DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL DG-5.2.0 (222 GENES)

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
OMIM disease identifiers and descriptions : October 25th, 2026.
This list is accurate for panel version DG 5.2.0
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
Ad 1. Blank field signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
- GQ 20: 99% confidence in the genotype call
- GQ 30: 99.9% confidence in the genotype call

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
AAAS	100% | 100%	97% | 97%	98% | 98%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS2	100% | 100%	97% | 97%	99% | 99%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
ABCD1	85% | 84%	98% | 98%	100% | 100%	Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100
ACTB	100% | 100%	97% | 97%	98% | 98%	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, 620470
ADCY3	100% | 100%	96% | 96%	98% | 97%	{Obesity, susceptibility to, BMIQ19}, 617885
AIRE	100% | 100%	96% | 96%	95% | 95%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKR1C2	86% | 84%	96% | 96%	99% | 99%	46XY sex reversal 8, 614279
AMH	100% | 100%	94% | 94%	97% | 97%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100% | 100%	96% | 96%	98% | 98%	Persistent Mullerian duct syndrome, type II, 261550
ANKRD31	100% | 100%	98% | 98%	99% | 99%	
ANOS1	99% | 99%	98% | 98%	100% | 100%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AR	97% | 97%	97% | 97%	99% | 99%	{Prostate cancer, hereditary, X-linked 3}, 301120;Androgen insensitivity, partial, with or without breast cancer, 312300;Spinal and bulbar muscular atrophy, X-linked 1, 313200;Androgen insensitivity, 300068;Hypospadias 1, X-linked, 300633

ARCN1	96% | 96%	97% | 97%	99% | 99%	Short stature-micrognathia syndrome, 617164
ARHGAP35	100% | 100%	98% | 98%	99% | 99%	
ARL6	100% | 100%	98% | 98%	100% | 100%	Retinitis pigmentosa 55, 613575;{Bardet-Biedl syndrome 1, modifier of}, 209900;Bardet-Biedl syndrome 3, 600151
ARMC5	100% | 100%	96% | 96%	98% | 98%	{ACTH-independent macronodular adrenal hyperplasia 2}, 615954
ARX	93% | 93%	96% | 96%	100% | 100%	Proud syndrome, 300004;Hydranencephaly with abnormal genitalia, 300215;Partington syndrome, 309510;Developmental and epileptic encephalopathy 1, 308350;Lissencephaly, X-linked 2, 300215;Intellectual developmental disorder, X-linked 29, 300419
ATF3	100% | 100%	97% | 97%	99% | 99%	
ATRX	100% | 100%	99% | 99%	100% | 100%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448;Intellectual disability-hypotonic facies syndrome, X-linked, 309580;Alpha-thalassemia/impaired intellectual development syndrome, 301040
AXL	100% | 100%	96% | 96%	99% | 99%	
B9D1	100% | 100%	96% | 96%	99% | 99%	?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120
BBS1	93% | 93%	94% | 94%	99% | 99%	Bardet-Biedl syndrome 1, 209900
BBS10	100% | 100%	97% | 97%	100% | 100%	Bardet-Biedl syndrome 10, 615987
BBS12	100% | 100%	98% | 98%	99% | 99%	Bardet-Biedl syndrome 12, 615989
BBS2	98% | 98%	98% | 98%	99% | 99%	Retinitis pigmentosa 74, 616562;Bardet-Biedl syndrome 2, 615981
BBS4	100% | 100%	97% | 97%	99% | 99%	Bardet-Biedl syndrome 4, 615982
BBS5	100% | 100%	98% | 98%	100% | 100%	Bardet-Biedl syndrome 5, 615983
BBS7	100% | 100%	98% | 98%	100% | 100%	Bardet-Biedl syndrome 7, 615984
BMP15	99% | 99%	99% | 99%	100% | 100%	Premature ovarian failure 4, 300510;Ovarian dysgenesis 2, 300510
BMP4	100% | 100%	97% | 97%	99% | 99%	Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932
BMP7	100% | 100%	95% | 95%	99% | 99%	
BNC1	100% | 100%	97% | 97%	99% | 99%	?Premature ovarian failure 16, 618723
C14orf39	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 52, 619202;Premature ovarian failure 18, 619203
CBX2	100% | 100%	96% | 96%	98% | 98%	?46XY sex reversal 5, 613080
CCDC141	100% | 100%	98% | 98%	100% | 100%	
CCNQ	98% | 98%	98% | 98%	99% | 99%	STAR syndrome, 300707
CDH2	100% | 100%	97% | 97%	99% | 99%	Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929

CDKN1C	99% | 99%	91% | 91%	97% | 97%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CEP41	100% | 100%	97% | 97%	99% | 99%	Joubert syndrome 15, 614464
CHD7	100% | 100%	98% | 98%	99% | 99%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CLPP	100% | 100%	96% | 96%	98% | 98%	Perrault syndrome 3, 614129
CNGA2	98% | 98%	97% | 97%	100% | 100%	
CREBBP	100% | 100%	97% | 97%	99% | 99%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849
CTU2	99% | 99%	94% | 94%	97% | 97%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUL4B	97% | 97%	99% | 99%	100% | 100%	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354
CUL7	100% | 100%	97% | 97%	99% | 99%	3-M syndrome 1, 273750
CYB5A	57% | 57%	96% | 96%	99% | 99%	Methemoglobinemia and ambiguous genitalia, 250790
CYP11A1	96% | 96%	97% | 97%	99% | 98%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100% | 100%	96% | 96%	99% | 99%	Aldosteronism, glucocorticoid-remediable, 103900;Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010
CYP11B2	99% | 99%	96% | 96%	98% | 98%	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400;Aldosterone to renin ratio raised;{Low renin hypertension, susceptibility to};Hypoaldosteronism, congenital, due to CMO II deficiency, 610600
CYP17A1	100% | 100%	96% | 96%	99% | 99%	17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100% | 100%	98% | 98%	100% | 100%	Aromatase deficiency, 613546
CYP21A2	99% | 99%	95% | 95%	98% | 98%	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910;Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910
DAP3	91% | 91%	97% | 97%	99% | 99%	Perrault syndrome 7, 621101
DCAF17	97% | 97%	97% | 97%	99% | 99%	Woodhouse-Sakati syndrome, 241080
DCC	100% | 100%	98% | 98%	100% | 100%	Mirror movements 1 and/or agenesis of the corpus callosum, 157600;Esophageal carcinoma, somatic, 133239;Colorectal cancer, somatic, 114500;Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542
DHCR7	95% | 95%	96% | 96%	99% | 99%	Smith-Lemli-Opitz syndrome, 270400
DHH	100% | 100%	97% | 97%	98% | 98%	46XY gonadal dysgenesis with minifascicular neuropathy, 607080;46XY sex reversal 7, 233420
DHX37	99% | 99%	95% | 95%	98% | 98%	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731;46XY sex reversal 11, 273250

DLK1	99% | 99%	96% | 96%	99% | 99%	
DMRT1	100% | 100%	96% | 96%	99% | 99%	
DMRT2	100% | 100%	97% | 97%	99% | 99%	Spondylocostal dysostosis 7, autosomal recessive, 621523
DUSP6	100% | 100%	97% | 97%	99% | 99%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DYNC2H1	100% | 100%	98% | 98%	99% | 99%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2I1	96% | 96%	97% | 97%	99% | 99%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
EIF2B5	100% | 100%	97% | 97%	99% | 99%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EIF4ENIF1	100% | 100%	96% | 96%	99% | 99%	
EMX2	100% | 100%	92% | 92%	99% | 99%	Schizencephaly, 269160
ERAL1	99% | 99%	97% | 97%	99% | 99%	Perrault syndrome 6, 617565
ERCC6	100% | 100%	98% | 98%	99% | 99%	UV-sensitive syndrome 1, 600630;Cerebrooculofacioskeletal 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
ESR1	100% | 100%	96% | 96%	99% | 99%	Breast cancer, somatic, 114480;{Migraine, susceptibility to}, 157300;Estrogen resistance, 615363;{Myocardial infarction, susceptibility to}, 608446
ESR2	100% | 100%	97% | 97%	99% | 99%	?Ovarian dysgenesis 8, 618187
FANCM	100% | 100%	98% | 98%	100% | 100%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FEZF1	100% | 100%	97% | 97%	99% | 99%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	100% | 100%	98% | 98%	99% | 99%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	100% | 100%	95% | 95%	99% | 99%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	99% | 98%	97% | 97%	99% | 99%	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% | 100%	96% | 96%	99% | 99%	Bent bone dysplasia syndrome, 614592;LADD syndrome 1, 149730;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410;Scaphocephaly and Axenfeld-Rieger anomaly;Jackson-Weiss syndrome, 123150;Gastric cancer, somatic, 613659;Craniofacial-skeletal-dermatologic dysplasia, 101600;Apert syndrome, 101200;Pfeiffer syndrome, 101600;Craniosynostosis, nonspecific;?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579;Beare-Stevenson cutis gyrata syndrome, 123790;Crouzon syndrome, 123500;Saethre-Chotzen syndrome, 101400
FIGLA	99% | 99%	96% | 96%	99% | 99%	Premature ovarian failure 6, 612310

FIGNL1	100% | 100%	98% | 98%	99% | 99%	
FLRT3	100% | 100%	98% | 98%	100% | 100%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FOXL2	100% | 100%	91% | 91%	99% | 99%	Premature ovarian failure 3, 608996;Blepharophimosis, epicanthus inversus, and ptosis, types 1 and 2, 110100
FRAS1	100% | 100%	97% | 97%	99% | 99%	Fraser syndrome 1, 219000
FREM2	100% | 100%	97% | 97%	100% | 100%	Fraser syndrome 2, 617666;Cryptophthalmos, unilateral or bilateral, isolated, 123570
FSHB	100% | 100%	97% | 97%	99% | 99%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	100% | 100%	97% | 97%	99% | 99%	Ovarian hyperstimulation syndrome, 608115;Ovarian dysgenesis 1, 233300
FZD2	100% | 100%	94% | 94%	98% | 98%	Omodysplasia 2, 164745
GALT	100% | 100%	97% | 97%	99% | 99%	Galactosemia, 230400
GATA4	100% | 100%	95% | 95%	99% | 99%	Tetralogy of Fallot, 187500;Atrial septal defect 2, 607941;Ventricular septal defect 1, 614429;Atrioventricular septal defect 4, 614430;?Testicular anomalies with or without congenital heart disease, 615542
GDF9	100% | 100%	97% | 97%	99% | 99%	Premature ovarian failure 14, 618014
GGPS1	100% | 100%	98% | 98%	99% | 99%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
GK	96% | 96%	99% | 99%	100% | 100%	Glycerol kinase deficiency, 307030
GLI2	100% | 100%	96% | 96%	99% | 99%	Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829
GNRH1	100% | 100%	98% | 98%	99% | 99%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	100% | 100%	98% | 98%	100% | 100%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GREB1L	100% | 100%	97% | 97%	99% | 99%	Deafness, autosomal dominant 80, 619274;Renal hypodysplasia/aplasia 3, 617805
GRIP1	100% | 100%	97% | 97%	99% | 99%	Fraser syndrome 3, 617667
HARS2	100% | 100%	97% | 97%	99% | 99%	Perrault syndrome 2, 614926
HESX1	100% | 100%	97% | 97%	98% | 98%	Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230
HFM1	100% | 100%	98% | 98%	99% | 99%	?Hydatidiform mole, recurrent, 5, 621720;Premature ovarian failure 9, 615724
HHAT	100% | 100%	96% | 96%	99% | 99%	Nivelon-Nivelon-Mabille syndrome, 600092
HOXA13	95% | 95%	78% | 78%	99% | 99%	Hand-foot-genital syndrome, 140000;?Guttmacher syndrome, 176305
HROB	96% | 96%	97% | 97%	99% | 99%	Ovarian dysgenesis 11, 620897
HS6ST1	98% | 98%	93% | 93%	99% | 99%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880

