DSD/PRIMARY ADRENAL INSUFFICIENCY GENE PANEL
DG 2.18 (135 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
AAAS	100%	99,90%	100%	100%	Achalasia-addisonianism-alacrimia syndrome, 231550
AARS2	100%	99,40%	100%	100%	Combined oxidative phosphorylation deficiency 8, 614096 Leukoencephalopathy, progressive, with ovarian failure, 615889
ABCD1	75,80%	71,60%	100%	100%	Adrenomyeloneuropathy, adult, 300100 Adrenoleukodystrophy, 300100
ADCY3	100%	99,10%	100%	100%	No OMIM disease ID
AIRE	100%	99,80%	100%	100%	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300
AKR1C2	94,90%	89,20%	100%	100%	46XY sex reversal 8, 614279
AMH	96,40%	83,80%	100%	99,80%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	100%	99,50%	100%	100%	Persistent Mullerian duct syndrome, type II, 261550
ANOS1	89,80%	88,90%	99,90%	99,40%	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700
AR	97,60%	93,20%	100%	99,20%	Hypospadias 1, X-linked, 300633 Androgen insensitivity, 300068 Androgen insensitivity, partial, with or without breast cancer, 312300 Spinal and bulbar muscular atrophy of Kennedy, 313200
ARMC5	100%	99,40%	100%	100%	ACTH-independent macronodular adrenal hyperplasia 2, 615954
ARX	81,00%	64,00%	91,50%	85,70%	Proud syndrome, 300004 Partington syndrome, 309510 Lissencephaly, X-linked 2, 300215 Epileptic encephalopathy, early infantile, 1, 308350 Mental retardation, X-linked 29 and others, 300419 Hydranencephaly with abnormal genitalia, 300215
ATF3	99,90%	97,50%	100%	100%	No OMIM disease ID
ATRX	99,40%	96,30%	100%	100%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580

B9D1	92,20%	92,00%	100%	100%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
BMP15	100%	99,30%	100%	100%	Ovarian dysgenesis 2, 300510 Premature ovarian failure 4, 300510
CBX2	100%	99,80%	100%	100%	?46XY sex reversal 5, 613080
CCDC141	100%	99,30%	100%	100%	No OMIM disease ID
CDKN1C	86,30%	74,80%	99,20%	96,90%	IMAGE syndrome, 614732 Beckwith-Wiedemann syndrome, 130650
CEP41	99,80%	97,40%	100%	100%	Joubert syndrome 15, 614464
CHD7	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 CHARGE syndrome, 214800
CLPP	100%	99,10%	100%	100%	Perrault syndrome 3, 614129
CYB5A	100%	100%	100%	100%	Methemoglobinemia and ambiguous genitalia, 250790
CYP11A1	99,30%	96,10%	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
CYP17A1	100%	99,50%	100%	100%	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	98,80%	96,80%	100%	100%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP21A2	97,80%	88,40%	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 partial gonadal dysgenesis, with minifascicular neuropathy, 607080
DMRT1	100%	99,80%	100%	100%	No OMIM disease ID
DMRT2	97,70%	88,40%	100%	100%	No OMIM disease ID
DUSP6	100%	100%	100%	100%	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269
DYNC2H1	98,80%	95,50%	100%	100%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091

EIF2B5	100%	99,00%	100%	100%	Leukoencephalopathy with vanishing white matter, 603896 Ovarioleukodystrophy, 603896
ESR1	100%	99,80%	100%	100%	Estrogen resistance, 615363 Breast cancer, somatic, 114480
ESR2	100%	99,70%	100%	100%	?Ovarian dysgenesis 8, 618187
FAM58A	83,10%	78,50%	98,90%	94,70%	STAR syndrome, 300707
FEZF1	100%	99,90%	100%	100%	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030
FGF17	100%	100%	100%	100%	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270
FGF8	98,20%	88,90%	100%	99,60%	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702
FGFR1	100%	99,90%	100%	100%	Pfeiffer syndrome, 101600 Jackson-Weiss syndrome, 123150 Trigonocephaly 1, 190440 Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 HEARTsfield syndrome, 615465 Osteoglophonic dysplasia, 166250 Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001
FGFR2	97,70%	97,10%	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	99,70%	95,50%	99,80%	98,00%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996 Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100
FRAS1	100%	99,40%	100%	100%	Fraser syndrome 1, 219000
FREM2	100%	99,30%	100%	100%	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
FSHB	100%	100%	100%	100%	Hypogonadotropic hypogonadism 24 without anosmia, 229070

FSHR	99,50%	97,20%	100%	100%	Ovarian hyperstimulation syndrome, 608115 Ovarian dysgenesis 1, 233300 Ovarian response to FSH stimulation, 276400
GATA4	84,10%	74,50%	100%	99,90%	?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	88,90%	70,40%	100%	99,90%	Glycerol kinase deficiency, 307030
GNRH1	100%	93,70%	100%	100%	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841
GNRHR	100%	100%	100%	100%	Hypogonadotropic hypogonadism 7 without anosmia, 146110
GRIP1	100%	99,70%	100%	100%	Fraser syndrome 3, 617667
HESX1	99,70%	97,30%	100%	100%	Pituitary hormone deficiency, combined, 5, 182230 Septooptic dysplasia, 182230 Growth hormone deficiency with pituitary anomalies, 182230
HFM1	96,30%	89,60%	100%	100%	Premature ovarian failure 9, 615724
HOXA13	77,70%	69,00%	89,70%	79,70%	?Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HS6ST1	92,90%	84,50%	100%	100%	No OMIM disease ID
HSD17B3	100%	100%	100%	100%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD17B4	96,00%	93,70%	96,60%	96,60%	D-bifunctional protein deficiency, 261515 Perrault syndrome 1, 233400
HSD3B2	100%	99,70%	100%	100%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
IGSF10	100%	100%	100%	100%	No OMIM disease ID
IL17RD	99,90%	99,10%	100%	100%	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267
MR *AP*	100%	100%	100%	100%	Glucocorticoid deficiency 2, 607398
IRF6	99,60%	95,90%	100%	100%	Popliteal pterygium syndrome 1, 119500 van der Woude syndrome, 119300
KISS1	100%	98,30%	100%	100%	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842
KISS1R	100%	99,50%	100%	100%	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 ?Precocious puberty, central, 1, 176400
KLB	100%	99,90%	100%	100%	No OMIM disease ID
LARS2	100%	100%	100%	100%	Perrault syndrome 4, 615300 ?Hydrops, lactic acidosis, and sideroblastic anemia, 617021
LEP	99,90%	97,30%	100%	100%	Obesity, morbid, due to leptin deficiency, 614962
LEPR	94,30%	92,60%	94,60%	94,60%	Obesity, morbid, due to leptin receptor deficiency, 614963

LHB	90,40%	38,90%	100%	100%	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300
LHCGR	94,10%	92,30%	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
LHX3	96,60%	96,50%	100%	100%	Pituitary hormone deficiency, combined, 3, 221750
LIPA	99,20%	95,20%	95,20%	95,20%	Wolman disease, 278000 Cholesteryl ester storage disease, 278000
MAMLD1	99,80%	98,20%	100%	100%	Hypospadias 2, X-linked, 300758
MAP3K1	96,10%	91,60%	99,70%	98,30%	46XY sex reversal 6, 613762
MC2R	99,90%	98,30%	100%	100%	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200
MCM8	100%	99,60%	94,40%	94,40%	?Premature ovarian failure 10, 612885
MCM9	99,90%	99,80%	100%	100%	Ovarian dysgenesis 4, 616185
MKKS	83,20%	83,20%	90,70%	90,70%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
MYRF	99,30%	98,50%	100%	100%	Cardiac-urogenital syndrome, 618280 Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113
NEK1	99,80%	98,00%	100%	100%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NNT	100%	99,40%	100%	100%	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736
NOBOX	99,90%	98,40%	100%	99,80%	Premature ovarian failure 5, 611548
NR0B1	100%	99,50%	100%	100%	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NR3C1	100%	99,90%	100%	100%	Glucocorticoid resistance, 615962
NR3C2	100%	99,70%	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	96,10%	95,60%	100%	100%	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838
PCSK1	100%	99,50%	100%	100%	Obesity with impaired prohormone processing, 600955
TAC3	100%	99,60%	100%	100%	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839
TACR3	100%	100%	100%	100%	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840
PLXNA1	100%	99,60%	100%	100%	No OMIM disease ID
PNPLA6	100%	99,70%	100%	100%	Spastic paraplegia 39, autosomal recessive, 612020 Boucher-Neuhauser syndrome, 215470

					Oliver-McFarlane syndrome, 275400 ?Laurence-Moon syndrome, 245800
POLE	100%	99,80%	100%	100%	FILS syndrome, 615139 IMAGE-I syndrome, 618336
POMC	100%	100%	100%	100%	Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734
POR	99,80%	98,60%	100%	100%	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750
PROK2	99,90%	98,50%	100%	100%	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628
PROKR2	100%	100%	100%	100%	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200
PROP1	92,60%	82,60%	100%	100%	Pituitary hormone deficiency, combined, 2, 262600
PSMC3IP	100%	100%	100%	100%	Ovarian dysgenesis 3, 614324
RIPK4	100%	99,90%	100%	100%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
ROR2	100%	99,90%	97,00%	97,00%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RSPO1	100%	99,90%	100%	100%	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 Palmoplantar hyperkeratosis and true hermaphroditism, 610644
SAMD9	100%	99,80%	100%	100%	MIRAGE syndrome, 617053 Tumoral calcinosis, familial, normophosphatemic, 610455
SEMA3A	100%	99,90%	100%	100%	No OMIM disease ID
SGPL1	100%	100%	100%	100%	Nephrotic syndrome, type 14, 617575
SOHLH1	99,70%	96,50%	100%	100%	Ovarian dysgenesis 5, 617690 Spermatogenic failure 32, 618115
SOX10	99,90%	97,90%	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	91,40%	75,20%	100%	99,50%	Panhypopituitarism, X-linked, 312000 Mental retardation, X-linked, with isolated growth hormone deficiency, 300123
SOX9	100%	98,60%	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	99,40%	98,90%	100%	100%	Floating-Harbor syndrome, 136140
SRD5A2	99,90%	99,00%	100%	100%	Pseudovaginal perineoscrotal hypospadias, 264600
SRY	50,00%	50,00%	60,00%	60,00%	46XY sex reversal 1, 400044
STAG3	93,50%	93,20%	100%	100%	Premature ovarian failure 8, 615723

STAR	100%	100%	100%	100%	Lipoid adrenal hyperplasia, 201710
TBX19	100%	100%	100%	100%	Adrenocorticotropic hormone deficiency, 201400
TBX3	99,20%	96,80%	100%	100%	Ulnar-mammary syndrome, 181450
TCF12	100%	99,90%	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	96,80%	95,90%	100%	100%	?Glucocorticoid deficiency 5, 617825
WDR11	98,00%	96,50%	100%	100%	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858
WDR60	99,50%	97,00%	100%	100%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNT4	99,10%	94,80%	98,90%	96,20%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	99,90%	98,30%	100%	100%	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-DNA coding genes.
non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.
This list is accurate for panel version DG 2.18
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors