DISORDERS OF SEX DEVELOPMENT GENE PANEL DG 2.5/2.6

Gene name	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AKR1C2	153.9	87%	84.00%	46XY sex reversal 8,614279 Obesity,hyperphagia and developmental delay
AMH	34.8	93%	72.00%	Persistent Mullerian duct syndrome, type I,261550
AMHR2	136.6	100%	100.00%	Persistent Mullerian duct syndrome, type II,261550
AR	111.1	94%	89.00%	Androgen insensitivity,300068 Androgen insensitivity,partial,with/without breast cancer,312300 Hypospadias 1,X-linked,300633 Spinal and bulbar muscular atrophy of Kennedy,313200 {Prostate cancer,susceptibility to},176807
ARX	45.4	84%	82.00%	Epileptic encephalopathy,early infantile,1,308350 Hydranencephaly with abnormal genitalia,300215 Lissencephaly,X-linked 2,300215 Mental retardation,X-linked 29 and others,300419 Partington syndrome,309510 Proud syndrome,300004
ATRX	110.4	98%	97.00%	Alpha-thalassemia myelodysplasia syndrome, somatic,300448 Alpha-thalassemia/mental retardation syndrome,301040 Mental retardation-hypotonic facies syndrome,X-linked,309580
B9D1	97.6	92%	91.00%	Meckel syndrome 9,614209
CBX2	76	100%	98.00%	46XY sex reversal 5,613080
CEP41	76	95%	86.00%	Joubert syndrome 15,614464
CYB5A	119.4	100%	100.00%	Methemoglobinemia, type IV,250790
CYP11A1	117.8	99%	96.00%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete,613743
CYP11B1	149.4	100%	100.00%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency,202010 Aldosteronism,glucocorticoid-remediable,103900
CYP17A1	129.4	100%	100.00%	17,20-lyase deficiency, isolated,202110 17-alpha-hydroxylase/17,20-lyase deficiency,202110

CYP19A1	178.1	100%	100.00%	Aromatase deficiency,613546 Aromatase excess syndrome,139300
DHCR7	148.3	100%	100.00%	Smith-Lemli-Opitz syndrome,270400
DHH	93.4	100%	100.00%	46XY sex reversal 7,233420 46XY partial gonadal dysgenesis, with minifascicular neuropathy,607080
DMRT1	83.3	98%	94.00%	No OMIM phenotype XY sex reversal (Raymond (1999) Hum Mol Genet 8, 989) XY gonadal dysgenesis (Ledig (2010) Hum Reprod 25,2637) Azoospermia (Lopes (2013) PLoS Genet 9,e1003349) Male infertility (Tewes (2014) Fertil Steril 102, 816)
DYNC2H1	91.1	98%	86.00%	Short-rib thoracic dysplasia 3 with or without polydactyly,613091
FRAS1	144.2	100%	99.00%	Fraser syndrome,219000
FREM2	162.2	99%	97.00%	Fraser syndrome,219000
GATA4	69.9	75%	59.00%	?Testicular anomalies with or without congenital heart disease,615542 Atrial septal defect 2,607941 Atrioventricular septal defect 4,614430 Tetralogy of Fallot,187500 Ventricular septal defect 1,614429
GRIP1	127.7	100%	98.00%	Fraser syndrome,219000
HOXA13	37.4	73%	62.00%	Hand-foot-uterus syndrome,140000 Guttmacher syndrome,176305
HSD17B3	162	100%	100.00%	Pseudohermaphroditism, male, with gynecomastia,264300
HSD3B2	148.9	100%	100.00%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency,201810
LHCGR	142.2	96%	92.00%	Leydig cell adenoma,somatic,with precocious puberty,176410 Leydig cell hypoplasia with hypergonadotropic hypogonadism,238320 Leydig cell hypoplasia with pseudohermaphroditism,238320 Luteinizing hormone resistance,female,238320 Precocious puberty,male,
MAMLD1	155	100%	100.00%	Hypospadias 2,X-linked,300758
MAP3K1	148.5	93%	86.00%	46XY sex reversal 6,613762
MKKS	224.7	89%	89.00%	McKusick-Kaufman syndrome,236700 Bardet-Biedl syndrome 6,605231
NEK1	104.2	98%	92.00%	Short-rib thoracic dysplasia 6 with or without polydactyly,263520
NR0B1	144	100%	96.00%	46XY sex reversal 2, dosage-sensitive,300018

				Adrenal hypoplasia,congenital,with hypogonadotropic hypogonadism,300200
NR3C1	137.3	100%	100.00%	Glucocorticoid resistance,615962
NR5A1	81.3	100%	96.00%	46XY sex reversal 3,612965 Aderenocortical insufficiency Premature ovarian failure 7,612964 Spermatogenic failure 8,613957
POR	144.2	100%	99.00%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis,201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase,613571
RIPK4	139.1	100%	99.00%	Popliteal pterygium syndrome 2, lethal type,263650
ROR2	158.5	99%	98.00%	Robinow syndrome, autosomal recessive,268310 Brachydactyly,type B1,113000
RSPO1	105.5	100%	100.00%	Palmoplantar hyperkeratosis and true hermaphroditism,610644 Palmoplantar hyperkeratosis with squamous cell carcinoma and sex reversal,610644
SOX3	49.9	97%	92.00%	Mental retardation,X-linked,with isolated growth hormone deficiency,300123 Panhypopituitarism,X-linked,312000
SOX9	106.9	94%	89.00%	Campomelic dysplasia with autosomal sex reversal,114290 Acampomelic campomelic dysplasia,114290 Campomelic dysplasia,114290
SRD5A2	80.6	100%	99.00%	Pseudovaginal perineoscrotal hypospadias,264600
SRY	1.7	1%	1.00%	46XX sex reversal 1,400045 46XY sex reversal 1,400044
STAR	114.6	100%	100.00%	Lipoid adrenal hyperplasia,201710
TCTN3	118.2	100%	99.00%	Joubert syndrome 18,614815 Orofaciodigital syndrome IV,258860
TSPYL1	125.4	100%	100.00%	Sudden infant death with dysgenesis of the testes syndrome,608800
WDR60	103.6	97%	96.00%	Short-rib thoracic dysplasia 8 with or without polydactyly,615503
WNT4	228	92%	92.00%	SERKAL syndrome,611812 Mullerian aplasia and hyperandrogenism,158330
WT1	81.9	99%	94.00%	Denys-Drash syndrome,194080 Frasier syndrome,136680 Meacham syndrome,608978 Mesothelioma,somatic,156240 Nephrotic syndrome,type 4,256370 Wilms tumor,type 1,194070

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.
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 : April 10th, 2016.
This list is accurate for panel versions DG 2.5 and DG 2.6. From DG 2.5 to DG 2.6 no changes were made to the content of the gene panels.

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