondroplasia, 146000

				LADD syndrome, 149730 Muenke syndrome, 602849 Nevus, epidermal, somatic, 162900 SADDAN, 616482 Spermatocytic seminoma, somatic, 273300 Thanatophoric dysplasia, type I, 187600 Thanatophoric dysplasia, type II, 187601
FOXC1	32.7	86	68.5	Anterior segment dysgenesis 3, multiple subtypes, 601631 Axenfeld-Rieger syndrome, type 3, 602482
FOXE1	29.3	72.3	56.2	Bamforth-Lazarus syndrome, 241850 {Thyroid cancer, nonmedullary, 4}, 616534
GDF3	134.9	100	100	Klippel-Feil syndrome 3, autosomal dominant, 613702 Microphthalmia with coloboma 6, 613703 Microphthalmia, isolated 7, 613704
GDF6	75.2	98.7	89	Klippel-Feil syndrome 1, autosomal dominant, 118100 Leber congenital amaurosis 17, 615360 Microphthalmia with coloboma 6, digenic, 613703 Microphthalmia, isolated 4, 613094 Multiple synostoses syndrome 4, 617898
GJA1	246.4	100	100	Atrioventricular septal defect 3, 600309 Craniometaphyseal dysplasia, autosomal recessive, 218400 Erythrokeratodermia variabilis et progressiva 3, 617525 Hypoplastic left heart syndrome 1, 241550 Oculodentodigital dysplasia, 164200 Oculodentodigital dysplasia, autosomal recessive, 257850 Palmoplantar keratoderma with congenital alopecia, 104100 Syndactyly, type III, 186100
GJB6	185.4	100	100	Deafness, autosomal dominant 3B, 612643 Deafness, autosomal recessive 1B, 612645 Deafness, digenic GJB2/GJB6, 220290 Ectodermal dysplasia 2, Clouston type, 129500
GLI2	138.5	99.4	97.4	Culler-Jones syndrome, 615849 Holoprosencephaly 9, 610829
GLI3	154.2	100	99.7	Greig cephalopolysyndactyly syndrome, 175700 Pallister-Hall syndrome, 146510

				Polydactyly, postaxial, types A1 and B, 174200 Polydactyly, preaxial, type IV, 174700 {Hypothalamic hamartomas, somatic}, 241800
GNAI3	110.6	99	92	Auriculocondylar syndrome 1, 602483
GRHL3	140.7	100	99.9	Van der Woude syndrome 2, 606713
GSC	85.4	86.9	74.5	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities, 602471
HOXA2	74.1	99.5	95.6	?Microtia, hearing impairment, and cleft palate (AR), 612290 Microtia with or without hearing impairment (AD), 612290
HUWE1	98.4	99.2	97	Mental retardation, X-linked syndromic, Turner type, 300706
HYAL2	223.2	100	100	No OMIM phenotype Orofacial clefting (Muggenthaler (2017) PLoS Genet 13,e1006470) ?Hypertelorism and high myopia (Shaheen (2016) Genet Med 18,686)
IFT122	152	100	99.9	Cranioectodermal dysplasia 1, 218330
IFT43	114.8	100	100	?Cranioectodermal dysplasia 3, 614099 ?Retinitis pigmentosa 81, 617871 Short-rib thoracic dysplasia 18 with polydactyly, 617866
IFT88	78.1	98.1	90.7	No OMIM phenotype ?Cleft lip and palate (Tian (2017) Hum Mol Genet 26,860)
IKBKG	52.5	84.6	73.2	Ectodermal dysplasia, hypohidrotic, with immune deficiency, 300291 Ectodermal, dysplasia, anhidrotic, lymphedema and immunodeficiency, 300301 Immunodeficiency 33, 300636 Immunodeficiency, isolated, 300584 Incontinentia pigmenti, 308300 Invasive pneumococcal disease, recurrent isolated, 2, 300640
IL11RA	139.9	100	99.5	Craniosynostosis and dental anomalies, 614188
INTU	122	99.7	96.6	?Orofaciodigital syndrome XVII, 617926 ?Short-rib throacic dysplasia 20 with polydactyly, 617925
IRF6	113.7	99.9	97.9	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300 {Orofacial cleft 6}, 608864
KAT6B	192.3	99.6	98.5	Genitopatellar syndrome, 606170 SBBYSS syndrome, 603736
KDM1A	129.7	96.2	93.4	Cleft palate, psychomotor retardation, and distinctive facial features, 616728

