

WES DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY DG 3.00

<i>Gene</i>	<i>Median coverage</i>	<i>% covered >10x</i>	<i>% covered >20x</i>	<i>OMIM disease ID</i>
AAAS	117.1	100.0	99.9	231550
AARS2	131.5	100.0	99.4	615889
ABCD1	78.1	75.8	71.6	300100
ADCY3	130.7	100.0	99.1	617885
AIRE	91.0	100.0	99.8	240300
AKR1C2	164.2	94.9	89.2	614279
AMH	54.3	96.4	83.8	261550
AMHR2	157.4	100.0	99.5	261550
ANOS1	97.2	89.8	88.9	308700
AR	93.5	97.6	93.3	300068;312300;300633
ARMC5	153.8	100.0	99.4	615954
ARX	38.6	81.0	64.0	300215
ATF3	119.3	99.9	97.5	-
ATRX	102.2	99.4	96.3	309580;301040
B9D1	94.7	85.2	85.1	614209
BMP15	126.1	100.0	99.3	300510
BMP4	178.6	100.0	100.0	-
BMP7	101.4	99.9	98.5	-
CBX2	148.2	100.0	99.8	613080
CCDC141	143.7	100.0	99.5	-
CCNQ	52.3	83.1	78.5	300707
CDKN1C	66.8	88.0	77.8	614732

CEP41	92.6	99.8	97.4	614464
CHD7	158.7	100.0	99.5	612370
CLPP	147.3	100.0	99.1	614129
CREBBP	124.1	99.7	98.5	180849;618332
CYB5A	157.5	100.0	100.0	250790
CYP11A1	129.8	99.3	96.1	613743
CYP11B1	146.2	100.0	100.0	202010
CYP11B2	138.4	100.0	100.0	610600;203400
CYP17A1	120.3	100.0	99.5	202110
CYP19A1	161.8	98.8	96.8	613546
CYP21A2	101.0	97.8	88.4	201910
DCC	147.1	100.0	100.0	157600
DHCR7	152.1	100.0	100.0	270400
DHH	140.3	100.0	100.0	233420
DMRT1	108.0	100.0	99.8	154230
DMRT2	158.9	97.7	88.4	-
DUSP6	157.9	100.0	100.0	615269
DYNC2H1	122.7	98.8	95.5	613091
EIF2B5	119.9	100.0	99.1	603896
ESR1	136.4	100.0	99.8	615363
ESR2	122.2	100.0	99.7	618187
FEZF1	172.6	100.0	99.9	616030
FGF17	150.4	100.0	100.0	615270
FGF8	127.7	98.2	88.9	612702
FGFR1	137.7	100.0	99.9	147950
FGFR2	134.1	97.7	97.1	101200
FLRT3	199.1	100.0	100.0	615271
FOXL2	68.7	99.7	95.5	110100;608996
FRAS1	140.3	100.0	99.4	219000
FREM2	167.6	100.0	99.3	219000

FSHB	115.6	100.0	100.0	229070
FSHR	91.9	99.5	97.2	233300
FZD2	145.0	99.9	98.2	164745
GATA4	80.9	84.1	74.5	615542
GDF9	171.0	100.0	100.0	618014
GK	56.7	88.9	70.4	307030
GNRH1	100.9	100.0	93.7	614841
GNRHR	156.3	100.0	100.0	138850
GRIP1	137.0	100.0	99.7	219000
HESX1	80.5	99.7	97.3	182230
HFM1	60.8	96.3	89.6	615724
HOXA13	66.0	77.7	69.0	140000
HS6ST1	54.3	92.9	84.5	614880
HSD17B3	142.1	97.8	97.8	264300
HSD17B4	130.0	95.4	93.1	233400
HSD3B2	151.4	100.0	99.7	201810
IGSF10	220.0	100.0	100.0	-
IL17RD	145.4	99.9	99.1	615267
IRF6	91.7	99.6	95.9	119500
KISS1	62.5	100.0	98.3	614842
KISS1R	109.4	100.0	99.5	614837;176400
KLB	225.5	100.0	99.9	-
LARS2	143.2	100.0	100.0	615300
LEP	198.4	99.9	97.3	614962
LEPR	128.3	94.3	92.6	614963
LHB	21.0	90.4	38.9	228300
LHCGR	142.3	94.1	92.3	238320;176410
LHX1	156.9	100.0	99.6	-
LHX3	86.2	96.6	96.5	221750
LIPA	131.2	99.2	95.2	278000

MAMLD1	125.4	99.8	98.2	300758
MAP3K1	162.9	96.1	91.6	613762
MC2R	183.7	99.9	98.3	202200
MCM8	136.8	100.0	99.6	612885
MCM9	153.0	99.9	99.8	616185
MKKS	232.6	100.0	100.0	236700
MKRN3	143.5	96.0	96.0	615346
MRAP	180.8	100.0	100.0	607398
MYRF	146.9	99.3	98.5	618280
NEK1	141.3	99.8	98.0	263520
NNT	143.9	96.4	95.9	614736
NOBOX	107.3	99.9	98.4	611548
NR0B1	132.2	100.0	99.5	300200;300018
NR3C1	164.1	100.0	99.9	615962
NR3C2	156.4	100.0	99.7	177735
NR5A1	84.4	100.0	100.0	612965
NSMF	105.0	96.1	95.6	614838
PBX1	146.9	100.0	99.4	617641
PCSK1	175.2	100.0	99.5	600955
PLXNA1	177.0	100.0	99.6	-
PNPLA6	139.7	100.0	99.7	275400;245800
POLE	140.6	100.0	99.8	618336
POMC	114.0	100.0	100.0	609734
POR	175.0	99.8	98.6	201750
PROK2	136.2	99.9	98.5	610628
PROKR2	228.4	100.0	100.0	244200
PROP1	98.8	92.6	82.6	262600
PSMC3IP	116.0	100.0	100.0	614324
RIPK4	144.6	100.0	99.9	263650
ROR2	168.7	100.0	99.9	268310

RSPO1	96.2	100.0	99.9	610644
SAMD9	185.7	100.0	99.8	617053
SEMA3A	196.5	100.0	99.9	614897
SGPL1	159.8	100.0	100.0	617575
SOHLH1	117.4	99.7	96.5	617690
SOX10	70.3	99.9	97.9	-
SOX2	207.1	100.0	100.0	206900
SOX3	60.2	91.4	75.2	300833
SOX9	161.0	100.0	98.6	114290
SPRY4	153.2	100.0	100.0	615266
SRCAP	168.1	99.4	98.9	136140
SRD5A2	81.7	99.9	99.0	264600
SRY	35.5	50.0	50.0	400045;400044
STAG3	121.4	93.5	93.2	615723
STAR	148.9	100.0	100.0	201710
SYCE1	114.0	100.0	98.6	616947
TAC3	79.3	100.0	99.6	614839
TACR3	150.3	100.0	100.0	614840
TBX19	185.4	100.0	100.0	201400
TBX3	105.8	99.2	96.8	181450
TCF12	155.4	100.0	99.9	615314
TCTN3	136.4	100.0	100.0	614815
TOE1	170.4	100.0	100.0	614969
TSPYL1	147.6	100.0	100.0	608800
TWNK	202.8	100.0	100.0	616138
TXNRD2	121.2	96.8	95.9	617825
WDR11	139.6	98.0	96.5	614858
WDR60	121.8	99.5	97.0	615503
WNT4	200.3	99.1	94.8	611812
WT1	81.1	97.3	95.4	136680;194080

ZFPM2	181.3	100.0	100.0	616067
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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.

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 Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : October 1st, 2016.

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