DISORDERS OF SEX DEVELOPMENT GENE PANEL DG 2.9

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated Phenotype description and OMIM disease ID*
AKR1C2	201.9	96%	90%	46XY sex reversal 8, 614279 Obesity, hyperphagia, and developmental delay
AMH	46	97%	85%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	172.3	100%	99%	Persistent Mullerian duct syndrome, type II, 261550
AR	98.2	95%	90%	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	38.3	81%	67%	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	105.7	98%	95%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
B9D1	127.2	92%	91%	?Meckel syndrome 9, 614209
CBX2	111.7	99%	97%	?46XY sex reversal 5, 613080
CEP41	98.2	98%	93%	Joubert syndrome 15, 614464
CYB5A	141	100%	100%	Methemoglobinemia, type IV, 250790
CYP11A1	142.4	99%	97%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	187.7	100%	99%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP17A1	143.7	100%	99%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	200.3	100%	100%	Aromatase deficiency, 613546

				Aromatase excess syndrome, 139300
DHCR7	173	100%	100%	Smith-Lemli-Opitz syndrome, 270400
DHH	134.9	100%	99%	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DMRT1	107.5	99%	96%	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	166.7	99%	96%	No OMIM phenotype
DYNC2H1	110.6	98%	91%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
FGFR2	155.6	97%	96%	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 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 Scaphocephaly, maxillary retrusion, and mental retardation, 609579
FOXL2	43	94%	78%	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996
FRAS1	168.5	100%	99%	Fraser syndrome, 219000
FREM2	194.3	99%	99%	Fraser syndrome, 219000
GATA4	98.9	74%	63%	Atrial septal defect 2, 607941 Atrioventricular septal defect 4, 614430 Tetralogy of Fallot, 187500 Ventricular septal defect 1, 614429 ?Testicular anomalies with or without congenital heart disease, 615542

GRIP1	148.9	100%	99%	Fraser syndrome, 219000
HOXA13	58	73%	66%	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HSD17B3	161.7	100%	100%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD3B2	181.3	100%	100%	3-beta-hydroxysteroid dehydrogenase, type II, deficiency, 201810
LHCGR	188	97%	93%	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	145.9	99%	98%	Hypospadias 2, X-linked, 300758
MAP3K1	187.2	94%	89%	46XY sex reversal 6, 613762
MKKS	216.9	89%	89%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
NEK1	142.4	99%	96%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NR0B1	130	99%	98%	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, with hypogonadotropic hypogonadism, 300200
NR3C1	165.1	100%	100%	Glucocorticoid resistance, 615962
NR5A1	102.1	100%	99%	46XY sex reversal 3, 612965 Adrenocortical insufficiency Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957
POR	182	100%	99%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
RIPK4	163.1	100%	99%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
ROR2	190.4	99%	99%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RSPO1	128.8	100%	100%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
SOX3	47	94%	81%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	138.7	97%	93%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290 Campomelic dysplasia, 114290
SRD5A2	96.1	99%	97%	Pseudovaginal perineoscrotal hypospadias, 264600

SRY	58.9	50%	50%	46XX sex reversal 1, 400045 46XY sex reversal 1, 400046
STAR	147.6	100%	100%	Lipoid adrenal hyperplasia, 201710
TCTN3	133.6	100%	99%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TSPYL1	154	100%	99%	Sudden infant death with dysgenesis of the testes syndrome, 608800
WDR60	131.2	99%	97%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNT4	284.8	93%	92%	Mullerian aplasia and hyperandrogenism, 158330 SERKAL syndrome, 611812
WT1	94.2	96%	89%	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
ZFPM2	208.3	100%	99%	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 : April 14th 2017
This list is accurate for panel version DG 2.9

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