KDM6A	109	93.2	84.3	Kabuki syndrome 2, 300867
KLK4	185.1	100	98.8	Amelogenesis imperfecta, type IIA1, 204700
KMT2D	142.1	99.9	99	Kabuki syndrome 1, 147920
KREMEN1	149.1	94.7	93.6	Ectodermal dysplasia 13, hair/tooth type, 617392
LRP2	176.3	100	99.8	Donnai-Barrow syndrome, 222448
LRP6	169.3	100	99.7	Tooth agenesis, selective, 7, 616724 {Coronary artery disease, autosomal dominant, 2}, 610947
LTBP3	113.5	98.7	94.7	Dental anomalies and short stature, 601216 Geleophysic dysplasia 3, 617809
MASP1	148.6	100	99.6	3MC syndrome 1, 257920
MED12	105.7	98	94.8	Lujan-Fryns syndrome, 309520 Ohdo syndrome, X-linked, 300895 Opitz-Kaveggia syndrome, 305450
MEGF8	127.6	99.9	98.6	Carpenter syndrome 2, 614976
MEOX1	76.8	96.6	91.2	Klippel-Feil syndrome 2, 214300
MID1	164.6	99.8	98.4	Opitz GBBB syndrome, type I, 300000
MITF	155.5	100	99.9	COMMAD syndrome, 617306 Tietz albinism-deafness syndrome, 103500 Waardenburg syndrome, type 2A, 193510 Waardenburg syndrome/ocular albinism, digenic, 103470 {Melanoma, cutaneous malignant, susceptibility to, 8}, 614456
MMP20	100.5	100	98.6	Amelogenesis imperfecta, type IIA2, 612529
MSX1	75.2	95.4	87.5	Ectodermal dysplasia 3, Witkop type, 189500 Orofacial cleft 5, 608874 Tooth agenesis, selective, 1, with or without orofacial cleft, 106600
MSX2	94.2	98	85.5	Craniosynostosis 2, 604757 Parietal foramina 1, 168500 Parietal foramina with cleidocranial dysplasia, 168550
NAA10	102.4	98.7	96.7	?Microphthalmia, syndromic 1, 309800 Ogden syndrome, 300855
NECTIN1	145.4	100	100	Cleft lip/palate-ectodermal dysplasia syndrome, 225060 Orofacial cleft 7, 225060
NFKBIA	116.3	98.5	93.8	Ectodermal dysplasia, anhidrotic, with T-cell immunodeficiency, 612132

NIPBL	116.1	96.5	94.5	Cornelia de Lange syndrome 1, 122470
NOG	191.9	100	100	Brachydactyly, type B2, 611377 Multiple synostoses syndrome 1, 186500 Stapes ankylosis with broad thumbs and toes, 184460 Symphalangism, proximal, 1A, 185800 Tarsal-carpal coalition syndrome, 186570
NSD1	155.2	100	99.9	Leukemia, acute myeloid, 601626 Sotos syndrome 1, 117550
OFD1	51.5	84	67.8	?Retinitis pigmentosa 23, 300424 Joubert syndrome 10, 300804 Orofaciodigital syndrome I, 311200 Simpson-Golabi-Behmel syndrome, type 2, 300209
OTX2	154.8	100	99.8	Microphthalmia, syndromic 5, 610125 Pituitary hormone deficiency, combined, 6, 613986 Retinal dystrophy, early-onset, with or without pituitary dysfunction, 610125
PAX3	118.5	100	100	Craniofacial-deafness-hand syndrome, 122880 Rhabdomyosarcoma 2, alveolar, 268220 Waardenburg syndrome, type 1, 193500 Waardenburg syndrome, type 3, 148820
PAX6	119.9	100	99.9	?Coloboma of optic nerve, 120430 ?Coloboma, ocular, 120200 ?Morning glory disc anomaly, 120430 Aniridia, 106210 Anterior segment dysgenesis 5, multiple subtypes, 604229 Cataract with late-onset corneal dystrophy, 106210 Foveal hypoplasia 1, 136520 Keratitis, 148190 Optic nerve hypoplasia, 165550
PAX7	117.8	100	100	Rhabdomyosarcoma 2, alveolar, 268220
PAX9	238.8	99.6	99.3	Tooth agenesis, selective, 3, 604625
PITX2	147.8	99.7	97.5	Anterior segment dysgenesis 4, 137600 Axenfeld-Rieger syndrome, type 1, 180500 Ring dermoid of cornea, 180550
PLCB4	126.1	99.2	95.7	Auriculocondylar syndrome 2, 614669

POLR1C	117	99.7	96.1	Leukodystrophy, hypomyelinating, 11, 616494 Treacher Collins syndrome 3, 248390
POLR1D	176.2	91.6	91.6	Treacher Collins syndrome 2, 613717
PORCN	117.7	100	99.3	Focal dermal hypoplasia, 305600
PTCH1	114.6	98.4	95.9	Basal cell carcinoma, somatic, 605462 Basal cell nevus syndrome, 109400 Holoprosencephaly 7, 610828
PTH1R	108.5	99.9	98.8	Chondrodysplasia, Blomstrand type, 215045 Eiken syndrome, 600002 Failure of tooth eruption, primary, 125350 Metaphyseal chondrodysplasia, Murk Jansen type, 156400
RAB23	110.3	99.7	98	Carpenter syndrome, 201000
RAD21	78.5	98.8	94.7	Cornelia de Lange syndrome 4, 614701
RECQL4	149.6	99.2	96.5	Baller-Gerold syndrome, 218600 RAPADILINO syndrome, 266280 Rothmund-Thomson syndrome, 268400
RIPK4	163.3	100	99.6	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
RUNX2	106.4	72.3	72.2	Cleidocranial dysplasia, 119600 Cleidocranial dysplasia, forme fruste, dental anomalies only, 119600 Cleidocranial dysplasia, forme fruste, with brachydactyly, 119600 Metaphyseal dysplasia with maxillary hypoplasia with or without brachydactyly, 156510
SALL1	138.5	99.3	98.4	Townes-Brocks branchiootorenal-like syndrome, 107480 Townes-Brocks syndrome 1, 107480
SALL4	147.5	97.6	96.3	Duane-radial ray syndrome, 607323 IVIC syndrome, 147750
SATB2	110.5	98.5	93.4	Glass syndrome, 612313
SEMA3E	142.6	99.9	99	?CHARGE syndrome, 214800
SF3B4	89.5	99.8	97.7	Acrofacial dysostosis 1, Nager type, 154400
SH3BP2	110.9	91.4	91.4	Cherubism, 118400
SHH	117.5	99	94	Holoprosencephaly 3, 142945 Microphthalmia with coloboma 5, 611638 Schizencephaly, 269160 Single median maxillary central incisor, 147250

SIX1	117.3	99.7	97.6	Branchiootic syndrome 3, 608389 Deafness, autosomal dominant 23, 605192
SIX3	145.3	100	98.9	Holoprosencephaly 2, 157170 Schizencephaly, 269160
SKI	85.3	96.4	90.8	Shprintzen-Goldberg syndrome, 182212
SMAD6	100.5	80	72	Aortic valve disease 2, 614823 {Craniosynostosis 7, susceptibility to}, 617439
SMC1A	99.4	99.9	98.8	Cornelia de Lange syndrome 2, 300590
SMC3	81.4	93.8	87.6	Cornelia de Lange syndrome 3, 610759
SMOC2	91.5	75.4	72.6	Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400
SNAI2	129.8	100	99.8	Piebaldism, 172800 Waardenburg syndrome, type 2D, 608890
SOX10	65.8	98.2	91.3	PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 Waardenburg syndrome, type 4C, 613266
SOX6	102.7	99.7	97.9	No OMIM phenotype Developmental delay and spinal syrinx (Scott (2014) J Child Neurol 29, NP164) Dystonia, dopa-responsive (Ebrahimi-Fakhari (2015) Pediatr Neurol 52,115) ?Craniosynostosis (Tagariello (2006) J Med Genet 43,534)
SPECC1L	157.9	100	100	?Facial clefting, oblique, 1, 600251 Opitz GBBB syndrome, type II, 145410
SUMO1	17.6	58.8	37	?Orofacial cleft 10, 613705
TBX22	121.8	99.2	96.3	?Abruzzo-Erickson syndrome, 302905 Cleft palate with ankyloglossia, 303400
TCF12	150.3	100	99.8	Craniosynostosis 3, 615314
TCOF1	98.6	99.5	97.3	Treacher Collins syndrome 1, 154500
TFAP2A	109.3	100	99.3	Branchiooculofacial syndrome, 113620
TGFBR1	173.4	93.7	93.6	Loeys-Dietz syndrome 1, 609192 {Multiple self-healing squamous epithelioma, susceptibility to}, 132800
TGFBR2	193.5	100	99.9	Colorectal cancer, hereditary nonpolyposis, type 6, 614331 Esophageal cancer, somatic, 133239 Loeys-Dietz syndrome 2, 610168
TGIF1	138.3	100	100	Holoprosencephaly 4, 142946

TP63	206.3	100	100	ADULT syndrome, 103285 Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 Hay-Wells syndrome, 106260 Limb-mammary syndrome, 603543 Orofacial cleft 8, 129400 Rapp-Hodgkin syndrome, 129400 Split-hand/foot malformation 4, 605289
TRAF6	106.9	96.1	87.1	No OMIM phenotype Ectodermal dysplasia, hypohidrotic (Wisniewski (2012) Br J Dermatol 166,1353)
TSHZ1	166.6	98.8	98.5	Aural atresia, congenital, 607842
TWIST1	134.4	96.6	87.2	Craniosynostosis 1, 123100 Robinow-Sorauf syndrome, 180750 Saethre-Chotzen syndrome with or without eyelid anomalies, 101400 Sweeney-Cox syndrome, 617746
UBB	61.9	100	99.7	Cleft palate, isolated, 119540
VAX1	52.2	88.4	78	?Microphthalmia, syndromic 11, 614402
WDR19	132.1	99.8	98.1	?Cranioectodermal dysplasia 4, 614378 ?Short-rib thoracic dysplasia 5 with or without polydactyly, 614376 Nephronophthisis 13, 614377 Senior-Loken syndrome 8, 616307
WDR35	145.1	99.3	97.7	Cranioectodermal dysplasia 2, 613610 Short-rib thoracic dysplasia 7 with or without polydactyly, 614091
WDR72	132.2	96.5	95.4	Amelogenesis imperfecta, type IIA3, 613211
WNT10A	114	100	99.1	Odontoonychodermal dysplasia, 257980 Schopf-Schulz-Passarge syndrome, 224750 Tooth agenesis, selective, 4, 150400
ZEB2	157	99.8	98.8	Mowat-Wilson syndrome, 235730
ZIC1	231.1	100	100	Craniosynostosis 6, 616602
ZIC2	122.5	90.5	78.9	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 : September 11th, 2018.
This list is accurate for panel version DG 2.14
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors