

WES DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY DG 3.2

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
AAAS	102.9	100.0	99.4	231550
AARS2	131.7	100.0	99.4	615889
ABCD1	80.1	76.0	72.6	300100
ADCY3	128.7	100.0	99.0	617885
AIRE	98.8	100.0	99.9	240300
AKR1C2	137.9	94.5	88.3	614279
AMH	67.1	99.4	92.9	261550
AMHR2	159.4	100.0	99.6	261550
ANOS1	81.6	89.8	88.3	308700
AR	90.1	98.1	93.6	300068;312300;300633
ARMC5	156.5	100.0	99.1	615954
ARX	41.3	82.1	67.5	300215
ATF3	108.8	99.9	97.3	-
ATRX	91.1	98.7	95.2	309580;301040
AXL	162.9	100.0	98.9	No OMIM phenotype
B9D1	91.9	85.2	85.2	614209
BMP15	102.8	100.0	98.7	300510
BMP4	175.3	100.0	100.0	-
BMP7	99.3	99.8	98.4	-
CBX2	154.6	100.0	100.0	613080
CCDC141	121.5	99.8	99.5	-
CCNQ	46.5	82.9	78.3	300707

CDKN1C	71.4	89.9	81.6	614732
CEP41	77.1	98.8	93.4	614464
CHD7	137.1	100.0	99.2	612370
CLPP	146.0	100.0	99.5	614129
CREBBP	104.9	99.6	97.8	180849;618332
CYB5A	134.1	100.0	100.0	250790
CYP11A1	119.9	99.2	94.5	613743
CYP11B1	155.9	100.0	99.9	202010
CYP11B2	154.9	100.0	99.9	610600;203400
CYP17A1	109.0	99.9	98.5	202110
CYP19A1	137.2	98.3	95.7	613546
CYP21A2	101.2	97.4	91.1	201910
DCC	124.0	100.0	100.0	157600
DHCR7	147.5	100.0	100.0	270400
DHH	134.6	100.0	100.0	233420
DMRT1	89.5	100.0	98.9	154230
DMRT2	126.1	98.9	91.6	-
DUSP6	150.2	100.0	100.0	615269
DYNC2H1	107.7	98.6	95.2	613091
EIF2B5	104.1	99.8	98.5	603896
ESR1	130.5	100.0	99.8	615363
ESR2	112.9	99.9	98.9	618187
FANCM	99.4	98.9	96.3	609644
FEZF1	152.5	100.0	100.0	616030
FGF17	153.9	100.0	100.0	615270
FGF8	123.7	97.1	87.2	612702
FGFR1	128.6	100.0	99.3	147950
FGFR2	113.3	97.6	97.0	101200
FLRT3	171.7	100.0	100.0	615271
FOXL2	72.5	99.4	94.7	110100;608996

FRAS1	124.8	100.0	99.2	219000
FREM2	158.0	99.8	98.7	219000
FSHB	104.6	100.0	100.0	229070
FSHR	87.9	99.2	97.0	233300
FZD2	146.2	100.0	97.8	164745
GATA4	84.9	87.4	78.5	615542
GDF9	144.2	100.0	100.0	618014
GK	44.9	84.2	61.8	307030
GNRH1	85.2	99.5	89.5	614841
GNRHR	133.1	100.0	100.0	138850
GRIP1	113.7	100.0	99.3	219000
HESX1	60.2	99.3	97.3	182230
HFM1	54.5	95.4	89.8	615724
HOXA13	54.9	76.4	67.3	140000
HS6ST1	59.3	93.6	86.7	614880
HSD17B3	112.2	97.8	97.8	264300
HSD17B4	114.3	95.3	92.8	233400
HSD3B2	128.3	100.0	99.7	201810
IGSF10	190.5	100.0	99.9	-
IL17RD	130.2	99.9	99.0	615267
IRF6	85.2	99.4	93.0	119500
KAT6B	148.0	99.4	98.0	606170;603736
KISS1	74.1	100.0	98.2	614842
KISS1R	109.1	100.0	99.6	614837;176400
KLB	205.0	100.0	99.9	-
LARS2	123.2	100.0	100.0	615300
LEP	172.5	100.0	99.6	614962
LEPR	113.2	94.1	92.3	614963
LHB	21.7	91.7	42.8	228300
LHCGR	114.6	96.6	92.4	238320;176410

LHX1	163.4	100.0	99.8	-
LHX3	85.9	96.6	96.2	221750
LIPA	105.7	96.9	94.6	278000
MAMLD1	128.4	99.7	97.8	300758
MAP3K1	137.3	97.0	93.0	613762
MC2R	152.7	99.7	97.4	202200
MCM8	113.6	99.9	98.8	612885
MCM9	132.6	99.9	99.0	616185
MKKS	204.9	100.0	100.0	236700
MKRN3	112.1	96.0	96.0	615346
MRAP	187.8	100.0	100.0	607398
MSH4	85.0	98.8	96.5	-
MYRF	144.5	99.0	97.8	618280
NEK1	120.2	99.5	98.2	263520
NNT	118.6	96.4	96.0	614736
NOBOX	95.3	99.3	96.6	611548
NR0B1	120.6	99.9	99.2	300200;300018
NR3C1	139.3	100.0	99.9	615962
NR3C2	136.9	99.9	99.8	177735
NR5A1	96.8	100.0	100.0	612965
NSMF	97.3	96.9	95.5	614838
PBX1	120.7	100.0	99.1	617641
PCSK1	147.3	99.9	99.4	600955
PLXNA1	190.3	100.0	99.9	-
PNPLA6	144.9	100.0	99.8	275400;245800
POLE	132.6	100.0	99.5	618336
POLG	113.2	99.9	98.8	157640
POMC	120.6	100.0	100.0	609734
POR	193.1	99.5	98.0	201750
PPP1R12A	122.1	97.8	95.9	618820

PROK2	106.6	99.9	98.9	610628
PROKR2	228.0	100.0	100.0	244200
PROP1	97.0	91.0	80.2	262600
PSMC3IP	94.7	100.0	100.0	614324
RIPK4	151.7	100.0	99.9	263650
ROR2	172.6	100.0	99.4	268310
RSPO1	100.7	100.0	99.9	610644
SAMD9	158.4	99.9	99.8	617053
SEMA3A	163.6	100.0	99.7	614897
SGPL1	142.2	100.0	100.0	617575
SOHLH1	123.1	99.7	96.8	617690
SOX10	69.2	99.9	97.2	-
SOX2	194.7	100.0	99.8	206900
SOX3	54.4	94.9	81.2	300833
SOX9	171.5	100.0	99.9	114290
SPRY4	155.9	100.0	100.0	615266
SRCAP	154.9	99.7	98.9	136140
SRD5A2	72.2	100.0	98.8	264600
SRY	30.3	50.0	49.9	400045;400044
STAG3	105.5	93.5	92.8	615723
STAR	135.7	100.0	99.9	201710
SYCE1	120.9	99.9	98.8	616947
TAC3	69.3	99.9	93.6	614839
TACR3	140.8	100.0	100.0	614840
TBX19	166.6	100.0	100.0	201400
TBX3	93.9	99.4	97.3	181450
TCF12	136.8	99.9	99.7	615314
TCTN3	118.9	100.0	100.0	614815
TOE1	156.3	100.0	100.0	614969
TSPYL1	142.4	100.0	100.0	608800

TWNK	173.4	100.0	99.9	616138
TXNRD2	123.8	96.8	95.9	617825
WDR11	121.8	98.2	96.5	614858
WDR60	106.7	99.3	95.8	615503
WNT4	228.0	97.8	93.6	611812
WT1	72.4	97.6	96.1	136680;194080
ZFPM2	151.2	100.0	99.9	616067

Gene symbols used follow HGNC 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.

[EAS.GenProductCoverage.pdf.footer.ad01](#)