HSD17B3	100% | 100%	98% | 98%	99% | 99%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	100% | 100%	96% | 96%	100% | 100%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSD3B2	100% | 100%	97% | 97%	100% | 100%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSF2BP	100% | 100%	97% | 97%	99% | 99%	Premature ovarian failure 19, 619245
IFT172	100% | 100%	97% | 97%	99% | 99%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IFT27	96% | 94%	97% | 97%	99% | 99%	Bardet-Biedl syndrome 19, 615996
IFT74	100% | 100%	98% | 98%	99% | 99%	Bardet-Biedl syndrome 22, 617119;Spermatogenic failure 58, 619585;Joubert syndrome 40, 619582
IGSF10	100% | 100%	98% | 98%	100% | 100%	
IL17RD	100% | 100%	97% | 97%	99% | 99%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
INSL3	80% | 80%	95% | 94%	97% | 97%	Cryptorchidism, 219050
IRF6	100% | 100%	96% | 96%	100% | 100%	{Orofacial cleft 6}, 608864;Popliteal pterygium syndrome 1, 119500;van der Woude syndrome 1, 119300;Ectodermal dysplasia 19, hypohydrotic/tooth/nail type, 621749
KASH5	100% | 100%	97% | 96%	98% | 98%	Hydatidiform mole, recurrent, 6, 621721;Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548
KAT6B	100% | 100%	97% | 97%	99% | 99%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KISS1	100% | 100%	93% | 93%	98% | 98%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100% | 100%	96% | 96%	97% | 93%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837;?Precocious puberty, central, 1, 176400
KLB	100% | 100%	98% | 98%	99% | 99%	
LARS2	96% | 96%	97% | 97%	99% | 99%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LEP	100% | 100%	96% | 96%	99% | 99%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94% | 94%	97% | 97%	99% | 99%	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	99% | 99%	95% | 95%	97% | 97%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	100% | 100%	98% | 98%	99% | 99%	Leydig cell adenoma, somatic, with precocious puberty, 176410;Leydig cell hypoplasia with pseudohermaphroditism, 238320;Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320;Luteinizing hormone resistance, female, 238320;Precocious puberty, male, 176410
LHX1	98% | 98%	92% | 92%	99% | 99%	
LHX3	100% | 100%	96% | 96%	98% | 98%	Pituitary hormone deficiency, combined, 3, 221750

LIPA	95% | 95%	97% | 97%	99% | 99%	Wolman disease, 620151;Cholesteryl ester storage disease, 278000
LZTFL1	100% | 100%	98% | 98%	100% | 100%	Bardet-Biedl syndrome 17, 615994
MAMLD1	97% | 97%	98% | 98%	100% | 100%	Hypospadias 2, X-linked, 300758
MAP3K1	100% | 100%	96% | 96%	99% | 99%	46XY sex reversal 6, 613762
MC2R	100% | 100%	98% | 98%	100% | 100%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM8	94% | 94%	98% | 98%	100% | 100%	?Premature ovarian failure 10, 612885
MCM9	100% | 100%	97% | 97%	99% | 99%	Ovarian dysgenesis 4, 616185
MEI4	100% | 100%	98% | 98%	100% | 100%	
MKKS	100% | 100%	98% | 98%	100% | 100%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231
MKRN3	100% | 100%	97% | 97%	99% | 99%	Precocious puberty, central, 2, 615346
MRAP	100% | 100%	98% | 98%	99% | 99%	Glucocorticoid deficiency 2, 607398
MSH4	100% | 100%	98% | 98%	99% | 99%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420
MYRF	99% | 99%	95% | 95%	98% | 97%	Nanophthalmos 1, 600165;Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280
NDNF	99% | 99%	97% | 97%	99% | 99%	Hypogonadotropic hypogonadism 25 with anosmia, 618841
NEK1	100% | 100%	98% | 98%	99% | 99%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NNT	96% | 96%	98% | 98%	100% | 100%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	100% | 100%	96% | 96%	97% | 97%	Premature ovarian failure 5, 611548
NOS1	100% | 100%	96% | 96%	99% | 99%	
NR0B1	98% | 98%	97% | 97%	100% | 100%	Adrenal hypoplasia, congenital, 300200;46XY sex reversal 2, dosage-sensitive, 300018
NR2F2	100% | 100%	96% | 96%	99% | 99%	46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779
NR3C1	100% | 100%	98% | 98%	99% | 99%	Glucocorticoid resistance, 615962
NR3C2	98% | 98%	94% | 94%	99% | 99%	Pseudohypoaldosteronism type I, autosomal dominant, 177735;Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR5A1	94% | 93%	96% | 96%	98% | 98%	46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957
NSMF	99% | 99%	96% | 96%	97% | 96%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NTN1	100% | 100%	97% | 97%	99% | 99%	Mirror movements 4, 618264
OBSL1	99% | 99%	95% | 95%	98% | 98%	3-M syndrome 2, 612921

PBX1	100% | 100%	97% | 97%	100% | 100%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCSK1	100% | 100%	96% | 96%	99% | 99%	{Obesity, susceptibility to, BMIQ12}, 612362;Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955
PHF6	100% | 100%	99% | 99%	100% | 100%	Borjeson-Forssman-Lehmann syndrome, 301900
PLXNA1	100% | 100%	97% | 97%	99% | 98%	Dworschak-Punetha neurodevelopmental syndrome, 619955
PMM2	95% | 95%	97% | 97%	99% | 99%	Congenital disorder of glycosylation, type Ia, 212065
PNPLA6	100% | 100%	95% | 95%	98% | 98%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470
POLE	100% | 100%	97% | 97%	99% | 99%	{Colorectal cancer, susceptibility to, 12}, 615083;FILS syndrome, 615139;IMAGE-I syndrome, 618336
POLG	100% | 100%	97% | 97%	99% | 99%	Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459;Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662;Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700;Progressive external ophthalmoplegia, autosomal dominant 1, 157640;Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POLR3A	100% | 100%	97% | 97%	99% | 99%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	95% | 95%	97% | 97%	99% | 99%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POLR3GL	100% | 100%	96% | 96%	99% | 99%	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234
POMC	100% | 100%	95% | 95%	99% | 99%	{Obesity, early-onset, susceptibility to}, 601665;Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POR	95% | 94%	96% | 96%	97% | 96%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PPP1R12A	100% | 100%	98% | 98%	100% | 100%	Genitourinary and/or/brain malformation syndrome, 618820
PPP2R3C	100% | 100%	98% | 98%	99% | 99%	Spermatogenic failure 36, 618420;Myoectodermal gonadal dysgenesis syndrome, 618419
PROK2	100% | 100%	98% | 98%	99% | 99%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100% | 100%	97% | 97%	99% | 99%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	99% | 99%	96% | 96%	98% | 98%	Pituitary hormone deficiency, combined, 2, 262600
PSMC3IP	100% | 100%	97% | 97%	99% | 99%	Ovarian dysgenesis 3, 614324

RIPK4	99% | 99%	96% | 96%	99% | 99%	CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RNF111	98% | 98%	97% | 97%	99% | 99%	
RNF216	100% | 100%	98% | 98%	99% | 99%	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840
ROR2	100% | 100%	96% | 96%	99% | 99%	Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive 1, 268310
RPL10	99% | 99%	98% | 98%	98% | 98%	{Autism, susceptibility to, X-linked 5}, 300847;Intellectual developmental disorder, X-linked syndromic 35, 300998
RSPO1	99% | 99%	97% | 97%	99% | 99%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644;Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RXFP2	100% | 100%	98% | 98%	99% | 99%	
SAMD9	100% | 100%	98% | 98%	100% | 100%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SDCCAG8	100% | 100%	97% | 97%	99% | 99%	Senior-Loken syndrome 7, 613615;Bardet-Biedl syndrome 16, 615993
SEMA3A	100% | 100%	98% | 98%	100% | 100%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SEMA3E	100% | 100%	97% | 97%	99% | 99%	
SGPL1	96% | 96%	97% | 97%	99% | 99%	RENI syndrome, 617575
SOHLH1	99% | 99%	95% | 95%	97% | 97%	Ovarian dysgenesis 5, 617690;Spermatogenic failure 32, 618115
SOX10	98% | 98%	95% | 95%	99% | 99%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX11	100% | 100%	93% | 93%	99% | 99%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866
SOX2	99% | 99%	95% | 95%	99% | 99%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900
SOX3	97% | 97%	95% | 95%	100% | 100%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123;Panhypopituitarism, X-linked, 312000
SOX9	98% | 98%	94% | 94%	98% | 98%	Campomelic dysplasia with autosomal sex reversal, 114290;46XY sex reversal 10, 616425;Acampomelic campomelic dysplasia, 114290;Campomelic dysplasia, 114290;46XX sex reversal 2, 278850
SPATA22	100% | 100%	97% | 97%	99% | 99%	Premature ovarian failure 25, 621002;Spermatogenic failure 96, 621001
SPIDR	100% | 100%	97% | 97%	99% | 99%	Ovarian dysgenesis 9, 619665
SPRY4	100% | 100%	97% | 97%	99% | 99%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SRCAP	100% | 100%	97% | 97%	99% | 99%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595;Floating-Harbor syndrome, 136140

SRD5A2	88% | 88%	97% | 97%	99% | 99%	Pseudovaginal perineoscrotal hypospadias, 264600
SRY	89% | 89%	100% | 100%	100% | 100%	46XY sex reversal 1, 400044;46XX sex reversal 1, 400045
STAG3	100% | 100%	96% | 96%	99% | 99%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723
STAR	100% | 100%	97% | 97%	99% | 99%	Lipoid adrenal hyperplasia, 201710
SWSAP1	100% | 100%	97% | 97%	98% | 98%	
SYCE1	99% | 99%	96% | 96%	99% | 99%	Premature ovarian failure 12, 616947;Spermatogenic failure 15, 616950
SYCP2L	100% | 100%	97% | 97%	99% | 99%	Premature ovarian failure 24, 620840
TAC3	100% | 100%	96% | 96%	99% | 99%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	100% | 100%	97% | 97%	100% | 100%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TBX19	100% | 100%	97% | 96%	99% | 99%	Adrenocorticotropic hormone deficiency, 201400
TBX3	100% | 100%	95% | 95%	99% | 99%	Ulnar-mammary syndrome, 181450
TCF12	96% | 96%	98% | 98%	99% | 99%	Craniosynostosis 3, 615314;Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TCTN3	100% | 100%	98% | 98%	99% | 99%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TENM1	99% | 99%	99% | 99%	100% | 100%	
TOE1	100% | 100%	97% | 97%	99% | 99%	Pontocerebellar hypoplasia, type 7, 614969
TP63	100% | 100%	98% | 98%	100% | 100%	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
TRIM32	100% | 100%	97% | 97%	99% | 99%	?Bardet-Biedl syndrome 11, 615988;Muscular dystrophy, limb-girdle, autosomal recessive 8, 254110
TSPYL1	100% | 100%	95% | 94%	99% | 99%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TTC8	91% | 91%	98% | 98%	100% | 100%	Bardet-Biedl syndrome 8, 615985;?Retinitis pigmentosa 51, 613464
TWNK	100% | 100%	97% | 97%	99% | 99%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138
TXNRD2	100% | 100%	96% | 96%	98% | 98%	?Glucocorticoid deficiency 5, 617825
WDPCP	92% | 92%	98% | 98%	100% | 100%	Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085
WDR11	100% | 100%	98% | 98%	99% | 99%	Intellectual developmental disorder, autosomal recessive 78, 620237;Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WNT4	100% | 100%	96% | 96%	99% | 99%	Mullerian aplasia and hyperandrogenism, 158330;SERKAL syndrome, 611812

WT1	99% | 99%	96% | 96%	99% | 99%	Mesothelioma, somatic, 156240;Meacham syndrome, 608978;Frasier syndrome, 136680;Nephrotic syndrome, type 4, 256370;Denys-Drash syndrome, 194080;Wilms tumor, type 1, 194070
ZFPM2	100% | 100%	98% | 98%	100% | 100%	Diaphragmatic hernia 3, 610187;46XY sex reversal 9, 616067;Tetralogy of Fallot, 187500
ZNF541	100% | 100%	97% | 97%	98% | 98%	
ZSWIM7	85% | 83%	97% | 97%	99% | 99%	Spermatogenic failure 71, 619831;?Ovarian dysgenesis 10, 619834