DISORDERS OF SEX DEVELOPMENT GENE PANEL DG 3.2.0 (149 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
AAAS	100	99,4	100	100	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS2	100	99,4	100	100	Leukoencephalopathy, progressive, with ovarian failure, 615889 Combined oxidative phosphorylation deficiency 8, 614096
ABCD1	76	72,6	100	100	Adrenoleukodystrophy, 300100 Adrenomyeloneuropathy, adult, 300100
ADCY3	100	99	100	100	No OMIM disease ID
AIRE	100	99,9	100	100	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKR1C2	94,5	88,3	100	100	46XY sex reversal 8, 614279
AMH	99,4	92,9	100	100	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100	99,6	100	100	Persistent Mullerian duct syndrome, type II, 261550
ANOS1	89,8	88,3	99,9	99,4	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AR	98,1	93,6	99,9	99,5	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	99,1	100	100	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARX	82,1	67,5	91,4	86,6	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	99,9	97,3	100	100	No OMIM disease ID
ATRX	98,7	95,2	100	99,9	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580

AXL	100	98,9	100	100	No OMIM disease ID
B9D1	85,2	85,2	95,8	94	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
BMP15	100	98,7	100	100	Premature ovarian failure 4, 300510 Ovarian dysgenesis 2, 300510
BMP4	100	100	100	100	Orofacial cleft 11, 600625 Microphthalmia, syndromic 6, 607932
BMP7	99,8	98,4	100	100	No OMIM disease ID
CBX2	100	100	100	100	?46XY sex reversal 5, 613080
CCDC141	99,8	99,5	100	99,9	No OMIM disease ID
CCNQ	82,9	78,3	99,8	98,2	STAR syndrome, 300707
CDKN1C	89,9	81,6	98,9	95,8	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CEP41	98,8	93,4	100	100	Joubert syndrome 15, 614464
CHD7	100	99,2	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CLPP	100	99,5	100	100	Perrault syndrome 3, 614129
CREBBP	99,6	97,8	100	100	Menke-Hennekam syndrome 1, 618332 Rubinstein-Taybi syndrome 1, 180849
CYB5A	100	100	100	100	Methemoglobinemia and ambiguous genitalia, 250790
CYP11A1	99,2	94,5	100	100	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100	99,9	100	100	Aldosteronism, glucocorticoid-remediable, 103900 Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010
CYP11B2	100	99,9	100	100	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Aldosterone to renin ratio raised,
CYP17A1	99,9	98,5	100	100	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	98,3	95,7	100	100	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP21A2	97,4	91,1	100	100	Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910
DCC	100	100	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	100	100	100	100	Smith-Lemli-Opitz syndrome, 270400
DHH	100	100	100	100	46XY gonadal dysgenesis with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DMRT1	100	98,9	100	100	No OMIM disease ID
DMRT2	98,9	91,6	100	100	No OMIM disease ID
DUSP6	100	100	100	100	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DYNC2H1	98,6	95,2	100	99,8	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
WDR60	99,3	95,8	100	100	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
EIF2B5	99,8	98,5	100	100	Ovarioleukodystrophy, 603896 Leukoencephalopathy with vanishing white matter, 603896
ESR1	100	99,8	100	100	Breast cancer, somatic, 114480 Estrogen resistance, 615363
ESR2	99,9	98,9	100	100	?Ovarian dysgenesis 8, 618187
FANCM	98,9	96,3	100	100	?Premature ovarian failure 15, 618096 Spermatogenic failure 28, 618086
FEZF1	100	100	100	100	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	100	100	100	100	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	97,1	87,2	100	99,9	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100	99,3	100	100	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	97,6	97	100	100	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	100	100	100	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FOXL2	99,4	94,7	99,9	99	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Premature ovarian failure 3, 608996
FRAS1	100	99,2	100	100	Fraser syndrome 1, 219000
FREM2	99,8	98,7	100	100	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FSHB	100	100	100	100	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	99,2	97	100	100	Ovarian response to FSH stimulation, 276400 Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300
FZD2	100	97,8	100	100	Omodysplasia 2, 164745
GATA4	87,4	78,5	100	100	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	?Premature ovarian failure 14, 618014
GK	84,2	61,8	100	99,6	Glycerol kinase deficiency, 307030
GNRH1	99,5	89,5	100	100	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	100	100	100	100	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GRIP1	100	99,3	100	100	Fraser syndrome 3, 617667
HESX1	99,3	97,3	100	100	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HFM1	95,4	89,8	100	99,9	Premature ovarian failure 9, 615724
HOXA13	76,4	67,3	90,3	81,4	Hand-foot-uterus syndrome, 140000 ?Guttmacher syndrome, 176305

HS6ST1	93,6	86,7	100	100	No OMIM disease ID
HSD17B3	97,8	97,8	100	100	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	95,3	92,8	96,6	96,6	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	100	99,7	100	100	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
IGSF10	100	99,9	100	100	No OMIM disease ID
IL17RD	99,9	99	100	100	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
IRF6	99,4	93	100	100	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300
KAT6B	99,4	98	100	100	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KISS1	100	98,2	100	100	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100	99,6	100	100	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KLB	100	99,9	100	100	No OMIM disease ID
LARS2	100	100	100	100	Perrault syndrome 4, 615300 Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LEP	100	99,6	100	100	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94,1	92,3	94,6	94,5	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	91,7	42,8	100	100	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	96,6	92,4	100	100	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	99,8	100	100	No OMIM disease ID
LHX3	96,6	96,2	100	100	Pituitary hormone deficiency, combined, 3, 221750
LIPA	96,9	94,6	95,2	95,2	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
MAMLD1	99,7	97,8	100	100	Hypospadias 2, X-linked, 300758
MAP3K1	97	93	100	99,9	46XY sex reversal 6, 613762
MC2R	99,7	97,4	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM8	99,9	98,8	94,4	94,3	?Premature ovarian failure 10, 612885

MCM9	99,9	99	100	100	Ovarian dysgenesis 4, 616185
MKKS	100	100	100	100	McKusick-Kaufman syndrome, 236700 Bardet-Biedl syndrome 6, 605231
MKRN3	96	96	96	96	Precocious puberty, central, 2, 615346
MRAP	100	100	100	100	Glucocorticoid deficiency 2, 607398
MSH4	98,8	96,5	100	99,5	No OMIM disease ID
MYRF	99	97,8	100	100	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 Cardiac-urogenital syndrome, 618280
NEK1	99,5	98,2	100	99,9	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NNT	96,4	96	96,4	96,4	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	99,3	96,6	100	100	Premature ovarian failure 5, 611548
NR0B1	99,9	99,2	100	100	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NR3C1	100	99,9	100	100	Glucocorticoid resistance, 615962
NR3C2	99,9	99,8	100	100	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR5A1	100	100	100	100	46, XX sex reversal 4, 617480 Premature ovarian failure 7, 612964 46XY sex reversal 3, 612965 Adrenocortical insufficiency, 612964 Spermatogenic failure 8, 613957
NSMF	96,9	95,5	100	100	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PBX1	100	99,1	100	100	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCSK1	99,9	99,4	100	100	Obesity with impaired prohormone processing, 600955
PLXNA1	100	99,9	100	100	No OMIM disease ID
PNPLA6	100	99,8	100	100	Spastic paraplegia 39, autosomal recessive, 612020 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800 Boucher-Neuhauser syndrome, 215470
POLE	100	99,5	100	100	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POLG	99,9	98,8	100	100	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	100	100	100	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POR	99,5	98	100	100	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
PPP1R12A	97,8	95,9	100	99,8	Genitourinary and/or/brain malformation syndrome, 618820
PROK2	99,9	98,9	100	100	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100	100	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	91	80,2	100	100	Pituitary hormone deficiency, combined, 2, 262600
PSMC3IP	100	100	100	100	Ovarian dysgenesis 3, 614324
RIPK4	100	99,9	100	100	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type 1, 263650
ROR2	100	99,4	97	97	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RSPO1	100	99,9	100	100	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
SAMD9	99,9	99,8	100	100	Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 MIRAGE syndrome, 617053
SEMA3A	100	99,7	100	100	No OMIM disease ID
SGPL1	100	100	100	100	Nephrotic syndrome, type 14, 617575
SOHLH1	99,7	96,8	100	100	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
SOX10	99,9	97,2	100	100	Waardenburg syndrome, type 4C, 613266 PCWH syndrome, 609136 Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584
SOX2	100	99,8	100	100	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 Microphthalmia, syndromic 3, 206900
SOX3	94,9	81,2	100	99,6	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	100	99,9	100	100	Campomelic dysplasia with autosomal sex reversal, 114290 Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290
SPRY4	100	100	100	100	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266

SRCAP	99,7	98,9	100	100	Floating-Harbor syndrome, 136140
SRD5A2	100	98,8	100	100	Pseudovaginal perineoscrotal hypospadias, 264600
SRY	50	49,9	50	50	46XY sex reversal 1, 400044
STAG3	93,5	92,8	100	100	Premature ovarian failure 8, 615723
STAR	100	99,9	100	100	Lipoid adrenal hyperplasia, 201710
SYCE1	99,9	98,8	100	100	?Spermatogenic failure 15, 616950 ?Premature ovarian failure 12, 616947
TAC3	99,9	93,6	100	100	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	100	100	100	100	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
TBX19	100	100	100	100	Adrenocorticotropic hormone deficiency, 201400
TBX3	99,4	97,3	100	100	Ulnar-mammary syndrome, 181450
TCF12	99,9	99,7	100	100	Craniosynostosis 3, 615314
TCTN3	100	100	100	100	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TOE1	100	100	100	100	Pontocerebellar hypoplasia, type 7, 614969
TSPYL1	100	100	100	100	Sudden infant death with dysgenesis of the testes syndrome, 608800
TWNK	100	99,9	100	100	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 Perrault syndrome 5, 616138
TXNRD2	96,8	95,9	100	100	?Glucocorticoid deficiency 5, 617825
WDR11	98,2	96,5	100	100	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WNT4	97,8	93,6	99,3	96,5	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	97,6	96,1	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	99,9	100	100	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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Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.
TWIST is the chemistry used for (in-house) TURBO/RAPID 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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors