WES DISORDERS/DIFFERENCES OF SEX DEVELOPMENT (DSD) / PRIMARY ADRENAL INSUFFICIENCY DG 3.4

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
AAAS	170.3	100.0	100.0	231550
AARS2	191.4	100.0	100.0	615889
ABCD1	212.8	100.0	100.0	300100
ADCY3	178.8	100.0	100.0	617885
AIRE	192.6	100.0	100.0	240300
AKR1C2	198.6	100.0	100.0	614279
AMH	234.7	100.0	100.0	261550
AMHR2	203.3	100.0	100.0	261550
ANOS1	166.7	100.0	100.0	308700
AR	195.5	100.0	100.0	300068;312300;300633
ARMC5	254.8	100.0	100.0	615954
ARX	176.0	99.0	96.8	300215
ATF3	147.0	100.0	100.0	-
ATRX	156.7	100.0	100.0	309580;301040
AXL	191.2	100.0	100.0	No OMIM phenotype
B9D1	189.5	96.6	94.1	614209
BMP15	161.6	100.0	100.0	300510
BMP4	222.6	100.0	100.0	-
BMP7	205.4	100.0	100.0	-
CBX2	233.8	100.0	100.0	613080
CCDC141	156.3	100.0	100.0	-
CCNQ	166.4	99.9	99.8	300707

CDH2	178.2	100.0	100.0	618929
CDKN1C	232.6	100.0	100.0	614732
CEP41	150.3	100.0	100.0	614464
CHD7	185.7	100.0	100.0	612370
CLPP	190.0	100.0	100.0	614129
CNGA2	181.7	100.0	100.0	No OMIM phenotype
CREBBP	224.8	100.0	100.0	180849;618332
CYB5A	137.6	100.0	100.0	250790
CYP11A1	188.0	100.0	100.0	613743
CYP11B1	252.6	100.0	100.0	202010
CYP11B2	247.2	100.0	100.0	610600;203400
CYP17A1	177.3	100.0	100.0	202110
CYP19A1	172.7	100.0	100.0	613546
CYP21A2	378.7	100.0	100.0	201910
DCC	175.2	100.0	100.0	157600
DHCR7	189.2	100.0	100.0	270400
DHH	247.5	100.0	100.0	233420
DMRT1	220.4	100.0	100.0	154230
DMRT2	214.3	100.0	100.0	-
DUSP6	205.3	100.0	100.0	615269
DYNC2H1	143.7	100.0	100.0	613091
EIF2B5	167.3	100.0	100.0	603896
ERAL1	171.6	100.0	100.0	617565
ESR1	211.9	100.0	100.0	615363
ESR2	191.2	100.0	100.0	618187
FANCM	152.5	100.0	100.0	609644
FEZF1	235.0	100.0	100.0	616030
FGF17	206.9	100.0	100.0	615270
FGF8	200.6	100.0	100.0	612702
FGFR1	214.8	100.0	100.0	147950

FGFR2	193.1	100.0	100.0	101200
FLRT3	191.9	100.0	100.0	615271
FOXL2	198.3	100.0	100.0	110100;608996
FRAS1	171.0	100.0	100.0	219000
FREM2	202.1	100.0	100.0	219000
FSHB	159.2	100.0	100.0	229070
FSHR	189.6	100.0	100.0	233300
FZD2	254.1	100.0	100.0	164745
GATA4	203.9	100.0	100.0	615542
GDF9	200.3	100.0	100.0	618014
GK	143.3	100.0	100.0	307030
GNRH1	168.3	100.0	100.0	614841
GNRHR	183.2	100.0	100.0	138850
GRIP1	168.6	100.0	100.0	219000
HARS2	154.5	100.0	100.0	614926
HESX1	156.6	100.0	100.0	182230
HFM1	134.4	100.0	100.0	615724
HOXA13	152.6	100.0	99.7	140000
HS6ST1	225.5	100.0	100.0	614880
HSD17B3	185.2	99.0	99.0	264300
HSD17B4	137.6	96.6	96.6	233400
HSD3B2	242.3	100.0	100.0	201810
IGSF10	196.5	100.0	100.0	-
IL17RD	173.0	100.0	100.0	615267
IRF6	187.9	100.0	100.0	119500
KAT6B	213.9	100.0	100.0	606170;603736
KISS1	222.3	100.0	100.0	614842
KISS1R	235.7	100.0	100.0	614837;176400
KLB	215.9	100.0	100.0	-
LARS2	171.4	100.0	100.0	615300

LEP	177.2	100.0	100.0	614962
LEPR	148.5	94.6	94.6	614963
LHB	352.7	100.0	100.0	228300
LHCGR	172.2	100.0	100.0	238320;176410
LHX1	289.5	100.0	100.0	-
LHX3	215.5	100.0	100.0	221750
LIPA	155.9	95.2	95.2	278000
MAMLD1	215.1	100.0	100.0	300758
MAP3K1	192.0	100.0	100.0	613762
MC2R	216.4	100.0	100.0	202200
MCM8	154.1	94.4	94.4	612885
MCM9	186.3	100.0	100.0	616185
MKKS	185.5	100.0	100.0	236700
MKRN3	222.5	96.0	96.0	615346
MRAP	208.8	100.0	100.0	607398
MSH4	128.9	100.0	100.0	-
MYRF	195.2	100.0	100.0	618280
NEK1	142.5	100.0	100.0	263520
NNT	158.2	96.4	96.4	614736
NOBOX	160.2	100.0	100.0	611548
NR0B1	233.8	100.0	100.0	300200;300018
NR3C1	163.9	100.0	100.0	615962
NR3C2	186.2	100.0	100.0	177735
NR5A1	225.9	100.0	100.0	612965
NSMF	194.9	100.0	100.0	614838
PBX1	199.2	100.0	100.0	617641
PCSK1	177.5	100.0	100.0	600955
PLXNA1	242.4	100.0	100.0	-
POLE	190.0	100.0	100.0	618336
POLG	195.1	100.0	100.0	157640

POMC	211.1	100.0	100.0	609734
POR	213.0	100.0	100.0	201750
PPP1R12A	154.5	100.0	100.0	618820
PROK2	152.9	100.0	100.0	610628
PROKR2	211.7	100.0	100.0	244200
PROP1	260.4	100.0	100.0	262600
PSMC3IP	160.4	100.0	100.0	614324
REC8	181.4	100.0	100.0	No OMIM phenotype
RIPK4	239.3	100.0	100.0	263650
ROR2	209.3	97.0	97.0	268310
RSPO1	204.2	100.0	100.0	610644
SAMD9	169.2	100.0	100.0	617053
SEMA3A	163.1	100.0	100.0	614897
SGPL1	161.5	100.0	100.0	617575
SOHLH1	199.6	100.0	100.0	617690
SOX10	254.5	100.0	100.0	-
SOX2	323.4	100.0	100.0	206900
SOX3	235.9	100.0	100.0	300833
SOX9	258.7	100.0	100.0	114290
SPRY4	231.0	100.0	100.0	615266
SRCAP	202.9	100.0	100.0	136140
SRD5A2	240.2	100.0	100.0	264600
SRY	66.6	50.0	50.0	400045;400044
STAG3	157.2	100.0	100.0	615723
STAR	160.3	100.0	100.0	201710
SYCE1	202.6	100.0	100.0	616947
TAC3	135.9	100.0	100.0	614839
TACR3	211.1	100.0	100.0	614840
TBX19	194.8	100.0	100.0	201400
TBX3	252.2	100.0	100.0	181450

TCF12	165.0	100.0	100.0	615314
TCTN3	169.8	100.0	100.0	614815
TOE1	181.6	100.0	100.0	614969
TSPYL1	198.7	100.0	100.0	608800
TWNK	197.1	100.0	100.0	616138
TXNRD2	175.0	100.0	100.0	617825
WDR11	159.1	100.0	100.0	614858
WDR60	166.4	100.0	100.0	615503
WNT4	199.3	100.0	99.8	611812
WT1	215.2	97.7	97.7	136680;194080
ZFPM2	194.5	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