WES DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY DG 3.8.1

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
AAAS	100.0%	100.0%	100.0%	99.6%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS2	100.0%	100.0%	100.0%	99.9%	Leukoencephalopathy, progressive, with ovarian failure, 615889;Combined oxidative phosphorylation deficiency 8, 614096
ABCD1	100.0%	99.6%	99.5%	83.3%	Adrenoleukodystrophy, 300100;Adrenomyeloneuropathy, adult, 300100
ADCY3	100.0%	100.0%	100.0%	99.6%	{Obesity, susceptibility to, BMIQ19}, 617885
AIRE	100.0%	100.0%	100.0%	99.9%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKR1C2	100.0%	100.0%	100.0%	98.5%	46XY sex reversal 8, 614279
AMH	100.0%	100.0%	100.0%	100.0%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100.0%	100.0%	100.0%	99.7%	Persistent Mullerian duct syndrome, type II, 261550
ANOS1	100.0%	99.8%	99.1%	73.9%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700

AR	99.5%	99.0%	97.5%	68.8%	Androgen insensitivity, partial, with or without breast cancer, 312300;{Prostate cancer, susceptibility to}, 176807;Androgen insensitivity, 300068;Spinal and bulbar muscular atrophy of Kennedy, 313200;Hypospadias 1, X-linked, 300633
ARHGAP35	100.0%	100.0%	100.0%	99.4%	
ARMC5	100.0%	100.0%	100.0%	99.9%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARX	99.0%	96.7%	93.9%	63.3%	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	100.0%	100.0%	100.0%	97.3%	
ATRX	99.9%	99.7%	96.9%	68.4%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448;Intellectual disability-hypotonic facies syndrome, X-linked, 309580;Alpha-thalassemia/impaired intellectual development syndrome, 301040
AXL	100.0%	100.0%	100.0%	99.6%	
B9D1	100.0%	100.0%	100.0%	99.8%	?Meckel syndrome 9, 614209;Joubert syndrome 27, 617120
BMP15	100.0%	100.0%	98.4%	74.4%	Premature ovarian failure 4, 300510;Ovarian dysgenesis 2, 300510
BMP4	100.0%	100.0%	100.0%	99.7%	Orofacial cleft 11, 600625;Microphthalmia, syndromic 6, 607932

BMP7	100.0%	100.0%	100.0%	99.8%	
BNC1	100.0%	99.9%	100.0%	99.6%	?Premature ovarian failure 16, 618723
C14orf39	100.0%	100.0%	100.0%	98.4%	Spermatogenic failure 52, 619202;?Premature ovarian failure 18, 619203
CBX2	100.0%	100.0%	100.0%	99.7%	?46XY sex reversal 5, 613080
CCDC141	99.5%	98.9%	100.0%	99.1%	
CCDC155	100.0%	100.0%	100.0%	99.5%	Spermatogenic failure 88, 620547;Premature ovarian failure 22, 620548
CCNQ	100.0%	99.9%	99.5%	79.6%	STAR syndrome, 300707
CDH2	100.0%	100.0%	100.0%	99.5%	Arrhythmogenic right ventricular dysplasia 14, 618920;?Attention deficit-hyperactivity disorder 8, 619957;Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CDKN1C	100.0%	100.0%	100.0%	99.8%	IMAGE syndrome, 614732;Beckwith-Wiedemann syndrome, 130650
CEP41	100.0%	100.0%	100.0%	99.0%	Joubert syndrome 15, 614464
CHD7	100.0%	100.0%	100.0%	99.5%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370;CHARGE syndrome, 214800
CLPP	100.0%	100.0%	100.0%	99.3%	Perrault syndrome 3, 614129
CNGA2	99.9%	99.7%	98.6%	72.1%	
CREBBP	100.0%	100.0%	100.0%	99.2%	Menke-Hennekam syndrome 1, 618332;Rubinstein-Taybi syndrome 1, 180849
CTU2	100.0%	100.0%	100.0%	99.9%	Microcephaly, facial dysmorphism, renal agenesis, and ambiguous genitalia syndrome, 618142

CUL4B	100.0%	99.9%	98.1%	72.0%	Intellectual developmental disorder, X-linked syndromic, Cabezas type, 300354
CUL7	100.0%	100.0%	100.0%	99.5%	3-M syndrome 1, 273750
CYB5A	100.0%	100.0%	100.0%	99.8%	Methemoglobinemia and ambiguous genitalia, 250790
CYP11A1	100.0%	100.0%	100.0%	99.8%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100.0%	100.0%	100.0%	99.9%	Aldosteronism, glucocorticoid-remediable, 103900;Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010
CYP11B2	100.0%	100.0%	100.0%	99.9%	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400;Hypoaldosteronism, congenital, due to CMO II deficiency, 610600;Aldosterone to renin ratio raised, ;{Low renin hypertension, susceptibility to},
CYP17A1	100.0%	100.0%	100.0%	99.7%	17,20-lyase deficiency, isolated, 202110;17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100.0%	99.9%	100.0%	99.4%	Aromatase deficiency, 613546;Aromatase excess syndrome, 139300
CYP21A2	100.0%	99.9%	100.0%	99.6%	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910;Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910
DCAF17	100.0%	100.0%	100.0%	99.8%	Woodhouse-Sakati syndrome, 241080

DCC	100.0%	100.0%	100.0%	99.5%	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	100.0%	100.0%	100.0%	99.9%	Smith-Lemli-Opitz syndrome, 270400
DHH	100.0%	100.0%	100.0%	100.0%	46XY gonadal dysgenesis with minifascicular neuropathy, 607080;46XY sex reversal 7, 233420
DHX37	100.0%	100.0%	100.0%	99.8%	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731;46XY sex reversal 11, 273250
DLK1	100.0%	100.0%	100.0%	99.5%	
DMRT1	100.0%	100.0%	100.0%	99.9%	
DMRT2	100.0%	100.0%	100.0%	99.4%	
DUSP6	100.0%	100.0%	100.0%	99.3%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DYNC2H1	99.8%	99.4%	100.0%	99.1%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
EIF2B5	100.0%	100.0%	100.0%	99.4%	Leukoencephalopathy with vanishing white matter 5, with or without ovarian failure, 620315
EIF4ENIF1	100.0%	100.0%	100.0%	99.2%	
ERAL1	100.0%	100.0%	100.0%	99.7%	Perrault syndrome 6, 617565

ESR1	100.0%	99.8%	100.0%	99.4%	Breast cancer, somatic, 114480;{Migraine, susceptibility to}, 157300;Estrogen resistance, 615363;{Myocardial infarction, susceptibility to}, 608446
ESR2	100.0%	100.0%	100.0%	99.7%	?Ovarian dysgenesis 8, 618187
FANCM	100.0%	100.0%	100.0%	98.8%	?Premature ovarian failure 15, 618096;Spermatogenic failure 28, 618086
FEZF1	100.0%	100.0%	100.0%	99.0%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	100.0%	100.0%	100.0%	99.9%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	100.0%	100.0%	100.0%	99.7%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100.0%	100.0%	100.0%	99.8%	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%	99.5%	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,
FIGLA	100.0%	100.0%	100.0%	99.6%	Premature ovarian failure 6, 612310
FIGNL1	100.0%	100.0%	100.0%	99.2%	
FLRT3	100.0%	99.7%	100.0%	99.9%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FOXL2	100.0%	100.0%	100.0%	98.7%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100;Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100;Premature ovarian failure 3, 608996
FRAS1	100.0%	99.9%	100.0%	99.6%	Fraser syndrome 1, 219000
FREM2	99.9%	99.7%	100.0%	99.6%	Fraser syndrome 2, 617666;Cryptophthalmos, unilateral or bilateral, isolated, 123570
FSHB	98.7%	98.0%	100.0%	100.0%	Hypogonadotropic hypogonadism 24 without anosmia, 229070

FSHR	100.0%	99.9%	100.0%	99.7%	Ovarian response to FSH stimulation, 276400;Ovarian hyperstimulation syndrome, 608115;Ovarian dysgenesis 1, 233300
FZD2	100.0%	100.0%	100.0%	99.2%	Omodysplasia 2, 164745
GALT	100.0%	100.0%	100.0%	99.6%	Galactosemia, 230400
GATA4	100.0%	100.0%	100.0%	99.8%	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	100.0%	100.0%	100.0%	99.4%	?Premature ovarian failure 14, 618014
GGPS1	100.0%	100.0%	100.0%	99.3%	Muscular dystrophy, congenital hearing loss, and ovarian insufficiency syndrome, 619518
GK	100.0%	100.0%	98.2%	72.2%	Glycerol kinase deficiency, 307030
GLI2	100.0%	100.0%	100.0%	99.9%	Culler-Jones syndrome, 615849;Holoprosencephaly 9, 610829
GNRH1	100.0%	100.0%	100.0%	97.9%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	100.0%	100.0%	100.0%	99.8%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GREB1L	100.0%	100.0%	100.0%	99.6%	Deafness, autosomal dominant 80, 619274;Renal hypodysplasia/aplasia 3, 617805
GRIP1	100.0%	100.0%	100.0%	99.5%	Fraser syndrome 3, 617667
HARS2	100.0%	100.0%	100.0%	99.6%	Perrault syndrome 2, 614926

HESX1	100.0%	100.0%	100.0%	97.4%	Pituitary hormone deficiency, combined, 5, 182230;Septooptic dysplasia, 182230;Growth hormone deficiency with pituitary anomalies, 182230
HFM1	100.0%	100.0%	100.0%	98.2%	Premature ovarian failure 9, 615724
HOXA13	99.9%	98.8%	99.5%	86.4%	Hand-foot-genital syndrome, 140000;?Guttmacher syndrome, 176305
HROB	100.0%	100.0%	100.0%	99.7%	
HS6ST1	100.0%	100.0%	100.0%	99.6%	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HSD17B3	100.0%	100.0%	100.0%	99.6%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	96.6%	96.6%	100.0%	99.3%	D-bifunctional protein deficiency, 261515;Perrault syndrome 1, 233400
HSD3B2	99.6%	99.4%	100.0%	99.7%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
HSF2BP	100.0%	100.0%	100.0%	99.7%	Premature ovarian failure 19, 619245
IGSF10	100.0%	100.0%	100.0%	99.6%	
IL17RD	100.0%	100.0%	100.0%	99.8%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IRF6	100.0%	100.0%	100.0%	99.6%	{Orofacial cleft 6}, 608864;Popliteal pterygium syndrome 1, 119500;van der Woude syndrome 1, 119300
KAT6B	100.0%	100.0%	100.0%	99.3%	SBBYSS syndrome, 603736;Genitopatellar syndrome, 606170
KISS1	100.0%	100.0%	100.0%	95.9%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842

KISS1R	100.0%	100.0%	100.0%	99.9%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837;?Precocious puberty, central, 1, 176400
KLB	100.0%	100.0%	100.0%	99.7%	
LARS2	100.0%	100.0%	100.0%	99.5%	Perrault syndrome 4, 615300;Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LEP	100.0%	100.0%	100.0%	99.4%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94.6%	94.6%	100.0%	99.3%	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	100.0%	100.0%	100.0%	100.0%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	100.0%	100.0%	100.0%	99.6%	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	100.0%	100.0%	100.0%	99.1%	
LHX3	100.0%	100.0%	100.0%	99.8%	Pituitary hormone deficiency, combined, 3, 221750
LIPA	96.6%	95.2%	100.0%	99.1%	Wolman disease, 620151;Cholesteryl ester storage disease, 278000
MAMLD1	100.0%	99.8%	99.1%	77.4%	Hypospadias 2, X-linked, 300758
MAP3K1	100.0%	100.0%	100.0%	99.3%	46XY sex reversal 6, 613762
MC2R	100.0%	100.0%	100.0%	99.5%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200

MCM8	94.4%	94.4%	100.0%	99.6%	?Premature ovarian failure 10, 612885
MCM9	100.0%	100.0%	100.0%	99.1%	Ovarian dysgenesis 4, 616185
MKKS	100.0%	100.0%	100.0%	99.6%	McKusick-Kaufman syndrome, 236700;Bardet-Biedl syndrome 6, 605231
MKRN3	100.0%	100.0%	100.0%	99.8%	Precocious puberty, central, 2, 615346
MRAP	100.0%	100.0%	100.0%	99.9%	Glucocorticoid deficiency 2, 607398
MSH4	100.0%	100.0%	100.0%	99.3%	Premature ovarian failure 20, 619938;Spermatogenic failure 2, 108420
MYRF	100.0%	100.0%	100.0%	99.6%	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113;Cardiac-urogenital syndrome, 618280
NDNF	100.0%	100.0%	100.0%	99.0%	Hypogonadotropic hypogonadism 25 with anosmia, 618841
NEK1	100.0%	100.0%	100.0%	99.1%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520;?Orofaciodigital syndrome II, 252100;{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NNT	96.4%	96.3%	100.0%	99.7%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	100.0%	100.0%	100.0%	99.8%	Premature ovarian failure 5, 611548
NOS1	100.0%	100.0%	100.0%	99.6%	
NR0B1	100.0%	99.8%	99.7%	81.5%	Adrenal hypoplasia, congenital, 300200;46XY sex reversal 2, dosage-sensitive, 300018

NR2F2	100.0%	100.0%	100.0%	99.3%	46XX sex reversal 5, 618901;Congenital heart defects, multiple types, 4, 615779
NR3C1	100.0%	100.0%	100.0%	99.6%	Glucocorticoid resistance, 615962
NR3C2	100.0%	100.0%	100.0%	99.6%	Pseudohypoaldosteronism type I, autosomal dominant, 177735;Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR5A1	100.0%	100.0%	100.0%	99.6%	46XX sex reversal 4, 617480;Premature ovarian failure 7, 612964;46XY sex reversal 3, 612965;Adrenocortical insufficiency, 612964;Spermatogenic failure 8, 613957
NSMF	100.0%	100.0%	100.0%	99.9%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
NTN1	100.0%	100.0%	100.0%	99.6%	Mirror movements 4, 618264
OBSL1	100.0%	100.0%	100.0%	99.9%	3-M syndrome 2, 612921
PBX1	100.0%	99.9%	100.0%	99.6%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCSK1	100.0%	100.0%	100.0%	99.5%	{Obesity, susceptibility to, BMIQ12}, 612362;Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955
PHF6	100.0%	100.0%	98.9%	75.6%	Borjeson-Forssman-Lehmann syndrome, 301900
PLXNA1	100.0%	100.0%	100.0%	100.0%	Dworschak-Punetha neurodevelopmental syndrome, 619955

PMM2	100.0%	100.0%	100.0%	98.7%	Congenital disorder of glycosylation, type Ia, 212065
PNPLA6	100.0%	100.0%	100.0%	99.9%	Spastic paraplegia 39, autosomal recessive, 612020;Oliver-McFarlane syndrome, 275400;?Laurence-Moon syndrome, 245800;Boucher-Neuhauser syndrome, 215470
POLE	100.0%	100.0%	100.0%	99.7%	{Colorectal cancer, susceptibility to, 12}, 615083;FILS syndrome, 615139;IMAGE-I syndrome, 618336
POLG	100.0%	100.0%	100.0%	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	100.0%	100.0%	100.0%	99.5%	Wiedemann-Rautenstrauch syndrome, 264090;Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694
POLR3B	100.0%	99.9%	100.0%	99.0%	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism, 614381;Charcot-Marie-Tooth disease, demyelinating, type 1I, 619742

POLR3GL	100.0%	100.0%	100.0%	99.7%	Short stature, oligodontia, dysmorphic facies, and motor delay, 619234
POMC	100.0%	100.0%	100.0%	99.8%	{Obesity, early-onset, susceptibility to}, 601665;Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POR	100.0%	100.0%	100.0%	100.0%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750;Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PPP1R12A	99.9%	99.3%	100.0%	99.4%	Genitourinary and/or/brain malformation syndrome, 618820
PPP2R3C	100.0%	100.0%	100.0%	99.3%	Spermatogenic failure 36, 618420;Myoectodermal gonadal dysgenesis syndrome, 618419
PROK2	100.0%	100.0%	100.0%	99.7%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100.0%	100.0%	100.0%	99.8%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	100.0%	100.0%	99.9%	96.7%	Pituitary hormone deficiency, combined, 2, 262600
PSMC3IP	100.0%	100.0%	100.0%	99.6%	Ovarian dysgenesis 3, 614324
RIPK4	100.0%	100.0%	100.0%	99.9%	CHAND syndrome, 214350;Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
ROR2	100.0%	100.0%	100.0%	99.8%	Brachydactyly, type B1, 113000;Robinow syndrome, autosomal recessive, 268310

RSPO1	100.0%	100.0%	100.0%	99.9%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644;Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
SAMD9	100.0%	100.0%	100.0%	99.0%	Tumoral calcinosis, familial, normophosphatemic, 610455;Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041;MIRAGE syndrome, 617053
SEMA3A	100.0%	100.0%	100.0%	99.7%	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SEMA3E	100.0%	100.0%	100.0%	99.7%	
SGPL1	100.0%	100.0%	100.0%	99.5%	RENI syndrome, 617575
SOHLH1	100.0%	100.0%	100.0%	99.9%	Ovarian dysgenesis 5, 617690;Spermatogenic failure 32, 618115
SOX10	100.0%	100.0%	100.0%	99.9%	Waardenburg syndrome, type 4C, 613266;PCWH syndrome, 609136;Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX11	100.0%	100.0%	100.0%	98.5%	Intellectual developmental disorder with microcephaly and with or without ocular malformations or hypogonadotropic hypogonadism, 615866
SOX2	100.0%	100.0%	100.0%	99.4%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900;Microphthalmia, syndromic 3, 206900
SOX3	100.0%	100.0%	98.2%	74.5%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123;Panhypopituitarism, X-linked, 312000

SOX9	100.0%	100.0%	100.0%	99.8%	Campomelic dysplasia with autosomal sex reversal, 114290;Acampomelic campomelic dysplasia, 114290;Campomelic dysplasia, 114290
SPATA22	100.0%	100.0%	100.0%	98.5%	
SPIDR	100.0%	100.0%	100.0%	99.6%	Ovarian dysgenesis 9, 619665
SPRY4	100.0%	100.0%	100.0%	99.8%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SRCAP	100.0%	100.0%	100.0%	99.6%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595;Floating-Harbor syndrome, 136140
SRD5A2	100.0%	100.0%	100.0%	99.6%	Pseudovaginal perineoscrotal hypospadias, 264600
SRY	50.0%	50.0%	49.1%	24.4%	46XY sex reversal 1, 400044;46XX sex reversal 1, 400045
STAG3	100.0%	100.0%	100.0%	99.3%	Spermatogenic failure 61, 619672;Premature ovarian failure 8, 615723
STAR	100.0%	100.0%	100.0%	99.9%	Lipoid adrenal hyperplasia, 201710
SYCE1	100.0%	100.0%	100.0%	99.7%	?Spermatogenic failure 15, 616950;?Premature ovarian failure 12, 616947
TAC3	100.0%	100.0%	100.0%	99.9%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	100.0%	99.8%	100.0%	99.3%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TBX19	100.0%	100.0%	100.0%	99.7%	Adrenocorticotropic hormone deficiency, 201400
TBX3	100.0%	100.0%	100.0%	99.4%	Ulnar-mammary syndrome, 181450

TCF12	100.0%	100.0%	100.0%	99.6%	Craniosynostosis 3, 615314;Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TCTN3	100.0%	100.0%	100.0%	99.4%	Joubert syndrome 18, 614815;Orofaciodigital syndrome IV, 258860
TENM1	99.9%	99.5%	98.8%	75.0%	
TOE1	100.0%	100.0%	100.0%	99.5%	Pontocerebellar hypoplasia, type 7, 614969
TP63	100.0%	99.9%	100.0%	99.7%	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	100.0%	100.0%	100.0%	99.7%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TWNK	100.0%	100.0%	100.0%	99.7%	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245;Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286;Perrault syndrome 5, 616138
TXNRD2	100.0%	100.0%	100.0%	99.8%	?Glucocorticoid deficiency 5, 617825
WDR11	100.0%	100.0%	100.0%	99.6%	Intellectual developmental disorder, autosomal recessive 78, 620237;Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR60	100.0%	100.0%	100.0%	99.2%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503

WNT4	100.0%	99.8%	100.0%	99.3%	?SERKAL syndrome, 611812;Mullerian aplasia and hyperandrogenism, 158330
WT1	100.0%	100.0%	100.0%	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	100.0%	100.0%	100.0%	99.6%	Diaphragmatic hernia 3, 610187;46XY sex reversal 9, 616067;Tetralogy of Fallot, 187500

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 3.7.0.