DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY PANEL DG-5.1.0 (207 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
AAAS	100%	100%	100%	100%	99.7%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS2	100%	100%	100%	100%	99.5%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
ABCD1	89.3%	86.3%	98.7%	89.2%	70.9%	Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100
ACTB	100%	100%	100%	99.9%	99.4%	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%	100%	100%	99.5%	{Obesity, susceptibility to, BMIQ19}, 617885

AIRE	100%	100%	100%	99.8%	99.1%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKR1C2	96.7%	92.5%	99.9%	99.7%	98.6%	46XY sex reversal 8, 614279
AMH	100%	100%	100%	99.9%	99%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100%	100%	100%	100%	99.4%	Persistent Mullerian duct syndrome, type II, 261550
ANKRD31	100%	100%	100%	99.9%	99.8%	
ANOS1	100%	100%	98.7%	89.8%	71.3%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AR	100%	100%	98.4%	87.6%	66.9%	Androgen insensitivity, partial, with or without breast cancer, 312300;Spinal and bulbar muscular atrophy, X-linked 1, 313200;{Prostate cancer, susceptibility to}, 301120;Androgen insensitivity, 300068;Hypospadias 1, X-linked, 300633
ARCN1	96.3%	96.1%	100%	100%	99.7%	Short stature-micrognathia syndrome, 617164
ARHGAP35	100%	100%	100%	100%	99.5%	
ARMC5	100%	100%	100%	100%	99.4%	{ACTH-independent macronodular adrenal hyperplasia 2}, 615954

ARX	99.9%	98.4%	96.3%	80.9%	59.5%	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%	100%	100%	99.6%	
ATRX	100%	100%	99.1%	91.6%	74.3%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448;Intellectual disability-hypotonic facies syndrome, X-linked, 309580;Alpha-thalassemia/impaired intellectual development syndrome, 301040
AXL	100%	100%	100%	100%	99.1%	
B9D1	100%	100%	100%	100%	99.4%	?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120
BMP15	100%	100%	98.6%	90.2%	70.4%	Premature ovarian failure 4, 300510;Ovarian dysgenesis 2, 300510
BMP4	100%	100%	100%	100%	99.5%	Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932
BMP7	100%	100%	100%	100%	99%	
BNC1	100%	100%	100%	100%	99.8%	?Premature ovarian failure 16, 618723
C14orf39	100%	100%	100%	100%	99.7%	Spermatogenic failure 52, 619202;Premature ovarian failure 18, 619203

CBX2	100%	100%	100%	100%	99.6%	?46XY sex reversal 5, 613080
CCDC141	100%	100%	100%	100%	99.8%	
CCNQ	96.7%	96.7%	99.5%	90.1%	70.1%	STAR syndrome, 300707
CDH2	100%	100%	100%	100%	99.7%	Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CDKN1C	100%	100%	100%	100%	99%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CEP41	100%	100%	100%	100%	99.8%	Joubert syndrome 15, 614464
CHD7	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CLPP	100%	100%	100%	100%	98.9%	Perrault syndrome 3, 614129
CNGA2	100%	99.9%	98%	87.1%	68.1%	
CREBBP	100%	100%	100%	99.9%	99.5%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849
CTU2	100%	100%	100%	100%	99.2%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142
CUL4B	96.7%	96.7%	99.2%	91.3%	74%	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354

CUL7	100%	100%	100%	100%	99.4%	3-M syndrome 1, 273750
CYB5A	57.7%	57.7%	100%	100%	99.9%	Methemoglobinemia and ambiguous genitalia, 250790
CYP11A1	100%	98%	100%	100%	99.3%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100%	100%	100%	100%	99.4%	Aldosteronism, glucocorticoid-remediable, 103900;Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010
CYP11B2	100%	100%	100%	100%	99.3%	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%	100%	100%	99.5%	17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100%	100%	100%	100%	99.8%	Aromatase deficiency, 613546
CYP21A2	100%	100%	100%	100%	99.4%	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910;Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910
DAP3	91.7%	90.8%	100%	100%	99.6%	Perrault syndrome 7, 621101
DCAF17	96.5%	96.5%	100%	99.9%	99.7%	Woodhouse-Sakati syndrome, 241080

DCC	100%	100%	100%	100%	99.7%	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	96.2%	96.2%	100%	100%	99.6%	Smith-Lemli-Opitz syndrome, 270400
DHH	100%	100%	100%	100%	99.7%	46XY gonadal dysgenesis with minifascicular neuropathy, 607080;46XY sex reversal 7, 233420
DHX37	100%	100%	100%	100%	99.4%	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731;46XY sex reversal 11, 273250
DLK1	100%	100%	100%	99.9%	98.9%	
DMRT1	100%	100%	100%	100%	99.5%	
DMRT2	100%	100%	100%	100%	99.7%	Spondylocostal dysostosis 7, autosomal recessive, 621523
DUSP6	100%	100%	100%	100%	99.9%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DYNC2H1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
DYNC2I1	95.7%	95.7%	100%	100%	99.5%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503

EIF2B5	100%	100%	100%	100%	99.6%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EIF4ENIF1	100%	100%	100%	100%	99.7%	
EMX2	100%	100%	100%	99.7%	96%	Schizencephaly, 269160
ERAL1	100%	100%	100%	99.9%	99.3%	Perrault syndrome 6, 617565
ERCC6	100%	100%	100%	100%	99.6%	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%	100%	100%	99.7%	Breast cancer, somatic, 114480;{Migraine, susceptibility to}, 157300;Estrogen resistance, 615363;{Myocardial infarction, susceptibility to}, 608446
ESR2	100%	100%	100%	100%	99.8%	?Ovarian dysgenesis 8, 618187
FANCM	100%	100%	100%	100%	99.8%	Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FEZF1	100%	100%	100%	99.9%	99.6%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030

FGF17	100%	100%	100%	100%	99.5%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	100%	100%	100%	99.8%	98.3%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100%	99.2%	100%	100%	99.4%	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%	100%	100%	99.7%	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	100%	100%	100%	100%	99.3%	Premature ovarian failure 6, 612310
FIGNL1	100%	100%	100%	100%	99.9%	
FLRT3	100%	100%	100%	100%	99.9%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FOXL2	100%	100%	99.9%	99.4%	96.8%	Premature ovarian failure 3, 608996;Blepharophimosis, epicanthus inversus, and ptosis, types 1 and 2, 110100
FRAS1	100%	100%	100%	100%	99.7%	Fraser syndrome 1, 219000

FREM2	100%	100%	100%	100%	99.7%	Fraser syndrome 2, 617666;Cryptophthalmos, unilateral or bilateral, isolated, 123570
FSHB	100%	100%	100%	100%	99.6%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	100%	100%	100%	100%	99.8%	Ovarian hyperstimulation syndrome, 608115;Ovarian dysgenesis 1, 233300
FZD2	100%	100%	100%	99.9%	98.6%	Omodysplasia 2, 164745
GALT	100%	100%	100%	100%	99.5%	Galactosemia, 230400
GATA4	100%	100%	100%	99.9%	98.2%	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%	100%	100%	99.9%	Premature ovarian failure 14, 618014
GGPS1	100%	100%	100%	100%	99.9%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
GK	96%	96%	99.5%	92.1%	73.6%	Glycerol kinase deficiency, 307030
GLI2	100%	100%	100%	99.9%	99.3%	Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829
GNRH1	100%	100%	100%	100%	99.8%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841

GNRHR	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GREB1L	100%	100%	100%	100%	99.7%	Deafness, autosomal dominant 80, 619274;Renal hypodysplasia/aplasia 3, 617805
GRIP1	100%	100%	100%	100%	99.9%	Fraser syndrome 3, 617667
HARS2	100%	100%	100%	99.9%	98.9%	Perrault syndrome 2, 614926
HESX1	100%	100%	100%	100%	99.7%	Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230
HFM1	100%	100%	100%	100%	99.7%	Premature ovarian failure 9, 615724
HHAT	100%	100%	100%	100%	99.6%	Nivelon-Nivelon-Mabille syndrome, 600092
HOXA13	100%	99.8%	99.6%	96.3%	86%	Hand-foot-genital syndrome, 140000;?Guttmacher syndrome, 176305
HROB	95.2%	95.2%	100%	100%	99.4%	Ovarian dysgenesis 11, 620897
HS6ST1	100%	100%	100%	100%	98.6%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HSD17B3	100%	100%	100%	100%	99.8%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	100%	100%	100%	100%	99.9%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400

HSD3B2	100%	100%	100%	100%	99.6%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSF2BP	100%	100%	100%	100%	99.9%	Premature ovarian failure 19, 619245
IFT172	100%	100%	100%	100%	99.7%	Retinitis pigmentosa 71, 616394;Bardet-Biedl syndrome 20, 619471;Short-rib thoracic dysplasia 10 with or without polydactyly, 615630
IGSF10	100%	100%	100%	100%	99.8%	
IL17RD	100%	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
INSL3	78.8%	78.8%	100%	100%	99.1%	Cryptorchidism, 219050
IRF6	100%	100%	100%	100%	99.4%	{Orofacial cleft 6}, 608864;Popliteal pterygium syndrome 1, 119500;van der Woude syndrome 1, 119300
KASH5	100%	100%	100%	100%	99.5%	Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548
KAT6B	100%	100%	100%	100%	99.5%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KISS1	100%	100%	100%	100%	99.5%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100%	100%	100%	100%	98.8%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837;?Precocious puberty, central, 1, 176400
KLB	100%	100%	100%	100%	99.8%	

LARS2	96.2%	96.2%	100%	100%	99.8%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LEP	100%	100%	100%	100%	99.5%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94.6%	94.6%	100%	100%	99.7%	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	100%	100%	100%	99.8%	98%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	100%	100%	100%	100%	99.7%	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	100%	100%	100%	99.9%	98.5%	
LHX3	100%	100%	100%	99.6%	98.9%	Pituitary hormone deficiency, combined, 3, 221750
LIPA	95.6%	95.2%	100%	100%	99.9%	Wolman disease, 620151;Cholesteryl ester storage disease, 278000
MAMLD1	100%	100%	98.8%	89.1%	70.8%	Hypospadias 2, X-linked, 300758
MAP3K1	100%	100%	100%	99.9%	99.5%	46XY sex reversal 6, 613762

MC2R	100%	100%	100%	100%	99.2%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM8	94.4%	94.4%	100%	100%	99.8%	?Premature ovarian failure 10, 612885
MCM9	100%	100%	100%	100%	99.8%	Ovarian dysgenesis 4, 616185
MEI4	100%	100%	100%	100%	99.9%	
MKKS	100%	100%	100%	100%	100%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231
MKRN3	100%	100%	100%	100%	99.7%	Precocious puberty, central, 2, 615346
MRAP	100%	100%	100%	100%	99.6%	Glucocorticoid deficiency 2, 607398
MSH4	100%	100%	100%	100%	99.8%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420
MYRF	100%	99.8%	100%	99.9%	99.2%	Nanophthalmos 1, 600165;Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280
NDNF	100%	100%	100%	100%	99.8%	Hypogonadotropic hypogonadism 25 with anosmia, 618841
NEK1	100%	100%	100%	100%	99.8%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892

NNT	96.3%	96.3%	100%	100%	99.8%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	100%	100%	100%	99.9%	99.1%	Premature ovarian failure 5, 611548
NOS1	100%	100%	100%	100%	99.2%	
NR0B1	100%	100%	98.6%	88%	68.3%	Adrenal hypoplasia, congenital, 300200;46XY sex reversal 2, dosage-sensitive, 300018
NR2F2	100%	100%	100%	100%	99.4%	46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779
NR3C1	100%	100%	100%	100%	99.8%	Glucocorticoid resistance, 615962
NR3C2	100%	100%	100%	99.8%	99.1%	Pseudohypoaldosteronism type I, autosomal dominant, 177735;Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR5A1	98.7%	95%	100%	100%	99.5%	46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957
NSMF	100%	100%	100%	100%	99.2%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NTN1	100%	100%	100%	100%	99.1%	Mirror movements 4, 618264
OBSL1	100%	100%	100%	99.8%	99.2%	3-M syndrome 2, 612921

PBX1	100%	100%	100%	100%	99.7%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCSK1	100%	100%	100%	100%	99.9%	{Obesity, susceptibility to, BMIQ12}, 612362;Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955
PHF6	100%	100%	99.4%	92.6%	74.6%	Borjeson-Forssman-Lehmann syndrome, 301900
PLXNA1	100%	100%	100%	100%	99.6%	Dworschak-Punetha neurodevelopmental syndrome, 619955
PMM2	94.6%	94.6%	100%	100%	99.7%	Congenital disorder of glycosylation, type Ia, 212065
PNPLA6	100%	100%	100%	100%	99.3%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470
POLE	100%	100%	100%	100%	99.3%	{Colorectal cancer, susceptibility to, 12}, 615083;FILS syndrome, 615139;IMAGE-I syndrome, 618336

POLG	100%	100%	100%	100%	99.8%	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%	100%	100%	99.6%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	95.4%	95.4%	100%	100%	99.7%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742
POLR3GL	100%	100%	100%	100%	99.5%	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234
POMC	100%	100%	100%	100%	99.7%	{Obesity, early-onset, susceptibility to}, 601665;Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734

POR	98%	94.9%	100%	100%	99.6%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PPP1R12A	100%	100%	100%	100%	99.8%	Genitourinary and/or/brain malformation syndrome, 618820
PPP2R3C	100%	100%	100%	100%	99.9%	Spermatogenic failure 36, 618420;Myoectodermal gonadal dysgenesis syndrome, 618419
PROK2	100%	100%	100%	100%	100%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100%	100%	100%	100%	99.3%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	100%	100%	100%	99.9%	99.1%	Pituitary hormone deficiency, combined, 2, 262600
PSMC3IP	100%	100%	100%	100%	99.5%	Ovarian dysgenesis 3, 614324
RIPK4	100%	100%	100%	100%	99.7%	CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
RNF111	97.5%	97.5%	100%	100%	99.6%	
ROR2	100%	100%	100%	100%	99.6%	Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive 1, 268310

RPL10	100%	100%	99.3%	91.1%	69.5%	{Autism, susceptibility to, X-linked 5}, 300847;Intellectual developmental disorder, X-linked syndromic 35, 300998
RSPO1	100%	100%	100%	100%	99.9%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644;Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
RXFP2	100%	100%	100%	100%	99.9%	
SAMD9	100%	100%	100%	100%	99.8%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SEMA3A	100%	100%	100%	100%	99.8%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SEMA3E	100%	100%	100%	100%	99.9%	
SGPL1	95.5%	95.5%	100%	100%	99.7%	RENI syndrome, 617575
SOHLH1	100%	100%	100%	100%	99.5%	Ovarian dysgenesis 5, 617690;Spermatogenic failure 32, 618115
SOX10	97.8%	97.8%	100%	100%	99.2%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584

SOX11	100%	100%	100%	99.7%	96%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866
SOX2	100%	100%	100%	99.9%	97.8%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900
SOX3	100%	100%	96.1%	81.1%	61.5%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123;Panhypopituitarism, X-linked, 312000
SOX9	100%	99.5%	100%	99.9%	97.9%	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%	100%	100%	99.6%	Premature ovarian failure 25, 621002;Spermatogenic failure 96, 621001
SPIDR	100%	100%	100%	100%	99.7%	Ovarian dysgenesis 9, 619665
SPRY4	100%	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SRCAP	100%	100%	100%	100%	99.3%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595;Floating-Harbor syndrome, 136140

SRD5A2	87.5%	87.5%	100%	100%	99.3%	Pseudovaginal perineoscrotal hypospadias, 264600
SRY	50%	50%	48.1%	37.3%	22.5%	46XY sex reversal 1, 400044;46XX sex reversal 1, 400045
STAG3	100%	100%	100%	100%	99.3%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723
STAR	100%	100%	100%	100%	99.4%	Lipoid adrenal hyperplasia, 201710
SWSAP1	100%	100%	100%	100%	99.1%	
SYCE1	100%	100%	100%	100%	99.3%	Premature ovarian failure 12, 616947;Spermatogenic failure 15, 616950
SYCP2L	100%	100%	100%	100%	99.7%	Premature ovarian failure 24, 620840
TAC3	100%	100%	100%	100%	99.4%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	100%	100%	100%	100%	99%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TBX19	100%	100%	100%	100%	99.6%	Adrenocorticotropic hormone deficiency, 201400
TBX3	100%	100%	100%	100%	99.2%	Ulnar-mammary syndrome, 181450
TCF12	96.1%	96.1%	100%	100%	99.7%	Craniosynostosis 3, 615314;Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TCTN3	100%	100%	100%	100%	99.5%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TENM1	100%	100%	99.2%	91%	72.1%	

TOE1	100%	100%	100%	99.9%	99.4%	Pontocerebellar hypoplasia, type 7, 614969
TP63	100%	100%	100%	100%	99.6%	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
TSPYL1	100%	100%	100%	100%	99.1%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TWNK	100%	100%	100%	100%	99.6%	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%	100%	100%	99.4%	?Glucocorticoid deficiency 5, 617825
WDPCP	92.1%	92.1%	100%	100%	99.8%	Bardet-Biedl syndrome 15, 615992;Congenital heart defects, hamartomas of tongue, and polysyndactyly, 217085

WDR11	100%	100%	100%	100%	99.9%	Intellectual developmental disorder, autosomal recessive 78, 620237;Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WNT4	100%	100%	100%	100%	99.1%	Mullerian aplasia and hyperandrogenism, 158330;SERKAL syndrome, 611812
WT1	100%	100%	100%	99.9%	99.3%	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%	100%	100%	99.8%	Diaphragmatic hernia 3, 610187;46XY sex reversal 9, 616067;Tetralogy of Fallot, 187500
ZNF541	100%	100%	100%	100%	99.5%	
ZSWIM7	91.9%	85.2%	100%	100%	99.6%	Spermatogenic failure 71, 619831;?Ovarian dysgenesis 10, 619834

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.
TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.
srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.
srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.
srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.
This list is accurate for panel version DG 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors