DISORDERS OF SEX DEVELOPMENT GENE PANEL DG 3.4.0 (153 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
AAAS	100,0%	100,0%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS2	100,0%	100,0%	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
ABCD1	100,0%	100,0%	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ADCY3	100,0%	100,0%	No OMIM Disease ID
AIRE	100,0%	100,0%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKR1C2	100,0%	100,0%	46XY sex reversal 8, 614279
AMH	100,0%	100,0%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100,0%	100,0%	Persistent Mullerian duct syndrome, type II, 261550
ANOS1	100,0%	100,0%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AR	100,0%	100,0%	Androgen insensitivity, partial, with or without breast cancer, 312300 Androgen insensitivity, 300068 Spinal and bulbar muscular atrophy of Kennedy, 313200 Hypospadias 1, X-linked, 300633
ARMC5	100,0%	100,0%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARX	99,0%	96,8%	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%	No OMIM Disease ID
ATRX	100,0%	100,0%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Intellectual disability-hypotonic facies syndrome, X-linked, 309580
AXL	100,0%	100,0%	No OMIM Disease ID
B9D1	96,6%	94,1%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120

BMP15	100,0%	100,0%	Premature ovarian failure 4, 300510 Ovarian dysgenesis 2, 300510
BMP4	100,0%	100,0%	Orofacial cleft 11, 600625 Microphthalmia, syndromic 6, 607932
BMP7	100,0%	100,0%	No OMIM Disease ID
CBX2	100,0%	100,0%	?46XY sex reversal 5, 613080
CCDC141	100,0%	100,0%	No OMIM Disease ID
CCNQ	99,9%	99,8%	STAR syndrome, 300707
CDH2	100,0%	100,0%	Arrhythmogenic right ventricular dysplasia, familial, 14, 618920 Agenesis of corpus callosum, cardiac, ocular, and genital syndrome, 618929
CDKN1C	100,0%	100,0%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CEP41	100,0%	100,0%	Joubert syndrome 15, 614464
CHD7	100,0%	100,0%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CLPP	100,0%	100,0%	Perrault syndrome 3, 614129
CNGA2	100,0%	100,0%	No OMIM Disease ID
CREBBP	100,0%	100,0%	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CYB5A	100,0%	100,0%	Methemoglobinemia and ambiguous genitalia, 250790
CYP11A1	100,0%	100,0%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100,0%	100,0%	Aldosteronism, glucocorticoid-remediable, 103900 Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010
CYP11B2	100,0%	100,0%	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Aldosterone to renin ratio raised,
CYP17A1	100,0%	100,0%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	100,0%	100,0%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP21A2	100,0%	100,0%	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910
DCC	100,0%	100,0%	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%	Smith-Lemli-Opitz syndrome, 270400

DHH	100,0%	100,0%	46XY gonadal dysgenesis with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DMRT1	100,0%	100,0%	No OMIM Disease ID
DMRT2	100,0%	100,0%	No OMIM Disease ID
DUSP6	100,0%	100,0%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DYNC2H1	100,0%	100,0%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
WDR60	100,0%	100,0%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
EIF2B5	100,0%	100,0%	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
ERAL1	100,0%	100,0%	Perrault syndrome 6, 617565
ESR1	100,0%	100,0%	Breast cancer, somatic, 114480 Estrogen resistance, 615363
ESR2	100,0%	100,0%	?Ovarian dysgenesis 8, 618187
FANCM	100,0%	100,0%	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FEZF1	100,0%	100,0%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	100,0%	100,0%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	100,0%	100,0%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100,0%	100,0%	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%	Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 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 Beare-Stevenson cutis gyrata syndrome, 123790 Crouzon syndrome, 123500 Saethre-Chotzen syndrome, 101400

			Scaphocephaly and Axenfeld-Rieger anomaly, Craniosynostosis, nonspecific,
FLRT3	100,0%	100,0%	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FOXL2	100,0%	100,0%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Premature ovarian failure 3, 608996
FRAS1	100,0%	100,0%	Fraser syndrome 1, 219000
FREM2	100,0%	100,0%	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FSHB	100,0%	100,0%	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	100,0%	100,0%	Ovarian response to FSH stimulation, 276400 Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300
FZD2	100,0%	100,0%	Omodysplasia 2, 164745
GATA4	100,0%	100,0%	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%	?Premature ovarian failure 14, 618014
GK	100,0%	100,0%	Glycerol kinase deficiency, 307030
GNRH1	100,0%	100,0%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	100,0%	100,0%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GRIP1	100,0%	100,0%	Fraser syndrome 3, 617667
HARS2	100,0%	100,0%	Perrault syndrome 2, 614926
HESX1	100,0%	100,0%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HFM1	100,0%	100,0%	Premature ovarian failure 9, 615724
HOXA13	100,0%	99,7%	Hand-foot-uterus syndrome, 140000 ?Guttmacher syndrome, 176305
HS6ST1	100,0%	100,0%	No OMIM Disease ID
HSD17B3	99,0%	99,0%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	96,6%	96,6%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	100,0%	100,0%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
IGSF10	100,0%	100,0%	No OMIM Disease ID

IL17RD	100,0%	100,0%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IRF6	100,0%	100,0%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome 1, 119300
KAT6B	100,0%	100,0%	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KISS1	100,0%	100,0%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100,0%	100,0%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KLB	100,0%	100,0%	No OMIM Disease ID
LARS2	100,0%	100,0%	Perrault syndrome 4, 615300 Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LEP	100,0%	100,0%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94,6%	94,6%	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	100,0%	100,0%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	100,0%	100,0%	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%	No OMIM Disease ID
LHX3	100,0%	100,0%	Pituitary hormone deficiency, combined, 3, 221750
LIPA	95,2%	95,2%	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
MAMLD1	100,0%	100,0%	Hypospadias 2, X-linked, 300758
MAP3K1	100,0%	100,0%	46XY sex reversal 6, 613762
MC2R	100,0%	100,0%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM8	94,4%	94,4%	?Premature ovarian failure 10, 612885
MCM9	100,0%	100,0%	Ovarian dysgenesis 4, 616185
MKKS	100,0%	100,0%	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231
MKRN3	96,0%	96,0%	Precocious puberty, central, 2, 615346
MRAP	100,0%	100,0%	Glucocorticoid deficiency 2, 607398
MSH4	100,0%	100,0%	No OMIM Disease ID
MYRF	100,0%	100,0%	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 Cardiac-urogenital syndrome, 618280
NEK1	100,0%	100,0%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NNT	96,4%	96,4%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736

NOBOX	100,0%	100,0%	Premature ovarian failure 5, 611548
NR0B1	100,0%	100,0%	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NR3C1	100,0%	100,0%	Glucocorticoid resistance, 615962
NR3C2	100,0%	100,0%	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR5A1	100,0%	100,0%	46, XX 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%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PBX1	100,0%	100,0%	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCSK1	100,0%	100,0%	Obesity with impaired prohormone processing, 600955
PLXNA1	100,0%	100,0%	No OMIM Disease ID
POLE	100,0%	100,0%	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLG	100,0%	100,0%	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
POMC	100,0%	100,0%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POR	100,0%	100,0%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PPP1R12A	100,0%	100,0%	Genitourinary and/or/brain malformation syndrome, 618820
PROK2	100,0%	100,0%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100,0%	100,0%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	100,0%	100,0%	Pituitary hormone deficiency, combined, 2, 262600
PSMC3IP	100,0%	100,0%	Ovarian dysgenesis 3, 614324
REC8	100,0%	100,0%	No OMIM Disease ID
RIPK4	100,0%	100,0%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
ROR2	97,0%	97,0%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310

RSPO1	100,0%	100,0%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
SAMD9	100,0%	100,0%	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SEMA3A	100,0%	100,0%	No OMIM Disease ID
SGPL1	100,0%	100,0%	Nephrotic syndrome, type 14, 617575
SOHLH1	100,0%	100,0%	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
SOX10	100,0%	100,0%	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX2	100,0%	100,0%	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 Microphthalmia, syndromic 3, 206900
SOX3	100,0%	100,0%	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	100,0%	100,0%	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290
SPRY4	100,0%	100,0%	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SRCAP	100,0%	100,0%	Developmental delay, hypotonia, musculoskeletal defects, and behavioral abnormalities, 619595 Floating-Harbor syndrome, 136140
SRD5A2	100,0%	100,0%	Pseudovaginal perineoscrotal hypospadias, 264600
SRY	50,0%	50,0%	46XY sex reversal 1, 400044
STAG3	100,0%	100,0%	Spermatogenic failure 61, 619672 Premature ovarian failure 8, 615723
STAR	100,0%	100,0%	Lipoid adrenal hyperplasia, 201710
SYCE1	100,0%	100,0%	?Spermatogenic failure 15, 616950 ?Premature ovarian failure 12, 616947
TAC3	100,0%	100,0%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	100,0%	100,0%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TBX19	100,0%	100,0%	Adrenocorticotropic hormone deficiency, 201400
TBX3	100,0%	100,0%	Ulnar-mammary syndrome, 181450
TCF12	100,0%	100,0%	Craniosynostosis 3, 615314 Hypogonadotropic hypogonadism 26 with or without anosmia, 619718
TCTN3	100,0%	100,0%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860

TOE1	100,0%	100,0%	Pontocerebellar hypoplasia, type 7, 614969
TSPYL1	100,0%	100,0%	Sudden infant death with dysgenesis of the testes syndrome, 608800
TWNK	100,0%	100,0%	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%	?Glucocorticoid deficiency 5, 617825
WDR11	100,0%	100,0%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WNT4	100,0%	99,8%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	97,7%	97,7%	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%	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.
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TWIST is the chemistry used for WES analysis.
Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with coverage denoting NC are non-protein coding genes.
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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors