DISORDERS OF SEX DEVELOPMENT GENE PANEL DG 2.15 (56 genes)

Gene	*Median Coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AKR1C2	179.5	96.3	89.6	46XY sex reversal 8, 614279
AMH	42.1	92.8	74.2	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	158	100	99.5	Persistent Mullerian duct syndrome, type II, 261550
AR	85.3	93.8	88.3	Androgen insensitivity, 300068 Androgen insensitivity, partial, with or without breast cancer, 312300 Hypospadias 1, X-linked, 300633 Spinal and bulbar muscular atrophy of Kennedy, 313200 {Prostate cancer, susceptibility to}, 176807
ARX	29.1	75.8	59.5	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
ATF3	138.1	99.8	98.7	No OMIM phenotype Eur J Endocrinol. 2008 May 158(5):729-39. doi
ATRX	82.6	98.2	92.2	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
B9D1	115.3	92.1	91.4	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
CBX2	96.6	99.9	98.4	?46XY sex reversal 5, 613080
CEP41	83.5	97.7	89.6	Joubert syndrome 15, 614464
CYB5A	133.5	100	100	Methemoglobinemia and ambiguous genitalia, 250790
CYP11A1	123.9	99.6	97.7	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	175.9	100	100	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP17A1	135	100	99.7	17,20-lyase deficiency, isolated, 202110

				17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	160.6	99.1	97.3	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP21A2	93.8	95.8	86.6	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
DHCR7	158.3	100	100	Smith-Lemli-Opitz syndrome, 270400
DHH	117.7	100	100	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DMRT1	99.4	99.3	92.4	No OMIM phenotype XY gonadal dysgenesis (Ledig (2010) Hum Reprod 25,2637) Azoospermia (Lopes (2013) PLoS Genet 9,e1003349) ?Male infertility (Tewes (2014) Fertil Steril 102, 816) ?XY sex reversal (Raymond (1999) Hum Mol Genet 8, 989)
DMRT2	154.3	98.4	90.6	No OMIM phenotype 46,XY DSD (deletion)
DYNC2H1	90.5	96.6	87	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
FAM58A	73.2	82.8	78.8	STAR syndrome, 300707
FGFR2	140.1	97.4	96.4	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Apert syndrome, 101200 Beare-Stevenson cutis gyrata syndrome, 123790 Bent bone dysplasia syndrome, 614592 Craniofacial-skeletal-dermatologic dysplasia, 101600 Craniosynostosis, nonspecific, 0 Crouzon syndrome, 123500 Gastric cancer, somatic, 613659 Jackson-Weiss syndrome, 123150 LADD syndrome, 149730 Pfeiffer syndrome, 101600 Saethre-Chotzen syndrome, 101400 Scaphocephaly and Axenfeld-Rieger anomaly, 0 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FOXL2	39.5	92.8	71	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996
FRAS1	147.8	100	99.7	Fraser syndrome 1, 219000

FREM2	182.4	100	99.5	Fraser syndrome 2, 617666
GATA4	87.4	68.6	60.7	?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	130.8	100	99.9	Fraser syndrome 3, 617667
HOXA13	49	69.2	61.7	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HSD17B3	156.4	100	100	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD3B2	189.2	100	100	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
LHCGR	154.5	95.4	92.8	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, 176410
MAMLD1	131.4	99.9	98.3	Hypospadias 2, X-linked, 300758
MAP3K1	166	92.9	89.2	46XY sex reversal 6, 613762
MKKS	208.5	83.2	83.1	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
NEK1	103.2	98.1	93	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NR0B1	119.3	99.9	98.6	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, 300200
NR3C1	137.4	100	99.8	Glucocorticoid resistance, 615962
NR5A1	79.9	100	98.3	46, XX sex reversal 4, 617480 46XY sex reversal 3, 612965 Adrenocortical insufficiency, 612964 Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957
POR	167.7	99.9	98.7	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
RIPK4	163.3	100	99.6	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type, 263650

ROR2	165.9	99.4	98	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RSPO1	109.7	100	100	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
SOX3	37.7	86.4	71.5	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	134	97.8	93.8	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SRCAP	153.9	99.8	99.1	Floating-Harbor syndrome, 136140
SRD5A2	77.6	100	96.4	Pseudovaginal perineoscrotal hypospadias, 264600
SRY	46.1	50	50	46XX sex reversal 1, 400045 46XY sex reversal 1, 400044
STAR	124	100	100	Lipoid adrenal hyperplasia, 201710
TCTN3	127.6	100	99.8	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TOE1	165.1	100	100	Pontocerebellar hypoplasia, type 7, 614969
TSPYL1	141.5	100	99.4	Sudden infant death with dysgenesis of the testes syndrome, 608800
WDR60	114.2	99.1	96.3	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNT4	263.1	93.4	92.7	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	76.5	91.8	81.4	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
ZFPM2	196.3	100	99.6	46XY sex reversal 9, 616067 Diaphragmatic hernia 3, 610187 Tetralogy of Fallot, 187500

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 : December 31st, 2018.
This list is accurate for panel version DG 2.15
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors