

WES DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY DG 3.6

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
AAAS	138.0	100.0	100.0	231550
AARS2	124.6	100.0	100.0	615889
ABCD1	100.2	100.0	99.6	300100
ADCY3	120.5	100.0	100.0	617885
AIRE	133.8	100.0	100.0	240300
AKR1C2	175.5	100.0	100.0	614279
AMH	156.3	100.0	100.0	261550
AMHR2	140.9	100.0	100.0	261550
ANOS1	102.3	100.0	99.8	308700
AR	99.1	99.5	99.0	300068;312300;300633
ARHGAP35	131.0	100.0	100.0	No OMIM phenotype
ARMC5	142.9	100.0	100.0	615954
ARX	109.5	99.0	96.7	300215
ATF3	125.2	100.0	100.0	-
ATRX	115.5	99.9	99.7	309580;301040
AXL	113.7	100.0	100.0	No OMIM phenotype
B9D1	135.5	100.0	100.0	614209
BMP15	111.0	100.0	100.0	300510
BMP4	129.1	100.0	100.0	-
BMP7	132.2	100.0	100.0	-
CBX2	117.2	100.0	100.0	613080
CCDC141	135.1	99.5	98.9	-

CCNQ	120.8	100.0	99.9	300707
CDH2	139.6	100.0	100.0	618929
CDKN1C	165.2	100.0	100.0	614732
CEP41	150.6	100.0	100.0	614464
CHD7	133.9	100.0	100.0	612370
CLPP	112.2	100.0	100.0	614129
CNGA2	91.9	99.9	99.7	No OMIM phenotype
CREBBP	126.6	100.0	100.0	180849;618332
CYB5A	117.6	100.0	100.0	250790
CYP11A1	132.8	100.0	100.0	613743
CYP11B1	143.3	100.0	100.0	202010
CYP11B2	142.1	100.0	100.0	610600;203400
CYP17A1	124.2	100.0	100.0	202110
CYP19A1	149.0	100.0	99.9	613546
CYP21A2	160.4	100.0	99.9	201910
DCC	142.6	100.0	100.0	157600
DHCR7	129.7	100.0	100.0	270400
DHH	142.3	100.0	100.0	233420
DHX37	116.9	100.0	100.0	273250
DLK1	121.6	100.0	100.0	No OMIM phenotype
DMRT1	140.3	100.0	100.0	154230
DMRT2	143.1	100.0	100.0	-
DUSP6	124.9	100.0	100.0	615269
DYNC2H1	152.6	99.8	99.4	613091
EIF2B5	132.6	100.0	100.0	603896
ERAL1	120.6	100.0	100.0	617565
ESR1	129.1	100.0	99.8	615363
ESR2	150.5	100.0	100.0	618187
FANCM	150.0	100.0	100.0	609644
FEZF1	148.4	100.0	100.0	616030

FGF17	132.6	100.0	100.0	615270
FGF8	128.5	100.0	100.0	612702
FGFR1	125.7	100.0	100.0	147950
FGFR2	138.1	100.0	100.0	101200
FLRT3	148.6	100.0	99.7	615271
FOXL2	123.1	100.0	100.0	110100;608996
FRAS1	126.6	100.0	99.9	219000
FREM2	138.3	99.9	99.7	219000
FSHB	132.8	98.7	98.0	229070
FSHR	149.3	100.0	99.9	233300
FZD2	157.9	100.0	100.0	164745
GATA4	164.3	100.0	100.0	615542
GDF9	148.4	100.0	100.0	618014
GK	120.3	100.0	100.0	307030
GNRH1	149.4	100.0	100.0	614841
GNRHR	153.8	100.0	100.0	138850
GRIP1	131.8	100.0	100.0	219000
HARS2	140.0	100.0	100.0	614926
HESX1	152.7	100.0	100.0	182230
HFM1	158.9	100.0	100.0	615724
HOXA13	89.9	99.9	98.8	140000
HS6ST1	143.6	100.0	100.0	614880
HSD17B3	143.8	100.0	100.0	264300
HSD17B4	142.8	96.6	96.6	233400
HSD3B2	191.1	99.6	99.4	201810
IGSF10	148.1	100.0	100.0	-
IL17RD	125.0	100.0	100.0	615267
IRF6	126.8	100.0	100.0	119500
KAT6B	137.4	100.0	100.0	606170;603736
KISS1	191.5	100.0	100.0	614842

KISS1R	158.7	100.0	100.0	614837;176400
KLB	135.6	100.0	100.0	-
LARS2	127.2	100.0	100.0	615300
LEP	116.6	100.0	100.0	614962
LEPR	144.9	94.6	94.6	614963
LHB	232.2	100.0	100.0	228300
LHCGR	152.7	100.0	100.0	238320;176410
LHX1	128.6	100.0	100.0	-
LHX3	146.3	100.0	100.0	221750
LIPA	127.4	96.6	95.2	278000
MAMLD1	92.0	100.0	99.8	300758
MAP3K1	143.0	100.0	100.0	613762
MC2R	141.2	100.0	100.0	202200
MCM8	137.7	94.4	94.4	612885
MCM9	137.7	100.0	100.0	616185
MKKS	152.8	100.0	100.0	236700
MKRN3	154.0	100.0	100.0	615346
MRAP	134.6	100.0	100.0	607398
MSH4	143.0	100.0	100.0	-
MYRF	114.4	100.0	100.0	618280
NEK1	151.1	100.0	100.0	263520
NNT	130.2	96.4	96.3	614736
NOBOX	115.6	100.0	100.0	611548
NOS1	118.1	100.0	100.0	No OMIM phenotype
NR0B1	109.1	100.0	99.8	300200;300018
NR3C1	150.9	100.0	100.0	615962
NR3C2	137.1	100.0	100.0	177735
NR5A1	136.8	100.0	100.0	612965
NSMF	135.1	100.0	100.0	614838
PBX1	134.0	100.0	99.9	617641

PCSK1	132.5	100.0	100.0	600955
PLXNA1	137.3	100.0	100.0	-
POLE	132.3	100.0	100.0	618336
POLG	138.3	100.0	100.0	157640
POMC	149.8	100.0	100.0	609734
POR	138.4	100.0	100.0	201750
PPP1R12A	148.5	99.9	99.3	618820
PPP2R3C	151.6	100.0	100.0	618419
PROK2	143.5	100.0	100.0	610628
PROKR2	155.8	100.0	100.0	244200
PROP1	131.4	100.0	100.0	262600
PSMC3IP	120.2	100.0	100.0	614324
RIPK4	136.5	100.0	100.0	263650
ROR2	125.6	100.0	100.0	268310
RSPO1	107.4	100.0	100.0	610644
SAMD9	158.3	100.0	100.0	617053
SEMA3A	148.2	100.0	100.0	614897
SGPL1	133.8	100.0	100.0	617575
SOHLH1	131.4	100.0	100.0	617690
SOX10	142.9	100.0	100.0	-
SOX2	135.3	100.0	100.0	206900
SOX3	113.1	100.0	100.0	300833
SOX9	138.4	100.0	100.0	114290
SPATA22	161.2	100.0	100.0	No OMIM phenotype
SPIDR	119.5	100.0	100.0	619665
SPRY4	114.1	100.0	100.0	615266
SRCAP	119.0	100.0	100.0	136140
SRD5A2	150.8	100.0	100.0	264600
SRY	36.8	50.0	50.0	400045;400044
STAG3	125.1	100.0	100.0	615723

STAR	108.1	100.0	100.0	201710
SYCE1	163.9	100.0	100.0	616947
TAC3	134.6	100.0	100.0	614839
TACR3	141.0	100.0	99.8	614840
TBX19	123.6	100.0	100.0	201400
TBX3	149.6	100.0	100.0	181450
TCF12	138.9	100.0	100.0	615314
TCTN3	141.7	100.0	100.0	614815
TENM1	104.9	99.9	99.5	301700
TOE1	117.1	100.0	100.0	614969
TSPYL1	126.3	100.0	100.0	608800
TWNK	117.5	100.0	100.0	616138
TXNRD2	138.5	100.0	100.0	617825
WDR11	143.8	100.0	100.0	614858
WDR60	142.0	100.0	100.0	615503
WNT4	109.9	100.0	99.8	611812
WT1	125.9	100.0	100.0	136680;194080
ZFPM2	135.6	100.0	100.0	616067

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 is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.

Median Coverage describes the average number of reads seen across 50 exomes.

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 no value for coverage 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 1st, 2021.

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors