DSD/PRIMARY ADRENAL INSUFFICIENCY GENE PANEL DG 3.1.0 (147 genes)

Releasedate: 23-03-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
AARS2	100	99,4	100	100	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABCD1	75,8	71,6	100	100	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ADCY3	100	99,1	100	100	{Obesity, susceptibility to, BMIQ19}, 617885
AIRE	100	99,8	100	100	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKR1C2	94,9	89,2	100	100	46XY sex reversal 8, 614279
AMH	96,4	83,8	100	99,8	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100	99,5	100	100	Persistent Mullerian duct syndrome, type II, 261550
ANOS1	89,8	88,9	99,9	99,4	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AR	97,6	93,3	100	99,2	Hypospadias 1, X-linked, 300633 Androgen insensitivity, 300068 {Prostate cancer, susceptibility to}, 176807 Androgen insensitivity, partial, with or without breast cancer, 312300 Spinal and bulbar muscular atrophy of Kennedy, 313200
ARMC5	100	99,4	100	100	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARX	81	64	91,5	85,7	Lissencephaly, X-linked 2, 300215 Proud syndrome, 300004 Partington syndrome, 309510 Developmental and epileptic encephalopathy 1, 308350 Mental retardation, X-linked 29 and others, 300419 Hydranencephaly with abnormal genitalia, 300215
ATF3	99,9	97,5	100	100	No OMIM disease ID
ATRX	99,4	96,3	100	100	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580

AXL	100	99,7	100	100	No OMIM disease ID
B9D1	85,2	85,1	94,2	93,9	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
BMP15	100	99,3	100	100	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
BMP4	100	100	100	100	Microphthalmia, syndromic 6, 607932 Orofacial cleft 11, 600625
BMP7	99,9	98,5	100	100	No OMIM disease ID
CBX2	100	99,8	100	100	?46XY sex reversal 5, 613080
CCDC141	100	99,5	100	100	No OMIM disease ID
CCNQ	83,1	78,5	98,9	94,6	STAR syndrome, 300707
CDKN1C	88	77,8	99,3	97,3	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CEP41	99,8	97,4	100	100	Joubert syndrome 15, 614464
CHD7	100	99,5	100	100	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CLPP	100	99,1	100	100	Perrault syndrome 3, 614129
CREBBP	99,7	98,5	100	100	Rubinstein-Taybi syndrome 1, 180849 Menke-Hennekam syndrome 1, 618332
CYB5A	100	100	100	100	Methemoglobinemia and ambiguous genitalia, 250790
CYP11A1	99,3	96,1	100	100	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	100	100	100	100	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP11B2	100	100	100	100	Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 Aldosterone to renin ratio raised, 0 {Low renin hypertension, susceptibility to}, 0
CYP17A1	100	99,5	100	100	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	98,8	96,8	100	100	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP21A2	97,8	88,4	100	100	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
DCC	100	100	100	100	Esophageal carcinoma, somatic, 133239 Gaze palsy, familial horizontal, with progressive scoliosis, 2, 617542 Mirror movements 1 and/or agenesis of the corpus callosum, 157600 Colorectal cancer, somatic, 114500

DHCR7	100	100	100	100	Smith-Lemli-Opitz syndrome, 270400
DHH	100	100	100	100	46XY sex reversal 7, 233420 46XY gonadal dysgenesis with minifascicular neuropathy, 607080
DMRT1	100	99,8	100	100	No OMIM disease ID
DMRT2	97,7	88,4	100	100	No OMIM disease ID
DUSP6	100	100	100	100	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DYNC2H1	98,8	95,5	100	100	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
EIF2B5	100	99,1	100	100	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
ESR1	100	99,8	100	100	{Myocardial infarction, susceptibility to}, 608446 Estrogen resistance, 615363 Breast cancer, somatic, 114480 {Migraine, susceptibility to}, 157300
ESR2	100	99,7	100	100	?Ovarian dysgenesis 8, 618187
FEZF1	100	99,9	100	100	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	100	100	100	100	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	98,2	88,9	100	99,6	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100	99,9	100	100	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 Hartsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	97,7	97,1	100	100	Apert syndrome, 101200 Jackson-Weiss syndrome, 123150 Saethre-Chotzen syndrome, 101400 Gastric cancer, somatic, 613659 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Craniofacial-skeletal-dermatologic dysplasia, 101600 Pfeiffer syndrome, 101600 Crouzon syndrome, 123500 Beare-Stevenson cutis gyrata syndrome, 123790 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Craniosynostosis, nonspecific, 0 Scaphocephaly and Axenfeld-Rieger anomaly, 0

FLRT3	100	100	100	100	Hypogonadotropic hypogonadism 21 with anosmia, 615271
FOXL2	99,7	95,5	99,8	98	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996 Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100
FRAS1	100	99,4	100	100	Fraser syndrome 1, 219000
FREM2	100	99,3	100	100	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FSHB	100	100	100	100	Hypogonadotropic hypogonadism 24 without anosmia, 229070
FSHR	99,5	97,2	100	100	Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300 Ovarian response to FSH stimulation, 276400
FZD2	99,9	98,2	100	100	Omodysplasia 2, 164745
GATA4	84,1	74,5	100	99,9	?Testicular anomalies with or without congenital heart disease, 615542 Tetralogy of Fallot, 187500 Atrioventricular septal defect 4, 614430 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429
GDF9	100	100	100	100	?Premature ovarian failure 14, 618014
GK	88,9	70,4	100	99,9	Glycerol kinase deficiency, 307030
GNRH1	100	93,7	100	100	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	100	100	100	100	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GRIP1	100	99,7	100	100	Fraser syndrome 3, 617667
HESX1	99,7	97,3	100	100	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HFM1	96,3	89,6	100	100	Premature ovarian failure 9, 615724
HOXA13	77,7	69	89,7	79,7	?Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HS6ST1	92,9	84,5	100	100	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880
HSD17B3	97,8	97,8	97,8	97,8	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	95,4	93,1	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	100	100	100	No OMIM disease ID
IL17RD	99,9	99,1	100	100	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267

IRF6	99,6	95,9	100	100	Popliteal pterygium syndrome 1, 119500 {Orofacial cleft 6}, 608864 van der Woude syndrome, 119300
KAT6B	99,6	98,3	100	100	SBBYSS syndrome, 603736 Genitopatellar syndrome, 606170
KISS1	100	98,3	100	100	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100	99,5	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	99,9	97,3	100	100	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94,3	92,6	94,6	94,6	Obesity, morbid, due to leptin receptor deficiency, 614963
LHB	90,4	38,9	100	100	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	94,1	92,3	100	100	Leydig cell adenoma, somatic, with precocious puberty, 176410 Precocious puberty, male, 176410 Luteinizing hormone resistance, female, 238320 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320
LHX1	100	99,6	100	100	No OMIM disease ID
LHX3	96,6	96,5	100	100	Pituitary hormone deficiency, combined, 3, 221750
LIPA	99,2	95,2	95,2	95,2	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
MAMLD1	99,8	98,2	100	100	Hypospadias 2, X-linked, 300758
MAP3K1	96,1	91,6	99,7	98,3	46XY sex reversal 6, 613762
MC2R	99,9	98,3	100	100	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM8	100	99,6	94,4	94,4	?Premature ovarian failure 10, 612885
MCM9	99,9	99,8	100	100	Ovarian dysgenesis 4, 616185
MKKS	100	100	100	100	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MKRN3	96	96	96	96	Precocious puberty, central, 2, 615346
MRAP	100	100	100	100	Glucocorticoid deficiency 2, 607398
MYRF	99,3	98,5	100	100	Cardiac-urogenital syndrome, 618280 Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113
NEK1	99,8	98	100	100	{Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892 Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NNT	96,4	95,9	96,4	96,4	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736

NOBOX	99,9	98,4	100	99,8	Premature ovarian failure 5, 611548
NR0B1	100	99,5	100	100	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NR3C1	100	99,9	100	100	Glucocorticoid resistance, 615962
NR3C2	100	99,7	100	100	Pseudohypoaldosteronism type I, autosomal dominant, 177735 Hypertension, early-onset, autosomal dominant, with exacerbation in pregnancy, 605115
NR5A1	100	100	100	100	Adrenocortical insufficiency, 612964 46, XX sex reversal 4, 617480 Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957 46XY sex reversal 3, 612965
NSMF	96,1	95,6	100	100	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PBX1	100	99,4	100	100	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641
PCSK1	100	99,5	100	100	{Obesity, susceptibility to, BMIQ12}, 612362 Obesity with impaired prohormone processing, 600955
PLXNA1	100	99,6	100	100	No OMIM disease ID
PNPLA6	100	99,7	100	100	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470 Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
POLE	100	99,8	100	100	FILS syndrome, 615139 IMAGE-I syndrome, 618336 {Colorectal cancer, susceptibility to, 12}, 615083
POLG	100	99,3	100	100	Progressive external ophthalmoplegia, autosomal dominant 1, 157640 Mitochondrial DNA depletion syndrome 4B (MNGIE type), 613662 Mitochondrial recessive ataxia syndrome (includes SANDO and SCAE), 607459 Mitochondrial DNA depletion syndrome 4A (Alpers type), 203700 Progressive external ophthalmoplegia, autosomal recessive 1, 258450
POMC	100	100	100	100	{Obesity, early-onset, susceptibility to}, 601665 Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POR	99,8	98,6	100	100	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750
PPP1R12A-AS1	NC	NC	NC	NC	No OMIM disease ID
PROK2	99,9	98,5	100	100	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100	100	100	100	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	92,6	82,6	100	100	Pituitary hormone deficiency, combined, 2, 262600
PSMC3IP	100	100	100	100	Ovarian dysgenesis 3, 614324

RIPK4	100	99,9	100	100	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
ROR2	100	99,9	97	97	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RSPO1	100	99,9	100	100	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 Palmoplantar hyperkeratosis and true hermaphroditism, 610644
SAMD9	100	99,8	100	100	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455 Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041
SEMA3A	100	99,9	100	100	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897
SGPL1	100	100	100	100	Nephrotic syndrome, type 14, 617575
SOHLH1	99,7	96,5	100	100	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
SOX10	99,9	97,9	100	100	Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 PCWH syndrome, 609136 Waardenburg syndrome, type 4C, 613266
SOX2	100	100	100	100	Microphthalmia, syndromic 3, 206900 Optic nerve hypoplasia and abnormalities of the central nervous system, 206900
SOX3	91,4	75,2	100	99,5	Panhypopituitarism, X-linked, 312000 Mental retardation, X-linked, with isolated growth hormone deficiency, 300123
SOX9	100	98,6	100	100	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SPRY4	100	100	100	100	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266
SRCAP	99,4	98,9	100	100	Floating-Harbor syndrome, 136140
SRD5A2	99,9	99	100	100	Pseudovaginal perineoscrotal hypospadias, 264600
SRY	50	50	60	60	46XY sex reversal 1, 400044 46XX sex reversal 1, 400045
STAG3	93,5	93,2	100	100	Premature ovarian failure 8, 615723
STAR	100	100	100	100	Lipoid adrenal hyperplasia, 201710
SYCE1	100	98,6	100	100	?Premature ovarian failure 12, 616947 ?Spermatogenic failure 15, 616950
TAC3	100	99,6	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	99,2	96,8	100	100	Ulnar-mammary syndrome, 181450
TCF12	100	99,9	100	100	Craniosynostosis 3, 615314

TCTN3	100	100	100	100	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
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	100	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	96,5	100	100	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR60	99,5	97	100	100	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNT4	99,1	94,8	98,9	96,2	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	97,3	95,4	97,7	97,7	Mesothelioma, somatic, 156240 Wilms tumor, type 1, 194070 Frasier syndrome, 136680 Denys-Drash syndrome, 194080 Meacham syndrome, 608978 Nephrotic syndrome, type 4, 256370
ZFPM2	100	100	100	100	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 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.
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 : March 23rd , 2021.
This list is accurate for panel version DG 3.1.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors