

SKIN DISORDERS PANEL¹ DG-4.0.0 (646 GENES)

| <i>Gene</i> | <i>Twist X2 covered >10x</i> | <i>Twist X2 covered >20x</i> | <i>WGS covered >10x</i> | <i>WGS covered >20x</i> | <i>Associated Phenotype description and OMIM disease ID</i> |
|-------------|---------------------------------|---------------------------------|----------------------------|----------------------------|--|
| AAAS | 100.0% | 100.0% | 100.0% | 99.3% | Achalasia-addisonianism-alacrimia syndrome, 231550 |
| AAGAB | 100.0% | 100.0% | 100.0% | 97.8% | Keratoderma, palmoplantar, punctate type IA, 148600 |
| ABCA12 | 100.0% | 100.0% | 100.0% | 98.5% | Ichthyosis, congenital, autosomal recessive 4B (harlequin), 242500;Ichthyosis, congenital, autosomal recessive 4A, 601277 |
| ABCB6 | 100.0% | 100.0% | 100.0% | 99.1% | Microphthalmia, isolated, with coloboma 7, 614497;Dyschromatosis universalis hereditaria 3, 615402;[Blood group, Langereis system], 111600;Pseudohyperkalemia, familial, 2, due to red cell leak, 609153 |
| ABCC6 | 98.4% | 98.4% | 100.0% | 99.3% | Pseudoxanthoma elasticum, 264800;Arterial calcification, generalized, of infancy, 2, 614473;Pseudoxanthoma elasticum, forme fruste, 177850 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| ABCC9 | 96.0% | 96.0% | 100.0% | 98.4% | Cardiomyopathy, dilated, 10, 608569;Hypertrichotic osteochondrodysplasia (Cantu syndrome), 239850;?Atrial fibrillation, familial, 12, 614050;Intellectual disability and myopathy syndrome, 619719 |
| ABHD5 | 100.0% | 100.0% | 100.0% | 99.0% | Chanarin-Dorfman syndrome, 275630 |
| ACD | 100.0% | 100.0% | 100.0% | 98.6% | ?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553 |
| ACTA2 | 99.9% | 99.1% | 100.0% | 99.2% | Smooth muscle dysfunction syndrome, 613834;Aortic aneurysm, familial thoracic 6, 611788;Moyamoya disease 5, 614042 |
| ACTB | 100.0% | 100.0% | 100.0% | 99.0% | Baraitser-Winter syndrome 1, 243310;Becker nevus, syndromic or isolated, somatic mosaic, 604919;Thrombocytopenia 8, with dysmorphic features and developmental delay, 620475;Dystonia-deafness syndrome 1, 607371;Congenital smooth muscle hamartoma with or without hemihypertrophy, somatic mosaic, 620479 |

| | | | | | |
|----------|--------|--------|--------|-------|--|
| ACVRL1 | 100.0% | 100.0% | 100.0% | 99.1% | Telangiectasia, hereditary hemorrhagic, type 2, 600376 |
| ADA2 | 93.6% | 93.1% | 100.0% | 99.3% | Sneddon syndrome, 182410; Vasculitis, autoinflammation, immunodeficiency, and hematologic defects syndrome, 615688 |
| ADAM10 | 100.0% | 100.0% | 100.0% | 97.6% | {Alzheimer disease 18, susceptibility to}, 615590; Reticulate acropigmentation of Kitamura, 615537 |
| ADAM17 | 99.2% | 99.2% | 100.0% | 98.7% | ?Inflammatory skin and bowel disease, neonatal, 1, 614328 |
| ADAMTS10 | 100.0% | 100.0% | 100.0% | 99.1% | Weill-Marchesani syndrome 1, recessive, 277600 |
| ADAMTS17 | 100.0% | 100.0% | 100.0% | 97.3% | Weill-Marchesani 4 syndrome, recessive, 613195 |
| ADAMTS2 | 97.9% | 97.9% | 100.0% | 98.5% | Ehlers-Danlos syndrome, dermatosparaxis type, 225410 |
| ADAMTS3 | 99.3% | 98.7% | 100.0% | 99.0% | Hennekam lymphangiectasia-lymphedema syndrome 3, 618154 |
| ADAMTSL2 | 100.0% | 99.7% | 100.0% | 99.5% | Geleophysic dysplasia 1, 231050 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| ADAR | 100.0% | 100.0% | 100.0% | 98.2% | Dyschromatosis symmetrica hereditaria, 127400;Aicardi-Goutieres syndrome 6, 615010 |
| AGA | 100.0% | 100.0% | 100.0% | 98.3% | Aspartylglucosaminuria, 208400 |
| AGPAT2 | 100.0% | 100.0% | 100.0% | 97.9% | Lipodystrophy, congenital generalized, type 1, 608594 |
| AIRE | 100.0% | 100.0% | 100.0% | 99.5% | Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 |
| AKT1 | 100.0% | 100.0% | 100.0% | 99.8% | Breast cancer, somatic, 114480;Cowden syndrome 6, 615109;Colorectal cancer, somatic, 114500;Proteus syndrome, somatic, 176920;Ovarian cancer, somatic, 167000 |
| AKT3 | 94.6% | 94.3% | 100.0% | 97.7% | Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2, 615937 |
| ALAD | 100.0% | 100.0% | 100.0% | 99.6% | Porphyria, acute hepatic, 612740;{Lead poisoning, susceptibility to}, 612740 |
| ALAS2 | 100.0% | 99.8% | 98.3% | 72.9% | Anemia, sideroblastic, 1, 300751;Protoporphyrin, erythropoietic, X-linked, 300752 |

| | | | | | |
|----------|--------|--------|--------|-------|--|
| ALDH18A1 | 100.0% | 100.0% | 100.0% | 99.5% | Spastic paraplegia 9A, autosomal dominant, 601162;Cutis laxa, autosomal recessive, type IIIA, 219150;Spastic paraplegia 9B, autosomal recessive, 616586;Cutis laxa, autosomal dominant 3, 616603 |
| ALDH3A2 | 93.5% | 93.5% | 100.0% | 98.4% | Sjogren-Larsson syndrome, 270200 |
| ALDOB | 100.0% | 100.0% | 100.0% | 99.4% | Fructose intolerance, hereditary, 229600 |
| ALOX12B | 100.0% | 100.0% | 100.0% | 98.5% | Ichthyosis, congenital, autosomal recessive 2, 242100 |
| ALOXE3 | 100.0% | 100.0% | 100.0% | 98.5% | Ichthyosis, congenital, autosomal recessive 3, 606545 |
| ALPL | 100.0% | 100.0% | 100.0% | 99.5% | Odontohypophosphatasia, 146300;Hypophosphatasia, infantile, 241500;Hypophosphatasia, childhood, 241510;Hypophosphatasia, adult, 146300 |
| ALX4 | 100.0% | 100.0% | 100.0% | 97.1% | Parietal foramina 2, 609597;{Craniosynostosis 5, susceptibility to}, 615529;Frontonasal dysplasia 2, 613451 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| AMELX | 100.0% | 100.0% | 97.7% | 67.0% | Amelogenesis imperfecta, type 1E, 301200 |
| ANGPT2 | 100.0% | 100.0% | 100.0% | 98.3% | Lymphatic malformation 10, 619369 |
| ANKRD11 | 100.0% | 100.0% | 100.0% | 98.0% | KBG syndrome, 148050 |
| ANOS1 | 100.0% | 99.8% | 97.6% | 68.8% | Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 |
| ANTXR1 | 100.0% | 99.8% | 99.7% | 94.8% | GAPO syndrome, 230740;{?Hemangioma, capillary infantile, susceptibility to}, 602089 |
| ANTXR2 | 96.3% | 96.3% | 100.0% | 98.1% | Hyaline fibromatosis syndrome, 228600 |
| AP1B1 | 100.0% | 100.0% | 100.0% | 99.3% | Keratitis-ichthyosis-deafness syndrome, autosomal recessive, 242150 |
| AP1S3 | 90.6% | 90.6% | 100.0% | 97.3% | {Psoriasis 15, pustular, susceptibility to}, 616106 |
| AP3B1 | 100.0% | 100.0% | 100.0% | 98.8% | Hermansky-Pudlak syndrome 2, 608233 |

| | | | | | |
|----------|--------|--------|--------|-------|--|
| APC | 100.0% | 100.0% | 100.0% | 98.1% | Colorectal cancer, somatic, 114500;Brain tumor-polyposis syndrome 2, 175100;Desmoid disease, hereditary, 135290;Adenoma, periampullary, somatic, 175100;Hepatoblastoma, somatic, 114550;Gastric cancer, somatic, 613659;Gastric adenocarcinoma and proximal polyposis of the stomach, 619182;Gardner syndrome, 175100;Adenomatous polyposis coli, 175100 |
| APCDD1 | 100.0% | 100.0% | 100.0% | 99.2% | Hypotrichosis 1, 605389 |
| AQP5 | 100.0% | 100.0% | 100.0% | 99.2% | Palmoplantar keratoderma, Bothnian type, 600231 |
| ARHGAP31 | 100.0% | 100.0% | 100.0% | 98.3% | Adams-Oliver syndrome 1, 100300 |
| ARID1A | 100.0% | 100.0% | 99.6% | 93.0% | Coffin-Siris syndrome 2, 614607 |
| ARID1B | 98.6% | 98.4% | 98.0% | 86.3% | Coffin-Siris syndrome 1, 135900 |
| ASIP | 100.0% | 100.0% | 99.9% | 97.4% | [Skin/hair/eye pigmentation 9, brown/nonbrown eyes], 611742;[Skin/hair/eye pigmentation 9, dark/light hair], 611742 |

| | | | | | |
|----------|--------|--------|--------|-------|--|
| ASL | 100.0% | 100.0% | 100.0% | 99.3% | Argininosuccinic aciduria, 207900 |
| ASPRV1 | 100.0% | 100.0% | 99.8% | 98.1% | Ichthyosis, lamellar, autosomal dominant, 146750 |
| ASXL1 | 100.0% | 100.0% | 100.0% | 99.1% | Myelodysplastic syndrome, somatic, 614286;Bohring-Opitz syndrome, 605039 |
| ASXL3 | 100.0% | 100.0% | 100.0% | 97.4% | Bainbridge-Ropers syndrome, 615485 |
| ATIC | 100.0% | 100.0% | 100.0% | 97.8% | AICA-ribosiduria due to ATIC deficiency, 608688 |
| ATP2A2 | 100.0% | 100.0% | 100.0% | 99.1% | Acrokeratosis verruciformis, 101900;Darier disease, 124200 |
| ATP2C1 | 100.0% | 99.9% | 100.0% | 98.2% | Hailey-Hailey disease, 169600 |
| ATP6V0A2 | 100.0% | 100.0% | 100.0% | 97.2% | Wrinkly skin syndrome, 278250;Cutis laxa, autosomal recessive, type IIA, 219200 |
| ATP7A | 94.9% | 94.5% | 98.1% | 71.7% | Occipital horn syndrome, 304150;Neuronopathy, distal hereditary motor, X-linked, 300489;Menkes disease, 309400 |
| ATR | 100.0% | 100.0% | 100.0% | 98.2% | Seckel syndrome 1, 210600;?Cutaneous telangiectasia and cancer syndrome, familial, 614564 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| AXIN2 | 100.0% | 100.0% | 100.0% | 99.1% | Colorectal cancer, somatic, 114500;Oligodontia-colorectal cancer syndrome, 608615 |
| B3GALT6 | 99.9% | 98.0% | 100.0% | 94.8% | Ehlers-Danlos syndrome, spondylodysplastic type, 2, 615349;Spondyloepimetaphyseal dysplasia with joint laxity, type 1, with or without fractures, 271640;Al-Gazali syndrome, 609465 |
| B4GALT7 | 100.0% | 100.0% | 100.0% | 99.0% | Ehlers-Danlos syndrome, spondylodysplastic type, 1, 130070 |
| BANF1 | 100.0% | 100.0% | 100.0% | 97.3% | Nestor-Guillermo progeria syndrome, 614008 |
| BAP1 | 100.0% | 100.0% | 100.0% | 99.3% | Kury-Isidor syndrome, 619762;Tumor predisposition syndrome 1, 614327;{Uveal melanoma, susceptibility to, 2}, 606661 |
| BCOR | 100.0% | 99.8% | 98.4% | 73.6% | Microphthalmia, syndromic 2, 300166 |
| BCS1L | 100.0% | 100.0% | 100.0% | 99.2% | GRACILE syndrome, 603358;Mitochondrial complex III deficiency, nuclear type 1, 124000;Bjornstad syndrome, 262000 |
| BLM | 96.7% | 96.6% | 100.0% | 98.4% | Bloom syndrome, 210900 |
| BLOC1S3 | 100.0% | 100.0% | 100.0% | 95.6% | Hermansky-Pudlak syndrome 8, 614077 |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| BLOC1S6 | 100.0% | 100.0% | 100.0% | 98.2% | Hermansky-Pudlak syndrome 9, 614171 |
| BMS1 | 100.0% | 100.0% | 100.0% | 97.7% | ?Aplasia cutis congenita, nonsyndromic, 107600 |
| BRAF | 100.0% | 100.0% | 99.9% | 96.7% | Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Non-small cell lung cancer, somatic, 211980 |
| BRIP1 | 96.0% | 96.0% | 100.0% | 97.6% | Fanconi anemia, complementation group J, 609054;{Breast cancer, early-onset, susceptibility to}, 114480 |
| BSCL2 | 100.0% | 100.0% | 100.0% | 99.3% | Lipodystrophy, congenital generalized, type 2, 269700;Neuronopathy, distal hereditary motor, autosomal dominant 13, 619112;Silver spastic paraplegia syndrome, 270685;Encephalopathy, progressive, with or without lipodystrophy, 615924 |
| BTD | 94.2% | 94.2% | 100.0% | 99.5% | Biotinidase deficiency, 253260 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| C1QA | 76.2% | 73.5% | 100.0% | 99.2% | C1q deficiency 1, 613652 |
| C1QB | 77.2% | 76.8% | 100.0% | 94.6% | C1q deficiency 2, 620321 |
| C1QC | 99.6% | 97.3% | 100.0% | 97.9% | C1q deficiency 3, 620322 |
| C2CD3 | 96.0% | 96.0% | 100.0% | 98.8% | Orofaciodigital syndrome XIV, 615948 |
| CA2 | 100.0% | 100.0% | 100.0% | 98.8% | Osteopetrosis, autosomal recessive 3, with renal tubular acidosis, 259730 |
| CAPN12 | 100.0% | 100.0% | 99.9% | 94.4% | |
| CARD11 | 100.0% | 100.0% | 100.0% | 99.1% | B-cell expansion with NFKB and T-cell anergy, 616452;Immunodeficiency 11B with atopic dermatitis, 617638;Immunodeficiency 11A, 615206 |
| CARD14 | 100.0% | 100.0% | 100.0% | 99.3% | Psoriasis 2, 602723;Pityriasis rubra pilaris, 173200 |
| CARD9 | 100.0% | 100.0% | 100.0% | 99.9% | Immunodeficiency 103, susceptibility to fungal infection, 212050 |
| CARMIL2 | 100.0% | 100.0% | 100.0% | 98.3% | Immunodeficiency 58, 618131 |
| CARS1 | 100.0% | 100.0% | 100.0% | 99.3% | Microcephaly, developmental delay, and brittle hair syndrome, 618891 |
| CASP14 | 100.0% | 100.0% | 100.0% | 96.9% | Ichthyosis, congenital, autosomal recessive 12, 617320 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| CAST | 100.0% | 100.0% | 99.9% | 98.3% | Peeling skin with leukonychia, acral punctate keratoses, cheilitis, and knuckle pads, 616295 |
| CAV1 | 74.6% | 74.6% | 100.0% | 98.7% | Lipodystrophy, congenital generalized, type 3, 612526;Pulmonary hypertension, primary, 3, 615343;Lipodystrophy, familial partial, type 7, 606721 |
| CAVIN1 | 100.0% | 100.0% | 100.0% | 98.1% | Lipodystrophy, congenital generalized, type 4, 613327 |
| CBL | 100.0% | 100.0% | 100.0% | 98.2% | Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia, 613563;?Juvenile myelomonocytic leukemia, 607785 |
| CBS | 100.0% | 100.0% | 100.0% | 99.5% | Thrombosis, hyperhomocysteinemic, 236200;Homocystinuria, B6-responsive and nonresponsive types, 236200 |
| CCBE1 | 100.0% | 100.0% | 100.0% | 99.3% | Hennekam lymphangiectasia-lymphedema syndrome 1, 235510 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| CD151 | 100.0% | 100.0% | 100.0% | 99.8% | [Blood group, Raph], 179620;Epidermolysis bullosa simplex 7, with nephropathy and deafness, 609057 |
| CDAN1 | 100.0% | 100.0% | 99.9% | 96.7% | Dyserythropoietic anemia, congenital, type Ia, 224120 |
| CDH3 | 100.0% | 100.0% | 100.0% | 98.9% | Hypotrichosis, congenital, with juvenile macular dystrophy, 601553;Ectodermal dysplasia, ectrodactyly, and macular dystrophy, 225280 |
| CDK4 | 100.0% | 100.0% | 100.0% | 99.6% | {Melanoma, cutaneous malignant, 3}, 609048 |
| CDKN2A | 100.0% | 100.0% | 100.0% | 97.4% | {Melanoma and neural system tumor syndrome}, 155755;{Melanoma, cutaneous malignant, 2}, 155601;{Melanoma-pancreatic cancer syndrome}, 606719 |
| CDSN | 100.0% | 100.0% | 100.0% | 99.4% | Hypotrichosis 2, 146520;Peeling skin syndrome 1, 270300 |
| CELSR1 | 100.0% | 100.0% | 100.0% | 98.4% | Lymphatic malformation 9, 619319 |
| CERS3 | 100.0% | 100.0% | 100.0% | 98.1% | Ichthyosis, congenital, autosomal recessive 9, 615023 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| CFTR | 100.0% | 100.0% | 100.0% | 98.6% | Cystic fibrosis, 219700;Congenital bilateral absence of vas deferens, 277180;{Pancreatitis, hereditary}, 167800;{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400;Sweat chloride elevation without CF, ;{Hypertrypsinemia, neonatal}, |
| CHKB | 100.0% | 100.0% | 100.0% | 98.7% | Muscular dystrophy, congenital, megaconial type, 602541 |
| CHST14 | 100.0% | 100.0% | 100.0% | 91.9% | Ehlers-Danlos syndrome, musculocontractural type 1, 601776 |
| CHST8 | 100.0% | 100.0% | 100.0% | 99.5% | |
| CHSY1 | 99.9% | 99.7% | 100.0% | 97.5% | Temtamy preaxial brachydactyly syndrome, 605282 |
| CHUK | 100.0% | 100.0% | 100.0% | 98.5% | ?Popliteal pterygium syndrome, Bartsocas-Papas type 2, 619339;?Cocoon syndrome, 613630 |
| CIB1 | 100.0% | 100.0% | 100.0% | 97.7% | {Epidermodysplasia verruciformis, susceptibility to, 3}, 618267 |
| CKAP2L | 100.0% | 100.0% | 100.0% | 98.4% | Filippi syndrome, 272440 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| CLDN1 | 100.0% | 100.0% | 100.0% | 99.7% | Ichthyosis, leukocyte vacuoles, alopecia, and sclerosing cholangitis, 607626 |
| CLDN10 | 100.0% | 100.0% | 100.0% | 99.2% | HELIX syndrome, 617671 |
| CNNM4 | 100.0% | 100.0% | 100.0% | 97.4% | Jalili syndrome, 217080 |
| COL14A1 | 100.0% | 100.0% | 100.0% | 98.8% | |
| COL17A1 | 100.0% | 100.0% | 100.0% | 98.9% | Epithelial recurrent erosion dystrophy, 122400;Epidermolysis bullosa, junctional 4, intermediate, 619787 |
| COL1A2 | 100.0% | 100.0% | 100.0% | 99.1% | Osteogenesis imperfecta, type III, 259420;{Osteoporosis, postmenopausal}, 166710;Ehlers-Danlos syndrome, arthrochalasia type, 2, 617821;Combined osteogenesis imperfecta and Ehlers-Danlos syndrome 2, 619120;Ehlers-Danlos syndrome, cardiac valvular type, 225320;Osteogenesis imperfecta, type IV, 166220;Osteogenesis imperfecta, type II, 166210 |
| COL3A1 | 100.0% | 100.0% | 100.0% | 98.1% | Ehlers-Danlos syndrome, vascular type, 130050;Polymicrogyria with or without vascular-type EDS, 618343 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| COL5A1 | 100.0% | 100.0% | 100.0% | 99.2% | Ehlers-Danlos syndrome, classic type, 1, 130000;Fibromuscular dysplasia, multifocal, 619329 |
| COL5A2 | 100.0% | 100.0% | 100.0% | 98.4% | Ehlers-Danlos syndrome, classic type, 2, 130010 |
| COL7A1 | 100.0% | 100.0% | 100.0% | 99.3% | Nail disorder, nonsyndromic congenital, 8, 607523;Epidermolysis bullosa dystrophica, Bart type, 132000;Epidermolysis bullosa dystrophica inversa, 226600;Epidermolysis bullosa dystrophica, autosomal recessive, 226600;Epidermolysis bullosa, pretibial, 131850;Epidermolysis bullosa dystrophica, autosomal dominant, 131750;Transient bullous of the newborn, 131705;Epidermolysis bullosa pruriginosa, 604129;Epidermolysis bullosa dystrophica, localisata variant, 226600 |
| COX4I2 | 100.0% | 100.0% | 100.0% | 98.8% | Exocrine pancreatic insufficiency, dyserythropoietic anemia, and calvarial hyperostosis, 612714 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| COX7B | 100.0% | 99.9% | 98.5% | 76.8% | Linear skin defects with multiple congenital anomalies 2, 300887 |
| CPOX | 100.0% | 100.0% | 100.0% | 97.3% | Coproporphyrinuria, 121300;Harderoporphyria, 618892 |
| CST6 | 100.0% | 100.0% | 100.0% | 98.1% | ?Ectodermal dysplasia 15, hypohidrotic/hair type, 618535 |
| CSTA | 100.0% | 100.0% | 100.0% | 97.8% | Peeling skin syndrome 4, 607936 |
| CTC1 | 100.0% | 100.0% | 100.0% | 98.8% | Cerebroretinal microangiopathy with calcifications and cysts, 612199 |
| CTSA | 100.0% | 99.9% | 100.0% | 98.7% | Galactosialidosis, 256540 |
| CTSB | 84.5% | 83.6% | 100.0% | 98.7% | Keratolytic winter erythema, 148370 |
| CTSC | 94.7% | 94.2% | 100.0% | 98.3% | Periodontitis 1, juvenile, 170650;Haim-Munk syndrome, 245010;Papillon-Lefevre syndrome, 245000 |
| CTSZ | 77.7% | 71.8% | 100.0% | 99.2% | |
| CXCR4 | 99.0% | 99.0% | 100.0% | 97.3% | WHIM syndrome 1, 193670;Myelokathexis, isolated, 193670 |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| CYLD | 100.0% | 100.0% | 100.0% | 98.3% | Brooke-Spiegler syndrome, 605041;Cylindromatosis, familial, 132700;Trichoepithelioma, multiple familial, 1, 601606;?Frontotemporal dementia and/or amyotrophic lateral sclerosis 8, 619132 |
| CYP26C1 | 100.0% | 100.0% | 100.0% | 99.2% | Focal facial dermal dysplasia 4, 614974 |
| CYP4F22 | 100.0% | 100.0% | 100.0% | 99.3% | Ichthyosis, congenital, autosomal recessive 5, 604777 |
| DCAF17 | 100.0% | 100.0% | 99.9% | 98.3% | Woodhouse-Sakati syndrome, 241080 |
| DCLRE1C | 97.1% | 97.1% | 100.0% | 98.3% | Severe combined immunodeficiency, Athabaskan type, 602450;Omenn syndrome, 603554 |
| DDB2 | 100.0% | 100.0% | 100.0% | 98.5% | Xeroderma pigmentosum, group E, DDB-negative subtype, 278740 |
| DHCR7 | 96.2% | 96.2% | 100.0% | 99.7% | Smith-Lemli-Opitz syndrome, 270400 |
| DKC1 | 100.0% | 99.9% | 97.9% | 71.5% | ?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| DLX3 | 100.0% | 100.0% | 100.0% | 98.5% | Trichodontoosseous syndrome, 190320;Amelogenesis imperfecta, type IV, 104510 |
| DLX5 | 100.0% | 100.0% | 100.0% | 99.1% | Split-hand/foot malformation 1, 183600;?Split-hand/foot malformation 1 with sensorineural hearing loss, 220600 |
| DOCK6 | 100.0% | 100.0% | 100.0% | 98.7% | Adams-Oliver syndrome 2, 614219 |
| DOCK8 | 98.6% | 98.6% | 100.0% | 98.9% | Hyper-IgE syndrome 2, autosomal recessive, with recurrent infections, 243700 |
| DOLK | 100.0% | 100.0% | 100.0% | 98.1% | Congenital disorder of glycosylation, type Im, 610768 |
| DSC2 | 100.0% | 100.0% | 100.0% | 98.6% | Arrhythmogenic right ventricular dysplasia 11 with mild palmoplantar keratoderma and woolly hair, 610476;Arrhythmogenic right ventricular dysplasia 11, 610476 |
| DSC3 | 100.0% | 100.0% | 100.0% | 98.7% | Hypotrichosis and recurrent skin vesicles, 613102 |
| DSE | 100.0% | 100.0% | 100.0% | 98.8% | Ehlers-Danlos syndrome, musculocontractural type 2, 615539 |

| | | | | | |
|------|--------|--------|--------|-------|--|
| DSG1 | 100.0% | 100.0% | 100.0% | 98.8% | Keratosis palmoplantaris striata I, AD, 148700;Erythroderma, congenital, with palmoplantar keratoderma, hypotrichosis, and hyper IgE, 615508 |
| DSG3 | 100.0% | 100.0% | 100.0% | 98.6% | Blistering, acantholytic, of oral and laryngeal mucosa, 619226 |
| DSG4 | 100.0% | 100.0% | 100.0% | 98.8% | Hypotrichosis 6, 607903 |
| DSP | 100.0% | 100.0% | 100.0% | 98.0% | Arrhythmogenic right ventricular dysplasia 8, 607450;Epidermolysis bullosa, lethal acantholytic, 609638;Keratosis palmoplantaris striata II, 612908;Dilated cardiomyopathy with woolly hair, keratoderma, and tooth agenesis, 615821;Cardiomyopathy, dilated, with woolly hair and keratoderma, 605676 |
| DSPP | 100.0% | 100.0% | 97.9% | 95.2% | Dentinogenesis imperfecta, Shields type III, 125500;Dentinogenesis imperfecta, Shields type II, 125490;Dentin dysplasia, type II, 125420;Deafness, autosomal dominant 39, with dentinogenesis, 605594 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| DST | 100.0% | 100.0% | 100.0% | 98.1% | Neuropathy, hereditary sensory and autonomic, type VI, 614653;Epidermolysis bullosa simplex 3, localized or generalized intermediate, with bp230 deficiency, 615425 |
| DTNBP1 | 100.0% | 100.0% | 99.9% | 97.9% | Hermansky-Pudlak syndrome 7, 614076 |
| DUSP6 | 100.0% | 100.0% | 100.0% | 98.2% | Hypogonadotropic hypogonadism 19 with or without anosmia, 615269 |
| EBP | 100.0% | 100.0% | 98.7% | 72.8% | MEND syndrome, 300960;Chondrodysplasia punctata, X-linked dominant, 302960 |
| ECM1 | 100.0% | 100.0% | 100.0% | 98.6% | Urbach-Wiethe disease, 247100 |
| EDA | 100.0% | 99.6% | 96.2% | 65.6% | Tooth agenesis, selective, X-linked 1, 313500;Ectodermal dysplasia 1, hypohidrotic, X-linked, 305100 |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| EDAR | 100.0% | 100.0% | 100.0% | 98.6% | [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 | 100.0% | 100.0% | 100.0% | 98.5% | Ectodermal dysplasia 11B, hypohidrotic/hair/tooth type, autosomal recessive, 614941;Ectodermal dysplasia 11A, hypohidrotic/hair/tooth type, autosomal dominant, 614940 |
| EDN3 | 100.0% | 100.0% | 100.0% | 99.6% | Waardenburg syndrome, type 4B, 613265;{Hirschsprung disease, susceptibility to, 4}, 613712 |
| EDNRA | 100.0% | 100.0% | 100.0% | 98.4% | {Migraine, resistance to}, 157300;Mandibulofacial dysostosis with alopecia, 616367 |
| EDNRB | 100.0% | 100.0% | 100.0% | 98.1% | {Hirschsprung disease, susceptibility to, 2}, 600155;?ABCD syndrome, 600501;Waardenburg syndrome, type 4A, 277580 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| EFEMP2 | 100.0% | 100.0% | 100.0% | 99.5% | Cutis laxa, autosomal recessive, type IB, 614437 |
| EFNB1 | 100.0% | 99.9% | 98.7% | 73.5% | Craniofrontonasal dysplasia, 304110 |
| EIF2AK3 | 100.0% | 100.0% | 100.0% | 98.2% | Wolcott-Rallison syndrome, 226980 |
| ELN | 100.0% | 100.0% | 100.0% | 98.8% | Cutis laxa, autosomal dominant, 123700;Supravalvar aortic stenosis, 185500 |
| ELOVL1 | 100.0% | 100.0% | 100.0% | 99.5% | Ichthyotic keratoderma, spasticity, hypomyelination, and dysmorphic facies, 618527 |
| ELOVL4 | 100.0% | 100.0% | 99.9% | 97.6% | Spinocerebellar ataxia 34, 133190;Stargardt disease 3, 600110;Ichthyosis, spastic quadriplegia, and impaired intellectual development, 614457 |
| ENAM | 100.0% | 100.0% | 100.0% | 97.6% | Amelogenesis imperfecta, type IC, 204650;Amelogenesis imperfecta, type IB, 104500 |
| ENG | 100.0% | 100.0% | 100.0% | 98.9% | Telangiectasia, hereditary hemorrhagic, type 1, 187300 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| ENPP1 | 100.0% | 99.7% | 100.0% | 97.7% | {Obesity, susceptibility to}, 601665;Hypophosphatemic rickets, autosomal recessive, 2, 613312;{Diabetes mellitus, non-insulin-dependent, susceptibility to}, 125853;Arterial calcification, generalized, of infancy, 1, 208000;Cole disease, 615522 |
| EPG5 | 100.0% | 100.0% | 100.0% | 98.4% | Vici syndrome, 242840 |
| EPHB4 | 100.0% | 100.0% | 100.0% | 99.5% | Capillary malformation-arteriovenous malformation 2, 618196;Lymphatic malformation 7, 617300 |
| EPS8L3 | 100.0% | 100.0% | 100.0% | 98.9% | ?Hypotrichosis 5, 612841 |
| ERCC2 | 99.8% | 96.9% | 100.0% | 99.0% | Xeroderma pigmentosum, group D, 278730;Trichothiodystrophy 1, photosensitive, 601675;?Cerebrooculofacio skeletal syndrome 2, 610756 |
| ERCC3 | 100.0% | 100.0% | 100.0% | 98.5% | Trichothiodystrophy 2, photosensitive, 616390;Xeroderma pigmentosum, group B, 610651 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| ERCC4 | 100.0% | 100.0% | 100.0% | 97.8% | Xeroderma pigmentosum, type F/Cockayne syndrome, 278760;XFE progeroid syndrome, 610965;Xeroderma pigmentosum, group F, 278760;Fanconi anemia, complementation group Q, 615272 |
| ERCC5 | 100.0% | 100.0% | 100.0% | 98.5% | Xeroderma pigmentosum, group G, 278780;Cerebrooculofacios keletal syndrome 3, 616570;Xeroderma pigmentosum, group G/Cockayne syndrome, 278780 |
| ERCC6 | 100.0% | 100.0% | 100.0% | 98.8% | UV-sensitive syndrome 1, 600630;Cerebrooculofacios keletal syndrome 1, 214150;?De Sanctis-Cacchione syndrome, 278800;Cockayne syndrome, type B, 133540;{Macular degeneration, age-related, susceptibility to, 5}, 613761;Premature ovarian failure 11, 616946;{Lung cancer, susceptibility to}, 211980 |
| ERCC8 | 95.2% | 95.2% | 100.0% | 98.1% | UV-sensitive syndrome 2, 614621;Cockayne syndrome, type A, 216400 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| EVC | 100.0% | 99.9% | 100.0% | 98.2% | Ellis-van Creveld syndrome, 225500;?Weyers acrofacial dysostosis, 193530 |
| EVC2 | 100.0% | 100.0% | 100.0% | 98.5% | Ellis-van Creveld syndrome, 225500;Weyers acrofacial dysostosis, 193530 |
| EXPH5 | 100.0% | 100.0% | 100.0% | 97.9% | Epidermolysis bullosa simplex 4, localized or generalized intermediate, autosomal recessive, 615028 |
| F13A1 | 100.0% | 100.0% | 100.0% | 99.2% | Factor XIIIa deficiency, 613225;{Myocardial infarction, protection against}, 608446;{Venous thrombosis, protection against}, 188050 |
| FAM111B | 100.0% | 100.0% | 100.0% | 98.1% | Poikiloderma, hereditary fibrosing, with tendon contractures, myopathy, and pulmonary fibrosis, 615704 |
| FAM20A | 100.0% | 100.0% | 100.0% | 97.8% | Amelogenesis imperfecta, type IG (enamel-renal syndrome), 204690 |
| FAM20C | 100.0% | 100.0% | 100.0% | 97.3% | Raine syndrome, 259775 |
| FAM83G | 100.0% | 100.0% | 100.0% | 99.6% | |
| FAM83H | 100.0% | 100.0% | 100.0% | 99.5% | Amelogenesis imperfecta, type IIIA, 130900 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| FANCA | 100.0% | 100.0% | 100.0% | 98.4% | Fanconi anemia, complementation group A, 227650 |
| FANCB | 96.2% | 96.1% | 96.6% | 67.9% | Fanconi anemia, complementation group B, 300514 |
| FANCC | 100.0% | 100.0% | 100.0% | 98.6% | Fanconi anemia, complementation group C, 227645 |
| FANCD2 | 100.0% | 100.0% | 100.0% | 98.7% | Fanconi anemia, complementation group D2, 227646 |
| FANCE | 100.0% | 100.0% | 100.0% | 98.0% | Fanconi anemia, complementation group E, 600901 |
| FANCF | 100.0% | 100.0% | 100.0% | 98.2% | Fanconi anemia, complementation group F, 603467 |
| FANCG | 100.0% | 100.0% | 100.0% | 98.4% | Fanconi anemia, complementation group G, 614082 |
| FANCI | 100.0% | 100.0% | 100.0% | 98.4% | Fanconi anemia, complementation group I, 609053 |
| FANCL | 90.4% | 87.3% | 100.0% | 98.4% | Fanconi anemia, complementation group L, 614083 |
| FANCM | 100.0% | 100.0% | 100.0% | 97.3% | Premature ovarian failure 15, 618096; Spermatogenic failure 28, 618086 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| FAT4 | 99.9% | 99.8% | 100.0% | 98.9% | Van Maldergem syndrome 2, 615546;Hennekam lymphangiectasia-lymphedema syndrome 2, 616006 |
| FBLN5 | 92.8% | 92.8% | 100.0% | 98.6% | Cutis laxa, autosomal recessive, type IA, 219100;Charcot-Marie-Tooth disease, demyelinating, type 1H, 619764;Macular degeneration, age-related, 3, 608895;?Cutis laxa, autosomal dominant 2, 614434 |
| FDPS | 100.0% | 100.0% | 100.0% | 98.8% | Porokeratosis 9, multiple types, 616631 |
| FECH | 100.0% | 100.0% | 100.0% | 99.1% | Protoporphyrria, erythropoietic, 1, 177000 |
| FERMT1 | 100.0% | 100.0% | 100.0% | 98.3% | Kindler syndrome, 173650 |
| FGF10 | 99.9% | 99.3% | 100.0% | 97.3% | LADD syndrome 3, 620193;Aplasia of lacrimal and salivary glands, 180920 |
| FGF23 | 100.0% | 100.0% | 100.0% | 99.3% | Tumoral calcinosis, hyperphosphatemic, familial, 2, 617993;Hypophosphatemic rickets, autosomal dominant, 193100 |
| FGF3 | 100.0% | 100.0% | 100.0% | 95.9% | Deafness, congenital with inner ear agenesis, microtia, and microdontia, 610706 |

| | | | | | |
|-------|--------|--------|--------|-------|---|
| FGF5 | 100.0% | 100.0% | 100.0% | 98.4% | Trichomegaly, 190330 |
| FGF8 | 100.0% | 100.0% | 99.9% | 96.8% | Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 |
| FGFR1 | 99.7% | 98.5% | 100.0% | 99.1% | 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 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| FGFR2 | 100.0% | 100.0% | 100.0% | 99.0% | Bent bone dysplasia syndrome, 614592;LADD syndrome 1, 149730;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410;Jackson-Weiss syndrome, 123150;Gastric cancer, somatic, 613659;Craniofacial-skeletal-dermatologic dysplasia, 101600;Apert syndrome, 101200;Pfeiffer syndrome, 101600;?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579;Beare-Stevenson cutis gyrata syndrome, 123790;Crouzon syndrome, 123500;Saethre-Chotzen syndrome, 101400;Scaphocephaly and Axenfeld-Rieger anomaly, ;Craniosynostosis, nonspecific, |
|-------|--------|--------|--------|-------|--|

| | | | | | |
|--------|--------|--------|--------|-------|---|
| FGFR3 | 100.0% | 100.0% | 100.0% | 99.8% | 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 |
| FH | 100.0% | 100.0% | 100.0% | 98.5% | Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812 |
| FKBP10 | 100.0% | 100.0% | 100.0% | 98.3% | Osteogenesis imperfecta, type XI, 610968;Bruck syndrome 1, 259450 |
| FKBP14 | 100.0% | 100.0% | 100.0% | 97.5% | Ehlers-Danlos syndrome, kyphoscoliotic type, 2, 614557 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| FLCN | 100.0% | 100.0% | 100.0% | 99.3% | Birt-Hogg-Dube syndrome, 135150;Colorectal cancer, somatic, 114500;Pneumothorax, primary spontaneous, 173600;Renal carcinoma, chromophobe, somatic, 144700 |
| FLG | 100.0% | 100.0% | 100.0% | 97.5% | Ichthyosis vulgaris, 146700;{Dermatitis, atopic, susceptibility to, 2}, 605803 |
| FLG2 | 100.0% | 100.0% | 100.0% | 99.2% | Peeling skin syndrome 6, 618084 |
| FLT4 | 100.0% | 100.0% | 100.0% | 99.2% | Hemangioma, capillary infantile, somatic, 602089;Lymphatic malformation 1, 153100;Congenital heart defects, multiple types, 7, 618780 |
| FNIP1 | 100.0% | 100.0% | 100.0% | 98.8% | Immunodeficiency 93 and hypertrophic cardiomyopathy, 619705 |
| FOXC2 | 100.0% | 100.0% | 99.9% | 92.9% | Lymphedema-distichiasis syndrome, 153400;Lymphedema-distichiasis syndrome with renal disease and diabetes mellitus, 153400 |
| FOXE1 | 100.0% | 100.0% | 99.8% | 90.9% | Bamforth-Lazarus syndrome, 241850;{Thyroid cancer, nonmedullary, 4}, 616534 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| FOXN1 | 100.0% | 100.0% | 100.0% | 99.5% | T-cell lymphopenia, infantile, with or without nail dystrophy, autosomal dominant, 618806;T-cell immunodeficiency, congenital alopecia, and nail dystrophy, 601705 |
| FOXP3 | 100.0% | 99.9% | 98.7% | 73.9% | Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked, 304790 |
| FREM1 | 100.0% | 100.0% | 100.0% | 98.9% | Manitoba oculotrichoanal syndrome, 248450;Bifid nose with or without anorectal and renal anomalies, 608980;Trigonocephaly 2, 614485 |
| FUCA1 | 100.0% | 100.0% | 100.0% | 98.6% | Fucosidosis, 230000 |
| FZD6 | 100.0% | 100.0% | 100.0% | 98.6% | Nail disorder, nonsyndromic congenital, 1, 161050 |
| GALNS | 100.0% | 100.0% | 100.0% | 98.6% | Mucopolysaccharidosis IVA, 253000 |
| GALNT3 | 100.0% | 100.0% | 100.0% | 97.9% | Tumoral calcinosis, hyperphosphatemic, familial, 1, 211900 |
| GAN | 100.0% | 100.0% | 100.0% | 98.5% | Giant axonal neuropathy-1, 256850 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| GATA2 | 85.7% | 85.7% | 100.0% | 98.8% | {Leukemia, acute myeloid, susceptibility to}, 601626;Emberger syndrome, 614038;Immunodeficiency 21, 614172;{Myelodysplastic syndrome, susceptibility to}, 614286 |
| GDF2 | 100.0% | 100.0% | 100.0% | 99.5% | Telangiectasia, hereditary hemorrhagic, type 5, 615506 |
| GDF5 | 100.0% | 100.0% | 100.0% | 99.1% | Acromesomelic dysplasia 2A, 200700;Acromesomelic dysplasia 2B, 228900;Multiple synostoses syndrome 2, 610017;Symphalangism, proximal, 1B, 615298;Brachydactyly, type A2, 112600;?Acromesomelic dysplasia 2C, Hunter-Thompson type, 201250;Brachydactyly, type C, 113100;{Osteoarthritis-5}, 612400;Brachydactyly, type A1, C, 615072 |

| | | | | | |
|------|--------|--------|--------|-------|---|
| GGCX | 100.0% | 100.0% | 100.0% | 98.8% | Vitamin K-dependent clotting factors, combined deficiency of, 1, 277450;Pseudoxanthoma elasticum-like disorder with multiple coagulation factor deficiency, 610842 |
| GJA1 | 100.0% | 100.0% | 100.0% | 97.6% | Erythrokeratoderma 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 |
| GJB2 | 100.0% | 100.0% | 100.0% | 99.4% | Keratoderma, palmoplantar, with deafness, 148350;Deafness, autosomal recessive 1A, 220290;Deafness, autosomal dominant 3A, 601544;Hystrix-like ichthyosis with deafness, 602540;Bart-Pumphrey syndrome, 149200;Keratitis-ichthyosis-deafness syndrome, 148210;Vohwinkel syndrome, 124500 |

| | | | | | |
|------|--------|--------|--------|-------|--|
| GJB3 | 100.0% | 100.0% | 100.0% | 99.5% | Deafness, digenic, GJB2/GJB3, 220290;Erythrokeratoderma variabilis et progressiva 1, 133200;Deafness, autosomal dominant 2B, with or without peripheral neuropathy, 612644 |
| GJB4 | 100.0% | 100.0% | 100.0% | 99.7% | Erythrokeratoderma variabilis et progressiva 2, 617524 |
| GJB6 | 100.0% | 100.0% | 99.9% | 97.9% | Ectodermal dysplasia 2, Clouston type, 129500;Deafness, autosomal dominant 3B, 612643;Deafness, autosomal recessive 1B, 612645;Deafness, digenic GJB2/GJB6, 220290 |
| GJC2 | 99.8% | 98.7% | 100.0% | 96.5% | Lymphatic malformation 3, 613480;?Spastic paraplegia 44, autosomal recessive, 613206;Leukodystrophy, hypomyelinating, 2, 608804 |
| GLA | 91.4% | 91.4% | 98.4% | 73.6% | Fabry disease, cardiac variant, 301500;Fabry disease, 301500 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| GLB1 | 100.0% | 100.0% | 100.0% | 98.9% | GM1-gangliosidosis, type I, 230500;GM1-gangliosidosis, type III, 230650;Mucopolysaccharidosis type IVB (Morquio), 253010;GM1-gangliosidosis, type II, 230600 |
| GLMN | 100.0% | 100.0% | 100.0% | 97.8% | Glomuvenous malformations, 138000 |
| GMPPA | 100.0% | 100.0% | 100.0% | 99.5% | Alacrima, achalasia, and impaired intellectual development syndrome, 615510 |
| GNA11 | 100.0% | 100.0% | 100.0% | 97.4% | Hypocalciuric hypercalcemia, type II, 145981;Hypocalcemia, autosomal dominant 2, 615361 |
| GNA14 | 100.0% | 100.0% | 100.0% | 97.8% | |
| GNAQ | 100.0% | 99.9% | 100.0% | 96.3% | Capillary malformations, congenital, 1, somatic, mosaic, 163000;Sturge-Weber syndrome, somatic, mosaic, 185300 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| GNAS | 100.0% | 99.6% | 99.6% | 94.2% | ACTH-independent macronodular adrenal hyperplasia, 219080;Pituitary adenoma 3, multiple types, somatic, 617686;Pseudohypoparathyroidism 1c, 612462;Pseudohypoparathyroidism 1a, 103580;Osseous heteroplasia, progressive, 166350;Pseudohypoparathyroidism 1b, 603233;McCune-Albright syndrome, somatic, mosaic, 174800;Pseudopseudohypoparathyroidism, 612463 |
| GORAB | 100.0% | 100.0% | 100.0% | 97.3% | Geroderma osteodysplasticum, 231070 |
| GPNMB | 95.1% | 95.1% | 100.0% | 99.2% | Amyloidosis, primary localized cutaneous, 3, 617920 |
| GPR143 | 100.0% | 99.9% | 97.1% | 67.8% | Ocular albinism, type I, Nettleship-Falls type, 300500;Nystagmus 6, congenital, X-linked, 300814 |
| GRHL2 | 100.0% | 100.0% | 100.0% | 98.4% | Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| GRHL3 | 100.0% | 99.9% | 100.0% | 99.3% | van der Woude syndrome 2, 606713 |
| GSN | 100.0% | 100.0% | 100.0% | 98.0% | Amyloidosis, Finnish type, 105120 |
| GTF2E2 | 100.0% | 100.0% | 100.0% | 95.3% | Trichothiodystrophy 6, nonphotosensitive, 616943 |
| GTF2H5 | 59.3% | 59.2% | 100.0% | 98.6% | Trichothiodystrophy 3, photosensitive, 616395 |
| HCCS | 100.0% | 100.0% | 97.8% | 69.8% | Linear skin defects with multiple congenital anomalies 1, 309801 |
| HDAC8 | 97.6% | 97.2% | 97.3% | 71.1% | Cornelia de Lange syndrome 5, 300882 |
| HERC2 | 100.0% | 99.9% | 100.0% | 99.0% | Intellectual developmental disorder, autosomal recessive 38, 615516;[Skin/hair/eye pigmentation 1, blond/brown hair], 227220;[Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220 |
| HLCS | 100.0% | 100.0% | 99.9% | 97.6% | Holocarboxylase synthetase deficiency, 253270 |
| HMBS | 100.0% | 100.0% | 100.0% | 99.1% | Leukoencephalopathy, porphyria-related, 620711;Encephalopathy, porphyria-related, 620704;Porphyria, acute intermittent, nonerythroid variant, 176000;Porphyria, acute intermittent, 176000 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| HMGB3 | 100.0% | 99.9% | 98.0% | 67.6% | ?Microphthalmia, syndromic 13, 300915 |
| HOXC13 | 100.0% | 100.0% | 100.0% | 94.8% | Ectodermal dysplasia 9, hair/nail type, 614931 |
| HPGD | 100.0% | 100.0% | 100.0% | 97.8% | ?Digital clubbing, isolated congenital, 119900;Hypertrophic osteoarthropathy, primary, autosomal recessive 1, 259100;Cranioosteoarthropathy, 259100 |
| HPS1 | 100.0% | 100.0% | 100.0% | 99.5% | Hermansky-Pudlak syndrome 1, 203300 |
| HPS3 | 100.0% | 100.0% | 100.0% | 97.9% | Hermansky-Pudlak syndrome 3, 614072 |
| HPS4 | 100.0% | 100.0% | 100.0% | 99.3% | Hermansky-Pudlak syndrome 4, 614073 |
| HPS5 | 100.0% | 100.0% | 100.0% | 98.4% | Hermansky-Pudlak syndrome 5, 614074 |
| HPS6 | 100.0% | 100.0% | 100.0% | 98.7% | Hermansky-Pudlak syndrome 6, 614075 |
| HR | 100.0% | 100.0% | 100.0% | 99.5% | Atrichia with papular lesions, 209500;Alopecia universalis, 203655 |

| | | | | | |
|-------|--------|--------|--------|-------|---|
| HRAS | 100.0% | 100.0% | 100.0% | 99.6% | Bladder cancer, somatic, 109800;Thyroid carcinoma, follicular, somatic, 188470;Congenital myopathy with excess of muscle spindles, 218040;Nevus sebaceous or woolly hair nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Spitz nevus or nevus spilus, somatic, 137550;Costello syndrome, 218040 |
| HTRA1 | 100.0% | 100.0% | 100.0% | 95.6% | {Macular degeneration, age-related, neovascular type}, 610149;{Macular degeneration, age-related, 7}, 610149;CARASIL syndrome, 600142;Cerebral arteriopathy, autosomal dominant, with subcortical infarcts and leukoencephalopathy, type 2, 616779 |
| HYAL1 | 100.0% | 100.0% | 100.0% | 98.2% | Mucopolysaccharidosis type IX, 601492 |
| IDUA | 100.0% | 100.0% | 100.0% | 97.9% | Mucopolysaccharidosis Is, 607016;Mucopolysaccharidosis Ih/s, 607015;Mucopolysaccharidosis Ih, 607014 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| IFT122 | 100.0% | 100.0% | 100.0% | 99.1% | Cranioectodermal dysplasia 1, 218330 |
| IFT43 | 100.0% | 100.0% | 100.0% | 98.7% | ?Cranioectodermal dysplasia 3, 614099;?Retinitis pigmentosa 81, 617871;Short-rib thoracic dysplasia 18 with polydactyly, 617866 |
| IKBKG | 96.4% | 94.9% | 98.7% | 77.2% | Incontinentia pigmenti, 308300;Ectodermal dysplasia and immunodeficiency 1, 300291;Immunodeficiency 33, 300636;Autoinflammatory disease, systemic, X-linked, 301081 |
| IL17RA | 100.0% | 100.0% | 100.0% | 98.7% | Immunodeficiency 51, 613953 |
| IL17RD | 100.0% | 100.0% | 100.0% | 99.1% | Hypogonadotropic hypogonadism 18 with or without anosmia, 615267 |
| IL1RN | 100.0% | 100.0% | 100.0% | 98.8% | Chronic recurrent multifocal osteomyelitis 2, with periostitis and pustulosis, 612852;{Gastric cancer risk after H. pylori infection}, 613659;{Microvascular complications of diabetes 4}, 612628;Interleukin 1 receptor antagonist deficiency, 612852 |

| | | | | | |
|--------|--------|--------|--------|--------|--|
| IL31RA | 100.0% | 100.0% | 100.0% | 98.2% | ?Amyloidosis, primary localized cutaneous, 2, 613955 |
| IL36RN | 100.0% | 100.0% | 100.0% | 99.2% | Psoriasis 14, pustular, 614204 |
| INSR | 100.0% | 100.0% | 100.0% | 98.4% | Rabson-Mendenhall syndrome, 262190;Diabetes mellitus, insulin-resistant, with acanthosis nigricans, 610549;Donohue syndrome, 246200;Hyperinsulinemic hypoglycemia, familial, 5, 609968 |
| IRF4 | 100.0% | 100.0% | 100.0% | 97.5% | [Skin/hair/eye pigmentation, variation in, 8], 611724 |
| IRF6 | 100.0% | 100.0% | 100.0% | 99.6% | {Orofacial cleft 6}, 608864;Popliteal pterygium syndrome 1, 119500;van der Woude syndrome 1, 119300 |
| ISG15 | 100.0% | 100.0% | 100.0% | 100.0% | Immunodeficiency 38, 616126 |
| ITGA3 | 100.0% | 100.0% | 100.0% | 99.4% | Epidermolysis bullosa, junctional 7, with interstitial lung disease and nephrotic syndrome, 614748 |
| ITGA6 | 100.0% | 100.0% | 100.0% | 98.8% | Epidermolysis bullosa, junctional 6, with pyloric atresia, 619817 |

| | | | | | |
|-------|--------|--------|--------|-------|---|
| ITGB4 | 100.0% | 100.0% | 100.0% | 98.8% | Epidermolysis bullosa, junctional 5B, with pyloric atresia, 226730;Epidermolysis bullosa, junctional 5A, intermediate, 619816 |
| ITGB6 | 100.0% | 100.0% | 100.0% | 98.9% | Amelogenesis imperfecta, type IH, 616221 |
| JUP | 100.0% | 100.0% | 100.0% | 99.4% | Naxos disease, 601214;?Arrhythmogenic right ventricular dysplasia 12, 611528 |
| KANK2 | 100.0% | 100.0% | 100.0% | 99.6% | Nephrotic syndrome, type 16, 617783;Palmoplantar keratoderma and woolly hair, 616099 |
| KAT6B | 100.0% | 100.0% | 100.0% | 98.4% | SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170 |
| KCNH1 | 98.6% | 98.6% | 100.0% | 98.8% | Zimmermann-Laband syndrome 1, 135500;Temple-Baraitser syndrome, 611816 |
| KCNK9 | 100.0% | 100.0% | 99.9% | 96.4% | Birk-Barel syndrome, 612292 |
| KDF1 | 100.0% | 100.0% | 100.0% | 99.0% | ?Ectodermal dysplasia 12, hypohidrotic/hair/tooth/nail type, 617337 |
| KDSR | 100.0% | 100.0% | 99.9% | 98.6% | Erythrokeratoderma variabilis et progressiva 4, 617526 |

| | | | | | |
|-------|--------|--------|--------|-------|---|
| KIF11 | 100.0% | 100.0% | 100.0% | 98.7% | Microcephaly with or without chorioretinopathy, lymphedema, or impaired intellectual development, 152950 |
| KIT | 100.0% | 100.0% | 100.0% | 99.2% | Gastrointestinal stromal tumor, familial, 606764;Mastocytosis, cutaneous, 154800;Piebaldism, 172800;Germ cell tumors, somatic, 273300;Mastocytosis, systemic, somatic, 154800;Leukemia, acute myeloid, somatic, 601626 |
| KITLG | 100.0% | 99.3% | 100.0% | 98.2% | Hyperpigmentation with or without hypopigmentation, 145250;Waardenburg syndrome, type 2F, 619947;Deafness, autosomal dominant 69, unilateral or asymmetric, 616697;[Skin/hair/eye pigmentation 7, blond/brown hair], 611664 |
| KLF4 | 100.0% | 100.0% | 100.0% | 97.2% | |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| KLHL24 | 100.0% | 100.0% | 100.0% | 99.5% | Cardiomyopathy, familial hypertrophic, 29, with polyglucosan bodies, 620236;Epidermolysis bullosa simplex 6, generalized intermediate, with or without cardiomyopathy, 617294 |
| KLK4 | 100.0% | 100.0% | 100.0% | 98.0% | Amelogenesis imperfecta, type IIA1, 204700 |
| KLLN | 100.0% | 100.0% | 100.0% | 95.1% | Cowden syndrome 4, 615107 |
| KMT2D | 100.0% | 100.0% | 100.0% | 98.8% | Branchial arch abnormalities, choanal atresia, athelia, hearing loss, and hypothyroidism syndrome, 620186;Kabuki syndrome 1, 147920 |

| | | | | | |
|------|--------|--------|--------|-------|--|
| KRAS | 100.0% | 100.0% | 100.0% | 99.7% | Gastric cancer, somatic, 613659;Oculoectodermal syndrome, somatic, 600268;Breast cancer, somatic, 114480;Noonan syndrome 3, 609942;RAS-associated autoimmune leukoproliferative disorder, 614470;Arteriovenous malformation of the brain, somatic, 108010;Lung cancer, somatic, 211980;Pancreatic carcinoma, somatic, 260350;Leukemia, acute myeloid, somatic, 601626;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Cardiofaciocutaneous syndrome 2, 615278;Bladder cancer, somatic, 109800 |
|------|--------|--------|--------|-------|--|

| | | | | | |
|-------|--------|--------|--------|-------|--|
| KRT1 | 100.0% | 100.0% | 100.0% | 98.7% | Ichthyosis, annular epidermolytic 2, 620148;Palmoplantar keratoderma, nonepidermolytic, 600962;Epidermolytic hyperkeratosis 1, 113800;Palmoplantar keratoderma, epidermolytic, 2, 620411;Keratosis palmoplantaris striata III, 607654;Ichthyosis histrix, Curth-Macklin type, 146590 |
| KRT10 | 100.0% | 100.0% | 99.7% | 92.4% | Ichthyosis, annular epidermolytic 1, 607602;Epidermolytic hyperkeratosis 2B, autosomal recessive, 620707;Epidermolytic hyperkeratosis 2A, autosomal dominant, 620150;?Ichthyosis histrix, Lambert type, 146600;Ichthyosis with confetti, 609165 |
| KRT13 | 100.0% | 100.0% | 100.0% | 99.6% | White sponge nevus 2, 615785 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| KRT14 | 100.0% | 100.0% | 100.0% | 99.4% | Epidermolysis bullosa simplex 1D, generalized, intermediate or severe, autosomal recessive, 601001;Epidermolysis bullosa simplex 1C, localized, 131800;Dermatopathia pigmentosa reticularis, 125595;Epidermolysis bullosa simplex 1A, generalized severe, 131760;Naegeli-Franceschetti-Jadassohn syndrome, 161000;Epidermolysis bullosa simplex 1B, generalized intermediate, 131900 |
| KRT16 | 100.0% | 100.0% | 100.0% | 99.8% | Palmoplantar keratoderma, nonepidermolytic, focal, 613000;Pachyonychia congenita 1, 167200 |
| KRT17 | 100.0% | 100.0% | 100.0% | 99.7% | Steatocystoma multiplex, 184500;Pachyonychia congenita 2, 167210 |
| KRT2 | 100.0% | 100.0% | 100.0% | 99.0% | Ichthyosis bullosa of Siemens, 146800 |
| KRT4 | 100.0% | 100.0% | 100.0% | 98.9% | White sponge nevus 1, 193900 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| KRT5 | 100.0% | 100.0% | 100.0% | 98.6% | Epidermolysis bullosa simplex 2A, generalized severe, 619555;Dowling-Degos disease 1, 179850;Epidermolysis bullosa simplex 2F, with mottled pigmentation, 131960;Epidermolysis bullosa simplex 2D, generalized, intermediate or severe, autosomal recessive, 619599;Epidermolysis bullosa simplex 2B, generalized intermediate, 619588;Epidermolysis bullosa simplex 2C, localized, 619594;Epidermolysis bullosa simplex 2E, with migratory circinate erythema, 609352 |
| KRT6A | 100.0% | 100.0% | 100.0% | 98.7% | Pachyonychia congenita 3, 615726 |
| KRT6B | 100.0% | 100.0% | 100.0% | 99.1% | Pachyonychia congenita 4, 615728 |
| KRT6C | 99.9% | 99.7% | 98.9% | 92.3% | Palmoplantar keratoderma, nonepidermolytic, focal or diffuse, 615735 |
| KRT71 | 100.0% | 100.0% | 100.0% | 99.5% | ?Hypotrichosis 13, 615896 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| KRT74 | 100.0% | 100.0% | 100.0% | 99.3% | Woolly hair, autosomal dominant, 194300;?Hypotrichosis 3, 613981;?Ectodermal dysplasia 7, hair/nail type, 614929 |
| KRT75 | 100.0% | 100.0% | 100.0% | 98.8% | {Pseudofolliculitis barbae, susceptibility to}, 612318 |
| KRT81 | 100.0% | 100.0% | 100.0% | 98.9% | Monilethrix, 158000 |
| KRT82 | 100.0% | 100.0% | 100.0% | 99.0% | |
| KRT83 | 100.0% | 100.0% | 100.0% | 99.6% | Monilethrix, 158000;Erythrokeratoderma variabilis et progressiva 5, 617756 |
| KRT85 | 100.0% | 100.0% | 100.0% | 99.4% | Ectodermal dysplasia 4, hair/nail type, 602032 |
| KRT86 | 100.0% | 100.0% | 100.0% | 99.4% | Monilethrix, 158000 |
| KRT9 | 100.0% | 100.0% | 100.0% | 97.2% | Palmoplantar keratoderma, epidermolytic, 1, 144200 |
| LAMA3 | 100.0% | 100.0% | 100.0% | 98.7% | Epidermolysis bullosa, junctional 2A, intermediate, 619783;Epidermolysis bullosa, junctional 2C, laryngoonychocutaneous, 245660;Epidermolysis bullosa, junctional 2B, severe, 619784 |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| LAMB3 | 100.0% | 100.0% | 100.0% | 99.5% | Epidermolysis bullosa, junctional 1B, severe, 226700;Epidermolysis bullosa, junctional 1A, intermediate, 226650;Amelogenesis imperfecta, type IA, 104530 |
| LAMC2 | 100.0% | 100.0% | 100.0% | 99.2% | Epidermolysis bullosa, junctional 3B, severe, 619786;Epidermolysis bullosa, junctional 3A, intermediate, 619785 |
| LAMTOR2 | 100.0% | 100.0% | 100.0% | 99.6% | Immunodeficiency due to defect in MAPBP-interacting protein, 610798 |
| LDHA | 100.0% | 100.0% | 100.0% | 98.4% | Glycogen storage disease XI, 612933 |
| LDLRAP1 | 100.0% | 100.0% | 100.0% | 99.0% | Hypercholesterolemia, familial, 4, 603813 |
| LEMD3 | 100.0% | 100.0% | 99.9% | 94.6% | Buschke-Ollendorff syndrome, 166700;Osteopoikilosis with or without melorheostosis, 166700 |
| LIPH | 100.0% | 100.0% | 100.0% | 98.3% | Hypotrichosis 7, 604379;Woolly hair, autosomal recessive 2 with or without hypotrichosis, 604379 |
| LIPN | 100.0% | 100.0% | 100.0% | 98.4% | Ichthyosis, congenital, autosomal recessive 8, 613943 |

| | | | | | |
|----------|--------|--------|--------|-------|---|
| LMBRD1 | 100.0% | 99.8% | 100.0% | 96.9% | Methylmalonic aciduria and homocystinuria, cbIF type, 277380 |
| LMNA | 100.0% | 100.0% | 100.0% | 99.2% | Mandibuloacral dysplasia, 248370;Heart-hand syndrome, Slovenian type, 610140;Cardiomyopathy, dilated, 1A, 115200;Emery-Dreifuss muscular dystrophy 3, autosomal recessive, 616516;Restrictive dermopathy 2, 619793;Charcot-Marie-Tooth disease, type 2B1, 605588;Emery-Dreifuss muscular dystrophy 2, autosomal dominant, 181350;Hutchinson-Gilford progeria, 176670;Lipodystrophy, familial partial, type 2, 151660;Muscular dystrophy, congenital, 613205;Malouf syndrome, 212112 |
| LMX1B | 100.0% | 100.0% | 99.9% | 94.8% | Focal segmental glomerulosclerosis 10, 256020;Nail-patella syndrome, 161200 |
| LONP1 | 100.0% | 100.0% | 100.0% | 99.1% | CODAS syndrome, 600373 |
| LORICRIN | 100.0% | 100.0% | 99.6% | 82.5% | Vohwinkel syndrome with ichthyosis, 604117 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| LPAR6 | 100.0% | 99.8% | 99.9% | 94.7% | Hypotrichosis 8, 278150;Woolly hair, autosomal recessive 1, with or without hypotrichosis, 278150 |
| LPIN2 | 99.3% | 99.2% | 100.0% | 98.6% | Majeed syndrome, 609628 |
| LRMDA | 97.8% | 97.8% | 100.0% | 99.3% | Albinism, oculocutaneous, type VII, 615179 |
| LSS | 100.0% | 100.0% | 100.0% | 99.5% | Hypotrichosis 14, 618275;Cataract 44, 616509;Alopecia-intellectual disability syndrome 4, 618840 |
| LTBP3 | 100.0% | 100.0% | 100.0% | 96.9% | Dental anomalies and short stature, 601216;Geleophysic dysplasia 3, 617809 |
| LTBP4 | 100.0% | 100.0% | 100.0% | 98.6% | Cutis laxa, autosomal recessive, type IC, 613177 |
| LTV1 | 100.0% | 100.0% | 100.0% | 98.8% | Inflammatory poikiloderma with hair abnormalities and acral keratoses, 620199 |
| LYST | 99.5% | 99.3% | 100.0% | 98.8% | Chediak-Higashi syndrome, 214500 |
| LYZ | 100.0% | 100.0% | 100.0% | 99.2% | Amyloidosis, hereditary systemic 5, 620658 |
| MAP2K1 | 95.8% | 95.8% | 100.0% | 98.9% | Cardiofaciocutaneous syndrome 3, 615279;Melorheostosis, isolated, somatic mosaic, 155950 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| MAP2K2 | 100.0% | 100.0% | 100.0% | 98.7% | Cardiofaciocutaneous syndrome 4, 615280 |
| MBTPS2 | 100.0% | 100.0% | 98.6% | 70.4% | Keratosis follicularis spinulosa decalvans, X-linked, 308800;Osteogenesis imperfecta, type XIX, 301014;IFAP syndrome with or without BRESHECK syndrome, 308205;?Olmsted syndrome, X-linked, 300918 |
| MDFIC | 100.0% | 99.3% | 100.0% | 97.8% | Lymphatic malformation 12, 620014 |
| MED12 | 100.0% | 99.8% | 97.5% | 69.0% | Lujan-Fryns syndrome, 309520;Ohdo syndrome, X-linked, 300895;Hardikar syndrome, 301068;Opitz-Kaveggia syndrome, 305450 |
| MEFV | 96.1% | 96.1% | 100.0% | 99.4% | Neutrophilic dermatosis, acute febrile, 608068;Familial Mediterranean fever, AR, 249100;Familial Mediterranean fever, AD, 134610 |
| MGP | 100.0% | 100.0% | 100.0% | 97.1% | Keutel syndrome, 245150 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| MITF | 99.9% | 99.7% | 100.0% | 98.5% | Waardenburg syndrome, type 2A, 193510;{Melanoma, cutaneous malignant, susceptibility to, 8}, 614456;Tietz albinism-deafness syndrome, 103500;COMMAD syndrome, 617306 |
| MLH1 | 100.0% | 100.0% | 100.0% | 97.6% | Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300 |
| MLPH | 100.0% | 100.0% | 100.0% | 99.2% | Griscelli syndrome, type 3, 609227 |
| MMACHC | 100.0% | 100.0% | 100.0% | 98.8% | Methylmalonic aciduria and homocystinuria, cb1C type, 277400 |
| MMP14 | 94.9% | 94.9% | 100.0% | 99.3% | Winchester syndrome, 277950 |
| MMP2 | 100.0% | 100.0% | 100.0% | 98.8% | Multicentric osteolysis, nodulosis, and arthropathy, 259600 |
| MMP20 | 100.0% | 100.0% | 100.0% | 98.9% | Amelogenesis imperfecta, type IIA2, 612529 |
| MPLKIP | 100.0% | 100.0% | 100.0% | 97.4% | Trichothiodystrophy 4, nonphotosensitive, 234050 |
| MRE11 | 100.0% | 100.0% | 100.0% | 97.3% | Ataxia-telangiectasia-like disorder 1, 604391 |

| | | | | | |
|-------|--------|--------|--------|-------|---|
| MSH2 | 100.0% | 100.0% | 100.0% | 98.0% | Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096 |
| MSX1 | 100.0% | 100.0% | 99.9% | 96.5% | Tooth agenesis, selective, 1, with or without orofacial cleft, 106600;Ectodermal dysplasia 3, Witkop type, 189500;Orofacial cleft 5, 608874 |
| MTOR | 100.0% | 100.0% | 100.0% | 99.3% | Focal cortical dysplasia, type II, somatic, 607341;Smith-Kingsmore syndrome, 616638 |
| MUTYH | 100.0% | 100.0% | 100.0% | 99.4% | Adenomas, multiple colorectal, 608456;Gastric cancer, somatic, 613659 |
| MVD | 100.0% | 100.0% | 100.0% | 99.8% | Porokeratosis 7, multiple types, 614714 |
| MVK | 100.0% | 100.0% | 100.0% | 99.7% | Hyper-IgD syndrome, 260920;Porokeratosis 3, multiple types, 175900;Mevalonic aciduria, 610377 |
| MYH8 | 100.0% | 100.0% | 100.0% | 98.5% | Carney complex variant, 608837;Trismus-pseudocamptodactyly syndrome, 158300 |
| MYO5A | 99.0% | 99.0% | 100.0% | 98.3% | Griscelli syndrome, type 1, 214450 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| NAA10 | 100.0% | 100.0% | 98.1% | 69.0% | Microphthalmia, syndromic 1, 309800;Ogden syndrome, 300855 |
| NAGA | 100.0% | 100.0% | 100.0% | 99.5% | Schindler disease, type I, 609241;Kanzaki disease, 609242;Schindler disease, type III, 609241 |
| NBAS | 100.0% | 99.8% | 100.0% | 98.7% | Short stature, optic nerve atrophy, and Pelger-Huet anomaly, 614800;Infantile liver failure syndrome 2, 616483 |
| NCF1 | 100.0% | 99.6% | 100.0% | 96.8% | Chronic granulomatous disease 1, autosomal recessive, 233700 |
| NCSTN | 100.0% | 100.0% | 100.0% | 99.1% | Acne inversa, familial, 1, 142690 |
| NDUFB11 | 99.7% | 97.9% | 88.1% | 61.0% | Linear skin defects with multiple congenital anomalies 3, 300952;?Mitochondrial complex I deficiency, nuclear type 30, 301021 |
| NECTIN1 | 93.4% | 93.4% | 100.0% | 99.0% | Cleft lip/palate-ectodermal dysplasia syndrome, 225060;Orofacial cleft 7, 225060 |
| NECTIN4 | 100.0% | 100.0% | 100.0% | 99.7% | Ectodermal dysplasia-syndactyly syndrome 1, 613573 |
| NEK11 | 100.0% | 99.9% | 100.0% | 98.1% | |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| NEK9 | 100.0% | 100.0% | 100.0% | 98.7% | ?Arthrogryposis, Perthes disease, and upward gaze palsy, 614262;Nevus comedonicus, somatic, 617025;Lethal congenital contracture syndrome 10, 617022 |
| NF1 | 99.4% | 99.4% | 100.0% | 98.6% | Watson syndrome, 193520;Leukemia, juvenile myelomonocytic, 607785;Neurofibromatosis, familial spinal, 162210;Neurofibromatosis, type 1, 162200;Neurofibromatosis-Noonan syndrome, 601321 |
| NFKBIA | 100.0% | 100.0% | 100.0% | 95.2% | Ectodermal dysplasia and immunodeficiency 2, 612132 |
| NHP2 | 100.0% | 100.0% | 100.0% | 98.7% | Dyskeratosis congenita, autosomal recessive 2, 613987 |
| NIPAL4 | 100.0% | 100.0% | 100.0% | 98.5% | Ichthyosis, congenital, autosomal recessive 6, 612281 |
| NIPBL | 100.0% | 100.0% | 100.0% | 98.4% | Cornelia de Lange syndrome 1, 122470 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| NLRP1 | 98.1% | 98.1% | 100.0% | 98.8% | {Vitiligo-associated multiple autoimmune disease susceptibility 1}, 606579;?Respiratory papillomatosis, juvenile recurrent, congenital, 618803;Autoinflammation with arthritis and dyskeratosis, 617388;Palmoplantar carcinoma, multiple self-healing, 615225 |
| NLRP12 | 100.0% | 100.0% | 100.0% | 97.9% | Familial cold autoinflammatory syndrome 2, 611762 |
| NLRP3 | 100.0% | 100.0% | 100.0% | 98.8% | CINCA syndrome, 607115;Familial cold inflammatory syndrome 1, 120100;Keratoendothelitis fugax hereditaria, 148200;Deafness, autosomal dominant 34, with or without inflammation, 617772;Muckle-Wells syndrome, 191900 |
| NME1 | 100.0% | 100.0% | 100.0% | 99.6% | |
| NOD2 | 100.0% | 100.0% | 100.0% | 99.5% | Blau syndrome, 186580;{Yao syndrome}, 617321;{Inflammatory bowel disease 1, Crohn disease}, 266600 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| NOP10 | 92.5% | 92.4% | 100.0% | 96.6% | ?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230 |
| NOTCH1 | 99.1% | 99.0% | 100.0% | 99.6% | Adams-Oliver syndrome 5, 616028;Aortic valve disease 1, 109730 |
| NRAS | 100.0% | 100.0% | 100.0% | 99.7% | Noonan syndrome 6, 613224;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic, 614470;Melanocytic nevus syndrome, congenital, somatic, 137550;Epidermal nevus, somatic, 162900;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic, 163200;Thyroid carcinoma, follicular, somatic, 188470;Neurocutaneous melanosis, somatic, 249400;Colorectal cancer, somatic, 114500 |
| NSD1 | 100.0% | 100.0% | 100.0% | 98.6% | Sotos syndrome, 117550 |

| | | | | | |
|-------|--------|--------|--------|-------|---|
| NSDHL | 100.0% | 99.9% | 99.5% | 74.7% | CK syndrome, 300831;CHILD syndrome, 308050 |
| OCA2 | 100.0% | 100.0% | 100.0% | 99.4% | [Skin/hair/eye pigmentation 1, blue/nonblue eyes], 227220;[Skin/hair/eye pigmentation 1, blond/brown hair], 227220;Albinism, brown oculocutaneous, 203200;Albinism, oculocutaneous, type II, 203200 |
| ODAM | 100.0% | 99.8% | 100.0% | 98.4% | |
| ODAPH | 100.0% | 100.0% | 100.0% | 97.4% | Amelogenesis imperfecta, type IIA4, 614832 |
| OFD1 | 100.0% | 100.0% | 96.1% | 66.2% | Simpson-Golabi-Behmel syndrome, type 2, 300209;?Retinitis pigmentosa 23, 300424;Orofaciodigital syndrome I, 311200;Joubert syndrome 10, 300804 |
| OSMR | 100.0% | 100.0% | 100.0% | 99.0% | Amyloidosis, primary localized cutaneous, 1, 105250 |
| PADI3 | 100.0% | 100.0% | 100.0% | 99.4% | Uncombable hair syndrome, 191480 |
| PAH | 100.0% | 100.0% | 100.0% | 99.2% | [Hyperphenylalaninemia, non-PKU mild], 261600;Phenylketonuria, 261600 |

| | | | | | |
|-------|--------|--------|--------|-------|---|
| PALB2 | 100.0% | 100.0% | 100.0% | 96.7% | {Breast-ovarian cancer, familial, susceptibility to, 5}, 620442;{Pancreatic cancer, susceptibility to, 3}, 613348;Fanconi anemia, complementation group N, 610832 |
| PAX3 | 100.0% | 99.8% | 100.0% | 98.5% | Craniofacial-deafness-hand syndrome, 122880;Waardenburg syndrome, type 3, 148820;Waardenburg syndrome, type 1, 193500;Rhabdomyosarcoma 2, alveolar, 268220 |
| PAX9 | 100.0% | 100.0% | 100.0% | 99.1% | Tooth agenesis, selective, 3, 604625 |
| PCNA | 100.0% | 100.0% | 100.0% | 99.2% | ?Ataxia-telangiectasia-like disorder 2, 615919 |
| PDGFB | 100.0% | 100.0% | 99.7% | 96.7% | Meningioma, SIS-related, 607174;Basal ganglia calcification, idiopathic, 5, 615483;Dermatofibrosarcoma protuberans, 607907 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| PDGFRB | 100.0% | 100.0% | 100.0% | 99.2% | Premature aging syndrome, Penttinen type, 601812;Kosaki overgrowth syndrome, 616592;Myofibromatosis, infantile, 1, 228550;Basal ganglia calcification, idiopathic, 4, 615007;Myeloproliferative disorder with eosinophilia, 131440 |
| PEPD | 93.9% | 93.9% | 100.0% | 99.5% | Prolidase deficiency, 170100 |
| PERP | 100.0% | 100.0% | 100.0% | 98.8% | Erythrokeratoderma variabilis et progressiva 7, 619209;Olmsted syndrome 2, 619208 |
| PEX7 | 97.9% | 97.9% | 100.0% | 98.8% | Rhizomelic chondrodysplasia punctata, type 1, 215100;Peroxisome biogenesis disorder 9B, 614879 |
| PHEX | 99.9% | 99.2% | 98.1% | 70.9% | Hypophosphatemic rickets, X-linked dominant, 307800 |
| PHGDH | 100.0% | 100.0% | 100.0% | 99.2% | Neu-Laxova syndrome 1, 256520;Phosphoglycerate dehydrogenase deficiency, 601815 |
| PHYH | 100.0% | 100.0% | 100.0% | 98.2% | Refsum disease, 266500 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| PIEZO1 | 100.0% | 100.0% | 100.0% | 99.7% | [ER blood group system], 620207;Lymphatic malformation 6, 616843;Dehydrated hereditary stomatocytosis with or without pseudohyperkalemia and/or perinatal edema, 194380 |
| PIGA | 100.0% | 100.0% | 97.7% | 73.6% | Paroxysmal nocturnal hemoglobinuria, somatic, 300818;Multiple congenital anomalies-hypotonia-seizures syndrome 2, 300868;Neurodevelopmental disorder with epilepsy and hemochromatosis, 301072 |
| PIGL | 100.0% | 100.0% | 100.0% | 98.2% | CHIME syndrome, 280000 |
| PIGN | 100.0% | 99.9% | 100.0% | 98.6% | Multiple congenital anomalies-hypotonia-seizures syndrome 1, 614080 |
| PIGV | 100.0% | 99.6% | 100.0% | 99.3% | Hyperphosphatasia with impaired intellectual development syndrome 1, 239300 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| PIK3CA | 100.0% | 100.0% | 100.0% | 98.0% | Hemifacial myohyperplasia, somatic, 606773;CLOVE syndrome, somatic, 612918;Hepatocellular carcinoma, somatic, 114550;Breast cancer, somatic, 114480;Cerebral cavernous malformations 4, somatic, 619538;Ovarian cancer, somatic, 167000;Colorectal cancer, somatic, 114500;Macrodactyly, somatic, 155500;CLAPO syndrome, somatic, 613089;Keratosis, seborrheic, somatic, 182000;Nevus, epidermal, somatic, 162900;Gastric cancer, somatic, 613659;Nonsmall cell lung cancer, somatic, 211980;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic, 602501;Cowden syndrome 5, 615108 |
| PITX2 | 100.0% | 100.0% | 100.0% | 98.1% | Ring dermoid of cornea, 180550;Axenfeld-Rieger syndrome, type 1, 180500;Anterior segment dysgenesis 4, 137600 |
| PKP1 | 100.0% | 100.0% | 100.0% | 99.0% | Ectodermal dysplasia/skin fragility syndrome, 604536 |

| | | | | | |
|-------|--------|--------|--------|-------|---|
| PLCD1 | 100.0% | 100.0% | 100.0% | 99.6% | Nail disorder, nonsyndromic congenital, 3, (leukonychia), 151600 |
| PLCG2 | 100.0% | 100.0% | 100.0% | 99.0% | Autoinflammation, antibody deficiency, and immune dysregulation syndrome, 614878;Familial cold autoinflammatory syndrome 3, 614468 |
| PLEC | 100.0% | 100.0% | 100.0% | 99.7% | ?Epidermolysis bullosa simplex 5D, generalized intermediate, autosomal recessive, 616487;Epidermolysis bullosa simplex 5B, with muscular dystrophy, 226670;Epidermolysis bullosa simplex 5C, with pyloric atresia, 612138;Epidermolysis bullosa simplex 5A, Ogna type, 131950;Muscular dystrophy, limb-girdle, autosomal recessive 17, 613723 |
| PLG | 100.0% | 100.0% | 100.0% | 98.9% | Dysplasminogenemia, 217090;Angioedema, hereditary, 4, 619360;Plasminogen deficiency, type I, 217090 |
| PLIN1 | 100.0% | 100.0% | 100.0% | 98.4% | Lipodystrophy, familial partial, type 4, 613877 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| PLOD1 | 100.0% | 100.0% | 100.0% | 98.5% | Ehlers-Danlos syndrome, kyphoscoliotic type, 1, 225400 |
| PLOD3 | 100.0% | 100.0% | 100.0% | 98.0% | BCARD syndrome (lysyl hydroxylase 3 deficiency), 612394 |
| PMS2 | 93.4% | 93.4% | 99.3% | 95.2% | Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101 |
| PMVK | 100.0% | 100.0% | 100.0% | 98.5% | Porokeratosis 1, multiple types, 175800 |
| PNPLA1 | 100.0% | 99.9% | 100.0% | 98.3% | Ichthyosis, congenital, autosomal recessive 10, 615024 |
| PNPLA2 | 100.0% | 100.0% | 100.0% | 99.5% | Neutral lipid storage disease with myopathy, 610717 |
| POC1A | 100.0% | 100.0% | 100.0% | 99.6% | Short stature, onychodysplasia, facial dysmorphism, and hypotrichosis, 614813 |
| POFUT1 | 100.0% | 100.0% | 100.0% | 98.7% | Dowling-Degos disease 2, 615327 |
| POGLUT1 | 100.0% | 100.0% | 100.0% | 98.8% | Dowling-Degos disease 4, 615696;Muscular dystrophy, limb-girdle, autosomal recessive 21, 617232 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| POLD1 | 100.0% | 100.0% | 100.0% | 99.2% | Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591 |
| POLH | 100.0% | 100.0% | 100.0% | 99.3% | Xeroderma pigmentosum, variant type, 278750 |
| POLR1C | 83.3% | 83.2% | 100.0% | 99.1% | Leukodystrophy, hypomyelinating, 11, 616494;Treacher Collins syndrome 3, 248390 |
| POLR1D | 100.0% | 100.0% | 100.0% | 98.8% | Treacher Collins syndrome 2, 613717 |
| POLR3A | 100.0% | 100.0% | 100.0% | 98.8% | Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 |
| POLR3B | 100.0% | 99.9% | 100.0% | 98.3% | Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| POMC | 100.0% | 100.0% | 100.0% | 99.2% | {Obesity, early-onset, susceptibility to}, 601665;Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 |
| POMP | 83.4% | 83.2% | 100.0% | 97.0% | Proteasome-associated autoinflammatory syndrome 2, 618048;Keratosi linearis with ichthyosis congenita and sclerosing keratoderma, 601952 |
| PORCN | 100.0% | 99.8% | 98.2% | 71.4% | Focal dermal hypoplasia, 305600 |
| POT1 | 100.0% | 100.0% | 99.9% | 98.3% | Tumor predisposition syndrome 3, 615848;?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368;?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367 |
| PPOX | 100.0% | 100.0% | 100.0% | 98.9% | Variegate porphyria, childhood-onset, 620483;Variegate porphyria, 176200 |
| PQBP1 | 100.0% | 100.0% | 97.9% | 68.4% | Renpenning syndrome, 309500 |
| PRDM10 | 100.0% | 100.0% | 100.0% | 99.4% | ?Birt-Hogg-Dube syndrome 2, 620459 |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| PRKAR1A | 100.0% | 100.0% | 100.0% | 98.9% | Pigmented nodular adrenocortical disease, primary, 1, 610489;Acrodysostosis 1, with or without hormone resistance, 101800;Carney complex, type 1, 160980;Myxoma, intracardiac, 255960;Adrenocortical tumor, somatic, |
| PSEN1 | 100.0% | 100.0% | 100.0% | 99.2% | Pick disease, 172700;Alzheimer disease, type 3, with spastic paraparesis and apraxia, 607822;Dementia, frontotemporal, 600274;?Acne inversa, familial, 3, 613737;Cardiomyopathy, dilated, 1U, 613694;Alzheimer disease, type 3, with spastic paraparesis and unusual plaques, 607822;Alzheimer disease, type 3, 607822 |
| PSENE1 | 100.0% | 100.0% | 100.0% | 98.2% | Acne inversa, familial, 2, with or without Dowling-Degos disease, 613736 |
| PSMB8 | 100.0% | 100.0% | 99.9% | 98.2% | Proteasome-associated autoinflammatory syndrome 1 and digenic forms, 256040 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| PSTPIP1 | 100.0% | 100.0% | 100.0% | 99.6% | Pyogenic sterile arthritis, pyoderma gangrenosum, and acne, 604416 |
| PTCH1 | 100.0% | 100.0% | 100.0% | 97.3% | Basal cell nevus syndrome 1, 109400;Basal cell carcinoma, somatic, 605462;Holoprosencephaly 7, 610828 |
| PTCH2 | 100.0% | 100.0% | 100.0% | 99.3% | Medulloblastoma, somatic, 155255;Basal cell carcinoma, somatic, 605462 |
| PTDSS1 | 100.0% | 100.0% | 100.0% | 98.2% | Lenz-Majewski hyperostotic dwarfism, 151050 |
| PTEN | 94.5% | 94.5% | 99.8% | 93.1% | {Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309 |
| PTHLH | 100.0% | 100.0% | 100.0% | 98.2% | Brachydactyly, type E2, 613382 |
| PTPN11 | 89.3% | 89.2% | 100.0% | 98.3% | Noonan syndrome 1, 163950;LEOPARD syndrome 1, 151100;Metachondromatosis, 156250;Leukemia, juvenile myelomonocytic, somatic, 607785 |
| PTPN14 | 100.0% | 100.0% | 100.0% | 99.3% | Choanal atresia and lymphedema, 613611 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| PTPRF | 100.0% | 100.0% | 100.0% | 99.6% | ?Breasts and/or nipples, aplasia or hypoplasia of, 2, 616001 |
| PYCR1 | 100.0% | 100.0% | 100.0% | 99.8% | Cutis laxa, autosomal recessive, type IIIB, 614438;Cutis laxa, autosomal recessive, type IIB, 612940 |
| RAB23 | 100.0% | 100.0% | 100.0% | 97.2% | Carpenter syndrome, 201000 |
| RAB27A | 100.0% | 100.0% | 100.0% | 99.1% | Griscelli syndrome, type 2, 607624 |
| RAD21 | 100.0% | 100.0% | 100.0% | 98.3% | Cornelia de Lange syndrome 4, 614701;?Mungan syndrome, 611376 |
| RAD50 | 100.0% | 100.0% | 100.0% | 96.9% | Nijmegen breakage syndrome-like disorder, 613078 |
| RAF1 | 95.6% | 92.7% | 100.0% | 98.7% | Cardiomyopathy, dilated, 1NN, 615916;Noonan syndrome 5, 611553;LEOPARD syndrome 2, 611554 |

| | | | | | |
|-------|--------|--------|--------|-------|--|
| RAG1 | 100.0% | 100.0% | 100.0% | 99.1% | Omenn syndrome, 603554; Severe combined immunodeficiency, B cell-negative, 601457; Combined cellular and humoral immune defects with granulomas, 233650; Alpha/beta T-cell lymphopenia with gamma/delta T-cell expansion, severe cytomegalovirus infection, and autoimmunity, 609889 |
| RAG2 | 100.0% | 100.0% | 100.0% | 98.3% | Severe combined immunodeficiency, B cell-negative, 601457; Combined cellular and humoral immune defects with granulomas, 233650; Omenn syndrome, 603554 |
| RAI1 | 100.0% | 100.0% | 100.0% | 98.9% | Smith-Magenis syndrome, 182290 |
| RBBP8 | 100.0% | 100.0% | 100.0% | 97.3% | Seckel syndrome 2, 606744; Jawad syndrome, 251255; Pancreatic carcinoma, somatic, |
| RBM28 | 100.0% | 100.0% | 100.0% | 98.7% | ? Alopecia, neurologic defects, and endocrinopathy syndrome, 612079 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| RBP4 | 100.0% | 100.0% | 100.0% | 98.8% | Microphthalmia, isolated, with coloboma 10, 616428;Retinal dystrophy, iris coloboma, and comedogenic acne syndrome, 615147 |
| RBPJ | 100.0% | 100.0% | 100.0% | 98.5% | Adams-Oliver syndrome 3, 614814 |
| RECQL4 | 100.0% | 100.0% | 100.0% | 99.2% | Baller-Gerold syndrome, 218600;Rothmund-Thomson syndrome, type 2, 268400;RAPADILINO syndrome, 266280 |
| RHBDF2 | 100.0% | 100.0% | 100.0% | 99.7% | Tylosis with esophageal cancer, 148500 |
| RHOA | 80.4% | 80.4% | 100.0% | 97.2% | Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic, 618727 |
| RIN2 | 100.0% | 100.0% | 100.0% | 98.5% | Macrocephaly, alopecia, cutis laxa, and scoliosis, 613075 |
| RIPK4 | 100.0% | 100.0% | 100.0% | 99.7% | CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650 |

| | | | | | |
|----------|--------|--------|--------|-------|--|
| RMRP | | | | | Anauxetic dysplasia 1, 607095;Metaphyseal dysplasia without hypotrichosis, 250460;Cartilage-hair hypoplasia, 250250 |
| RNASEH2A | 100.0% | 100.0% | 100.0% | 99.3% | Aicardi-Goutieres syndrome 4, 610333 |
| RNASEH2B | 91.4% | 91.4% | 100.0% | 97.3% | Aicardi-Goutieres syndrome 2, 610181 |
| RNASEH2C | 100.0% | 100.0% | 100.0% | 97.1% | Aicardi-Goutieres syndrome 3, 610329 |
| RNU4ATAC | | | | | Roifman syndrome, 616651;Lowry-Wood syndrome, 226960;Microcephalic osteodysplastic primordial dwarfism, type I, 210710 |
| ROGDI | 100.0% | 100.0% | 100.0% | 99.1% | Kohlschutter-Tonz syndrome, 226750 |
| RPL21 | 100.0% | 100.0% | 100.0% | 99.6% | Hypotrichosis 12, 615885 |
| RSPO1 | 100.0% | 100.0% | 100.0% | 99.6% | Palmoplantar hyperkeratosis and true hermaphroditism, 610644;Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 |
| RSPO4 | 100.0% | 100.0% | 100.0% | 98.4% | Anonychia congenita, 206800 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| RTEL1 | 100.0% | 100.0% | 100.0% | 99.4% | Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373 |
| RUNX2 | 100.0% | 100.0% | 99.9% | 95.3% | 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 |
| SAMD9 | 100.0% | 100.0% | 100.0% | 97.2% | Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053 |
| SAMHD1 | 100.0% | 100.0% | 100.0% | 98.1% | ?Chilblain lupus 2, 614415;Aicardi-Goutieres syndrome 5, 612952 |
| SART3 | 100.0% | 100.0% | 100.0% | 99.0% | |

| | | | | | |
|-----------|--------|--------|--------|-------|---|
| SASH1 | 100.0% | 100.0% | 100.0% | 98.2% | Dyschromatosis universalis hereditaria 1, 127500;?Cancer, alopecia, pigment dyscrasia, onychodystrophy, and keratoderma, 618373 |
| SAT1 | 100.0% | 100.0% | 97.1% | 66.3% | |
| SATB2 | 100.0% | 99.7% | 100.0% | 98.6% | Glass syndrome, 612313 |
| SCN10A | 100.0% | 100.0% | 100.0% | 98.8% | Episodic pain syndrome, familial, 2, 615551 |
| SCN11A | 100.0% | 99.9% | 99.9% | 97.2% | Episodic pain syndrome, familial, 3, 615552;Neuropathy, hereditary sensory and autonomic, type VII, 615548 |
| SCN9A | 100.0% | 99.9% | 100.0% | 97.5% | Erythralgia, primary, 133020;Insensitivity to pain, congenital, 243000;Small fiber neuropathy, 133020;Paroxysmal extreme pain disorder, 167400;Neuropathy, hereditary sensory and autonomic, type IID, 243000 |
| SDR9C7 | 100.0% | 100.0% | 100.0% | 99.8% | Ichthyosis, congenital, autosomal recessive 13, 617574 |
| SEC23B | 100.0% | 100.0% | 100.0% | 98.4% | ?Cowden syndrome 7, 616858;Dyserythropoietic anemia, congenital, type II, 224100 |
| SERPINA12 | 100.0% | 100.0% | 100.0% | 98.0% | |

| | | | | | |
|----------|--------|--------|--------|-------|---|
| SERPINA3 | 100.0% | 100.0% | 100.0% | 98.8% | Alpha-1-antichymotrypsin deficiency, ;Cerebrovascular disease, occlusive, |
| SERPINB7 | 100.0% | 100.0% | 100.0% | 98.4% | Palmoplantar keratoderma, Nagashima type, 615598 |
| SERPINB8 | 100.0% | 100.0% | 100.0% | 98.2% | Peeling skin syndrome 5, 617115 |
| SERPING1 | 100.0% | 100.0% | 100.0% | 99.0% | Angioedema, hereditary, 1 and 2, 106100;Complement component 4, partial deficiency of, 120790 |
| SERPINH1 | 100.0% | 100.0% | 100.0% | 99.4% | {Preterm premature rupture of the membranes, susceptibility to}, 610504;Osteogenesis imperfecta, type X, 613848 |
| SGPL1 | 96.6% | 96.6% | 100.0% | 99.1% | RENI syndrome, 617575 |
| SHOC2 | 100.0% | 100.0% | 100.0% | 97.1% | Noonan syndrome-like with loose anagen hair 1, 607721 |
| SKI | 100.0% | 99.9% | 99.7% | 92.7% | Shprintzen-Goldberg syndrome, 182212 |
| SKIC2 | 100.0% | 100.0% | 100.0% | 99.3% | Trichohepatoenteric syndrome 2, 614602 |
| SKIC3 | 98.9% | 98.9% | 100.0% | 98.3% | Trichohepatoenteric syndrome 1, 222470 |
| SLC17A9 | 100.0% | 100.0% | 100.0% | 98.9% | Porokeratosis 8, disseminated superficial actinic type, 616063 |

| | | | | | |
|---------|--------|--------|--------|-------|--|
| SLC24A4 | 100.0% | 100.0% | 100.0% | 98.9% | [Skin/hair/eye pigmentation 6, blond/brown hair], 210750;Amelogenesis imperfecta, type IIA5, 615887;[Skin/hair/eye pigmentation 6, blue/green eyes], 210750 |
| SLC24A5 | 100.0% | 99.6% | 100.0% | 98.7% | [Skin/hair/eye pigmentation 4, fair/dark skin], 113750;Albinism, oculocutaneous, type VI, 113750 |
| SLC26A2 | 100.0% | 100.0% | 100.0% | 98.3% | 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 |
| SLC27A4 | 100.0% | 100.0% | 100.0% | 99.2% | Ichthyosis prematurity syndrome, 608649 |
| SLC29A3 | 100.0% | 100.0% | 100.0% | 99.5% | Histiocytosis-lymphadenopathy plus syndrome, 602782 |
| SLC2A10 | 100.0% | 100.0% | 100.0% | 99.3% | Arterial tortuosity syndrome, 208050 |

| | | | | | |
|----------|--------|--------|--------|-------|--|
| SLC39A13 | 100.0% | 100.0% | 100.0% | 99.3% | Ehlers-Danlos syndrome, spondylodysplastic type, 3, 612350 |
| SLC39A4 | 100.0% | 100.0% | 100.0% | 99.5% | Acrodermatitis enteropathica, 201100 |
| SLC45A2 | 100.0% | 100.0% | 100.0% | 99.7% | [Skin/hair/eye pigmentation 5, dark/light eyes], 227240;[Skin/hair/eye pigmentation 5, black/nonblack hair], 227240;Albinism, oculocutaneous, type IV, 606574;[Skin/hair/eye pigmentation 5, dark/fair skin], 227240 |
| SLC4A4 | 97.3% | 97.0% | 100.0% | 98.2% | Proximal renal tubular acidosis-ocular anomaly syndrome, 604278 |
| SLC6A19 | 100.0% | 100.0% | 100.0% | 99.4% | Hartnup disorder, 234500 |
| SLC7A7 | 100.0% | 100.0% | 100.0% | 98.8% | Lysinuric protein intolerance, 222700 |
| SLCO2A1 | 100.0% | 100.0% | 100.0% | 99.0% | Hypertrophic osteoarthropathy, primary, autosomal dominant, 167100;PHOAR2-enteropathy syndrome, 614441 |
| SLURP1 | 100.0% | 100.0% | 100.0% | 99.5% | Meleda disease, 248300 |
| SLX4 | 100.0% | 100.0% | 100.0% | 99.0% | Fanconi anemia, complementation group P, 613951 |

| | | | | | |
|----------|--------|--------|--------|-------|--|
| SMAD3 | 100.0% | 100.0% | 100.0% | 96.8% | Loeys-Dietz syndrome 3, 613795 |
| SMARCA2 | 98.0% | 97.9% | 100.0% | 98.8% | Nicolaiides-Baraitser syndrome, 601358;Blepharophimosis-impaired intellectual development syndrome, 619293 |
| SMARCA4 | 100.0% | 100.0% | 100.0% | 99.6% | Coffin-Siris syndrome 4, 614609;{Rhabdoid tumor predisposition syndrome 2}, 613325;?Otosclerosis 12, 620792 |
| SMARCAD1 | 100.0% | 100.0% | 100.0% | 97.9% | Basan syndrome, 129200;Huriez syndrome, 181600;Adermatoglyphia, 136000 |
| SMARCAL1 | 100.0% | 100.0% | 100.0% | 98.8% | Schimke immunoosseous dysplasia, 242900 |
| SMARCB1 | 100.0% | 100.0% | 100.0% | 98.2% | Rhabdoid tumors, somatic, 609322;{Schwannomatosis-1, susceptibility to}, 162091;Coffin-Siris syndrome 3, 614608;{Rhabdoid tumor predisposition syndrome 1}, 609322 |
| SMO | 100.0% | 100.0% | 100.0% | 98.4% | Pallister-Hall-like syndrome, 241800;Basal cell carcinoma, somatic, 605462;Curry-Jones syndrome, somatic mosaic, 601707 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| SMOC2 | 100.0% | 100.0% | 100.0% | 98.6% | Dentin dysplasia, type I, with microdontia and misshapen teeth, 125400 |
| SNAI2 | 100.0% | 100.0% | 100.0% | 99.1% | |
| SNAP29 | 100.0% | 100.0% | 100.0% | 96.8% | Cerebral dysgenesis, neuropathy, ichthyosis, and palmoplantar keratoderma syndrome, 609528 |
| SNRPE | 100.0% | 100.0% | 100.0% | 98.8% | Hypotrichosis 11, 615059 |
| SNX10 | 89.3% | 89.3% | 100.0% | 98.3% | Osteopetrosis, autosomal recessive 8, 615085 |
| SOS1 | 98.7% | 98.1% | 100.0% | 96.9% | Noonan syndrome 4, 610733;?Fibromatosis, gingival, 1, 135300 |
| SOX10 | 97.8% | 97.8% | 100.0% | 97.9% | Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 |
| SOX18 | 99.8% | 98.8% | 100.0% | 92.6% | Hypotrichosis-lymphedema-telangiectasia syndrome, 607823;Hypotrichosis-lymphedema-telangiectasia-renal defect syndrome, 137940 |
| SOX2 | 100.0% | 100.0% | 99.9% | 95.2% | Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| SP7 | 100.0% | 100.0% | 100.0% | 99.2% | Osteogenesis imperfecta, type XII, 613849 |
| SPINK5 | 100.0% | 100.0% | 100.0% | 97.8% | Netherton syndrome, 256500 |
| SPINT2 | 100.0% | 100.0% | 100.0% | 98.8% | Diarrhea 3, secretory sodium, congenital, syndromic, 270420 |
| SPRED1 | 100.0% | 100.0% | 100.0% | 98.5% | Legius syndrome, 611431 |
| SPRY4 | 100.0% | 100.0% | 100.0% | 99.8% | Hypogonadotropic hypogonadism 17 with or without anosmia, 615266 |
| SRD5A3 | 100.0% | 100.0% | 100.0% | 97.6% | Kahrizi syndrome, 612713;Congenital disorder of glycosylation, type Iq, 612379 |
| SREBF1 | 100.0% | 100.0% | 99.9% | 97.9% | Ichthyosis, follicular, with atrichia and photophobia syndrome 2, 619016;Mucoepithelial dysplasia, hereditary, 158310 |
| ST14 | 100.0% | 100.0% | 100.0% | 99.2% | Ichthyosis, congenital, autosomal recessive 11, 602400 |
| ST3GAL5 | 98.3% | 98.3% | 100.0% | 97.8% | Salt and pepper developmental regression syndrome, 609056 |
| STAMPB | 96.3% | 96.3% | 100.0% | 99.0% | Microcephaly-capillary malformation syndrome, 614261 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| STAT3 | 100.0% | 100.0% | 100.0% | 98.0% | Hyper-IgE syndrome 1, autosomal dominant, with recurrent infections, 147060;Autoimmune disease, multisystem, infantile-onset, 1, 615952 |
| STAT5B | 100.0% | 100.0% | 100.0% | 98.8% | Growth hormone insensitivity with immune dysregulation 1, autosomal recessive, 245590;Growth hormone insensitivity with immune dysregulation 2, autosomal dominant, 618985;Leukemia, acute promyelocytic, somatic, 102578 |
| STIM1 | 100.0% | 99.6% | 100.0% | 99.0% | Myopathy, tubular aggregate, 1, 160565;Stormorken syndrome, 185070;Immunodeficiency 10, 612783 |
| STING1 | 100.0% | 100.0% | 100.0% | 97.3% | STING-associated vasculopathy, infantile-onset, 615934 |
| STK11 | 100.0% | 100.0% | 100.0% | 98.5% | Melanoma, malignant, somatic, 155600;Pancreatic cancer, somatic, 260350;Peutz-Jeghers syndrome, 175200;Testicular tumor, somatic, 273300 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| STK4 | 100.0% | 100.0% | 100.0% | 99.2% | T-cell immunodeficiency, recurrent infections, autoimmunity, and cardiac malformations, 614868 |
| STS | 96.9% | 96.5% | 98.2% | 72.4% | Ichthyosis, X-linked, 308100 |
| SUFU | 100.0% | 100.0% | 99.9% | 98.5% | {Meningioma, familial, susceptibility to}, 607174;Joubert syndrome 32, 617757;Basal cell nevus syndrome 2, 620343;{Medulloblastoma}, 155255 |
| SULT2B1 | 100.0% | 100.0% | 99.9% | 98.8% | Ichthyosis, congenital, autosomal recessive 14, 617571 |
| SUMF1 | 100.0% | 100.0% | 100.0% | 99.3% | Multiple sulfatase deficiency, 272200 |
| TALDO1 | 100.0% | 100.0% | 100.0% | 98.2% | Transaldolase deficiency, 606003 |
| TAP1 | 99.4% | 96.8% | 100.0% | 98.7% | MHC class I deficiency 1, 604571 |
| TAP2 | 97.9% | 97.9% | 100.0% | 98.4% | MHC class I deficiency 2, 620813 |
| TAPBP | 89.0% | 88.8% | 99.9% | 97.3% | ?MHC class I deficiency 3, 620814 |
| TAT | 100.0% | 100.0% | 100.0% | 98.8% | Tyrosinemia, type II, 276600 |

| | | | | | |
|---------|--------|--------|--------|-------|---|
| TBC1D24 | 100.0% | 100.0% | 100.0% | 99.6% | Deafness, autosomal recessive 86, 614617;Epilepsy, rolandic, with paroxysmal exercise-induce dystonia and writer's cramp, 608105;Myoclonic epilepsy, infantile, familial, 605021;Deafness, autosomal dominant 65, 616044;Developmental and epileptic encephalopathy 16, 615338;DOORS syndrome, 220500 |
| TBX3 | 100.0% | 100.0% | 100.0% | 98.2% | Ulnar-mammary syndrome, 181450 |
| TCHH | 100.0% | 100.0% | 99.7% | 88.2% | ?Uncombable hair syndrome 3, 617252 |
| TCIRG1 | 100.0% | 100.0% | 100.0% | 99.6% | Osteopetrosis, autosomal recessive 1, 259700 |
| TEK | 100.0% | 99.9% | 100.0% | 98.8% | Venous malformations, multiple cutaneous and mucosal, 600195;Glaucoma 3, primary congenital, E, 617272 |
| TERC | | | | | Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743;Dyskeratosis congenita, autosomal dominant 1, 127550 |
| TERF2IP | 99.7% | 96.0% | 100.0% | 97.9% | |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| TERT | 100.0% | 100.0% | 100.0% | 99.8% | Dyskeratosis congenita, autosomal dominant 2, 613989;Dyskeratosis congenita, autosomal recessive 4, 613989;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742;{Melanoma, cutaneous malignant, 9}, 615134;{Leukemia, acute myeloid}, 601626 |
| TFAP2A | 100.0% | 100.0% | 99.8% | 93.8% | Branchiooculofacial syndrome, 113620 |
| TGFB2 | 100.0% | 100.0% | 100.0% | 98.4% | Loeys-Dietz syndrome 4, 614816 |
| TGFBR1 | 100.0% | 100.0% | 100.0% | 96.7% | {Multiple self-healing squamous epithelioma, susceptibility to}, 132800;Loeys-Dietz syndrome 1, 609192 |
| TGFBR2 | 100.0% | 100.0% | 100.0% | 98.5% | Loeys-Dietz syndrome 2, 610168;Colorectal cancer, hereditary nonpolyposis, type 6, 614331;Esophageal cancer, somatic, 133239 |
| TGM1 | 100.0% | 100.0% | 100.0% | 99.5% | Ichthyosis, congenital, autosomal recessive 1, 242300 |
| TGM3 | 100.0% | 100.0% | 100.0% | 99.2% | ?Uncombable hair syndrome 2, 617251 |

| | | | | | |
|-----------|--------|--------|--------|-------|--|
| TGM5 | 100.0% | 100.0% | 100.0% | 98.8% | Peeling skin syndrome 2, 609796 |
| TINF2 | 100.0% | 100.0% | 100.0% | 98.4% | Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130 |
| TMC6 | 100.0% | 100.0% | 100.0% | 99.3% | {Epidermodysplasia verruciformis, susceptibility to, 1}, 226400 |
| TMC8 | 100.0% | 100.0% | 100.0% | 99.2% | {Epidermodysplasia verruciformis, susceptibility to, 2}, 618231 |
| TMEM165 | 100.0% | 100.0% | 100.0% | 97.8% | Congenital disorder of glycosylation, type IIk, 614727 |
| TNFRSF11A | 100.0% | 99.6% | 99.9% | 98.3% | Osteopetrosis, autosomal recessive 7, 612301;{Paget disease of bone 2, early-onset}, 602080;Osteolysis, familial expansile, 174810 |
| TNFRSF11B | 100.0% | 100.0% | 100.0% | 98.7% | Paget disease of bone 5, juvenile-onset, 239000 |
| TNFRSF1A | 100.0% | 100.0% | 100.0% | 99.7% | {Multiple sclerosis, susceptibility to, 5}, 614810;Periodic fever, familial, 142680 |
| TNFSF11 | 100.0% | 100.0% | 100.0% | 98.6% | Osteopetrosis, autosomal recessive 2, 259710 |

| | | | | | |
|-------|--------|--------|--------|-------|---|
| TNXB | 100.0% | 100.0% | 100.0% | 98.9% | Ehlers-Danlos syndrome, classic-like, 1, 606408;Vesicoureteral reflux 8, 615963 |
| TP63 | 100.0% | 99.9% | 100.0% | 99.3% | 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 |
| TPCN2 | 100.0% | 100.0% | 100.0% | 99.5% | [Skin/hair/eye pigmentation 10, blond/brown hair], 612267 |
| TREX1 | 100.0% | 100.0% | 100.0% | 99.8% | Vasculopathy, retinal, with cerebral leukoencephalopathy and systemic manifestations, 192315;Aicardi-Goutieres syndrome 1, dominant and recessive, 225750;{Systemic lupus erythematosus, susceptibility to}, 152700;Chilblain lupus, 610448 |

| | | | | | |
|--------|--------|--------|--------|-------|--|
| TRIM32 | 100.0% | 100.0% | 100.0% | 99.9% | ?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110 |
| TRIM37 | 98.3% | 98.3% | 100.0% | 98.5% | Mulibrey nanism, 253250 |
| TRPM4 | 100.0% | 100.0% | 100.0% | 98.9% | Progressive familial heart block, type IB, 604559;Erythrokeratoderma variabilis et progressiva 6, 618531 |
| TRPS1 | 100.0% | 99.9% | 100.0% | 98.4% | Trichorhinophalangeal syndrome, type III, 190351;Trichorhinophalangeal syndrome, type I, 190350 |
| TRPV3 | 100.0% | 100.0% | 100.0% | 99.1% | ?Palmoplantar keratoderma, nonepidermolytic, focal 2, 616400;Olmsted syndrome 1, 614594 |
| TSC1 | 100.0% | 100.0% | 100.0% | 98.8% | Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-1, 191100;Lymphangiomyomatosis, 606690 |
| TSC2 | 100.0% | 100.0% | 100.0% | 99.5% | Lymphangiomyomatosis, somatic, 606690;?Focal cortical dysplasia, type II, somatic, 607341;Tuberous sclerosis-2, 613254 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| TSPEAR | 100.0% | 100.0% | 100.0% | 98.9% | Tooth agenesis, selective, 10, 620173;?Deafness, autosomal recessive 98, 614861;Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis, 618180 |
| TTI2 | 100.0% | 100.0% | 100.0% | 98.5% | Intellectual developmental disorder, autosomal recessive 39, 615541 |
| TUFT1 | 100.0% | 100.0% | 100.0% | 99.2% | Woolly hair-skin fragility syndrome, 620415 |
| TWIST2 | 100.0% | 100.0% | 100.0% | 94.3% | Ablepharon-macrostomia syndrome, 200110;Barber-Say syndrome, 209885;Focal facial dermal dysplasia 3, Setleis type, 227260 |
| TYR | 100.0% | 99.9% | 100.0% | 98.8% | [Skin/hair/eye pigmentation 3, light/dark/freckling skin], 601800;[Skin/hair/eye pigmentation 3, blue/green eyes], 601800;{Melanoma, cutaneous malignant, susceptibility to, 8}, 601800;Albinism, oculocutaneous, type IB, 606952;Albinism, oculocutaneous, type IA, 203100 |

| | | | | | |
|-------|--------|--------|--------|-------|---|
| TYRP1 | 100.0% | 100.0% | 100.0% | 98.9% | [Skin/hair/eye pigmentation, variation in, 11 (Melanesian blond hair)], 612271;Albinism, oculocutaneous, type III, 203290 |
| UBE2A | 94.4% | 90.1% | 96.4% | 68.4% | Intellectual developmental disorder, X-linked syndromic, Nascimento type, 300860 |
| UBR1 | 98.0% | 98.0% | 100.0% | 98.2% | Johanson-Blizzard syndrome, 243800 |
| UROD | 100.0% | 100.0% | 100.0% | 99.2% | Porphyria, hepatoerythropoietic, 176100;Porphyria cutanea tarda, 176100 |
| UROS | 100.0% | 100.0% | 100.0% | 98.2% | Porphyria, congenital erythropoietic, 263700 |
| USB1 | 93.2% | 93.2% | 100.0% | 98.6% | Poikiloderma with neutropenia, 604173 |
| UVSSA | 100.0% | 100.0% | 100.0% | 99.2% | UV-sensitive syndrome 3, 614640 |
| VDR | 100.0% | 100.0% | 100.0% | 98.1% | Rickets, vitamin D-resistant, type IIA, 277440 |
| VEGFC | 100.0% | 100.0% | 100.0% | 98.3% | Lymphatic malformation 4, 615907 |

| | | | | | |
|--------|--------|--------|--------|-------|---|
| VHL | 88.0% | 87.9% | 100.0% | 99.3% | Erythrocytosis, familial, 2, 263400;von Hippel-Lindau syndrome, 193300;Renal cell carcinoma, somatic, 144700;Pheochromocytoma, 171300;Hemangioblastoma, cerebellar, somatic, |
| VPS13B | 100.0% | 99.8% | 100.0% | 98.7% | Cohen syndrome, 216550 |
| VPS33B | 100.0% | 100.0% | 100.0% | 98.5% | Keratoderma-ichthyosis-deafness syndrome, autosomal recessive, 620009;Cholestasis, progressive familial intrahepatic, 12, 620010;Arthrogryposis, renal dysfunction, and cholestasis 1, 208085 |
| WAS | 98.2% | 93.8% | 97.0% | 65.8% | Wiskott-Aldrich syndrome, 301000;Neutropenia, severe congenital, X-linked, 300299;Thrombocytopenia, X-linked, intermittent, 313900;Thrombocytopenia, X-linked, 313900 |
| WDR19 | 100.0% | 100.0% | 99.9% | 97.7% | 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 | 100.0% | 100.0% | 100.0% | 98.9% | Short-rib thoracic dysplasia 7 with or without polydactyly, 614091;Cranioectodermal dysplasia 2, 613610 |
| WDR72 | 96.8% | 96.8% | 100.0% | 98.3% | Amelogenesis imperfecta, type IIA3, 613211 |
| WIPF1 | 100.0% | 100.0% | 100.0% | 98.8% | Wiskott-Aldrich syndrome 2, 614493 |
| WNT10A | 100.0% | 100.0% | 100.0% | 99.5% | Schopf-Schulz-Passarge syndrome, 224750;Tooth agenesis, selective, 4, 150400;Ectodermal dysplasia 16 (odontoonychodermal dysplasia), 257980 |
| WNT10B | 100.0% | 100.0% | 100.0% | 99.0% | Tooth agenesis, selective, 8, 617073;Split-hand/foot malformation 6, 225300 |
| WNT5A | 100.0% | 100.0% | 100.0% | 97.7% | Robinow syndrome, autosomal dominant 1, 180700 |
| WNT7A | 100.0% | 100.0% | 100.0% | 99.1% | Fuhrmann syndrome, 228930;Ulna and fibula, absence of, with severe limb deficiency, 276820 |
| WRAP53 | 100.0% | 100.0% | 100.0% | 98.3% | Dyskeratosis congenita, autosomal recessive 3, 613988 |
| WRN | 100.0% | 100.0% | 100.0% | 97.7% | Werner syndrome, 277700 |

| | | | | | |
|----------|--------|--------|--------|-------|---|
| XPA | 100.0% | 100.0% | 100.0% | 97.7% | Xeroderma pigmentosum, group A, 278700 |
| XPC | 100.0% | 100.0% | 99.9% | 95.7% | Xeroderma pigmentosum, group C, 278720 |
| XYLT1 | 100.0% | 99.8% | 99.6% | 93.5% | Desbuquois dysplasia 2, 615777;{Pseudoxanthoma elasticum, modifier of severity of}, 264800 |
| XYLT2 | 99.9% | 99.2% | 100.0% | 98.9% | {Pseudoxanthoma elasticum, modifier of severity of}, 264800;Spondyloocular syndrome, 605822 |
| YWHAZ | 100.0% | 100.0% | 100.0% | 98.1% | |
| ZBTB20 | 100.0% | 100.0% | 100.0% | 99.4% | Primrose syndrome, 259050 |
| ZMPSTE24 | 100.0% | 100.0% | 100.0% | 98.7% | Mandibuloacral dysplasia with type B lipodystrophy, 608612;Restrictive dermopathy 1, 275210 |
| ZNF469 | 100.0% | 100.0% | 100.0% | 98.7% | Brittle cornea syndrome 1, 229200 |
| ZNF592 | 100.0% | 100.0% | 100.0% | 99.2% | |
| ZNF750 | 100.0% | 100.0% | 100.0% | 99.3% | ?Seborrhea-like dermatitis with psoriasiform elements, 610227 |

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.

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.

srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.

This list is accurate for panel version DG 4.0.0

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