DISORDERS OF SEX DEVELOPMENT GENE PANEL DG 2.17 (56 genes)

Releasedate: 06-12-2019

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
AKR1C2	137.7	95.9%	90.1%	46XY sex reversal 8, 614279
AMH	97.3	100.0%	99.3%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	152.1	100.0%	99.4%	Persistent Mullerian duct syndrome, type II, 261550
AR	97.8	98.9%	95.3%	Hypospadias 1, X-linked, 300633 Androgen insensitivity, 300068 Androgen insensitivity, partial, with or without breast cancer, 312300 Spinal and bulbar muscular atrophy of Kennedy, 313200
ARX	58.2	90.9%	83.3%	Proud syndrome, 300004 Lissencephaly, X-linked 2, 300215 Partington syndrome, 309510 Epileptic encephalopathy, early infantile, 1, 308350 Mental retardation, X-linked 29 and others, 300419 Hydranencephaly with abnormal genitalia, 300215
ATF3	123.9	99.8%	95.6%	No OMIM Disease ID
ATRX	86.2	99.1%	95.1%	Alpha-thalassemia/mental retardation syndrome, 301040 Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Mental retardation-hypotonic facies syndrome, X-linked, 309580
B9D1	111.4	92.2%	92.2%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
CBX2	168.8	100.0%	100.0%	?46XY sex reversal 5, 613080
CEP41	79.1	98.7%	94.4%	Joubert syndrome 15, 614464
CYB5A	137.4	100.0%	100.0%	Methemoglobinemia and ambiguous genitalia, 250790
CYP11A1	130.1	99.2%	94.8%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	171.1	100.0%	100.0%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP17A1	116.2	100.0%	99.8%	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 17,20-lyase deficiency, isolated, 202110
CYP19A1	128.2	99.7%	97.7%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300
CYP21A2	102.4	99.9%	97.2%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910

DHCR7	158.7	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DHH	181.5	100.0%	100.0%	46XY sex reversal 7, 233420 46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080
DMRT1	108.2	100.0%	99.7%	No OMIM Disease ID
DMRT2	155.1	100.0%	99.7%	No OMIM Disease ID
DYNC2H1	98.0	98.9%	94.3%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
FAM58A	57.8	83.7%	78.2%	STAR syndrome, 300707
FGFR2	118.0	97.7%	97.1%	Apert syndrome, 101200 Jackson-Weiss syndrome, 123150 Saethre-Chotzen syndrome, 101400 Gastric cancer, somatic, 613659 Scaphocephaly, maxillary retrusion, and mental retardation, 609579 Bent bone dysplasia syndrome, 614592 LADD syndrome, 149730 Craniofacial-skeletal-dermatologic dysplasia, 101600 Pfeiffer syndrome, 101600 Crouzon syndrome, 123500 Beare-Stevenson cutis gyrata syndrome, 123790 Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 Craniosynostosis, nonspecific, 0 Scaphocephaly and Axenfeld-Rieger anomaly, 0
FOXL2	144.6	100.0%	99.9%	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996 Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100
FRAS1	123.1	99.9%	99.3%	Fraser syndrome 1, 219000
FREM2	160.8	100.0%	99.6%	Fraser syndrome 2, 617666 Cryptophthalmos, unilateral or bilateral, isolated, 123570
GATA4	95.7	98.9%	90.6%	?Testicular anomalies with or without congenital heart disease, 615542 Tetralogy of Fallot, 187500 Atrioventricular septal defect 4, 614430 Atrial septal defect 2, 607941 Ventricular septal defect 1, 614429
GRIP1	114.2	100.0%	99.4%	Fraser syndrome 3, 617667
HOXA13	90.2	95.8%	84.5%	?Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HSD17B3	119.0	100.0%	100.0%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD3B2	137.1	100.0%	99.9%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810

LHCGR	141.5	98.5%	94.3%	Leydig cell adenoma, somatic, with precocious puberty, 176410 Precocious puberty, male, 176410 Luteinizing hormone resistance, female, 238320 Leydig cell hypoplasia with pseudohermaphroditism, 238320 Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320
MAMLD1	134.7	99.8%	98.4%	Hypospadias 2, X-linked, 300758
MAP3K1	147.4	99.8%	98.2%	46XY sex reversal 6, 613762
MKKS	161.5	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
NEK1	111.2	99.9%	98.0%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520
NR0B1	155.1	100.0%	99.3%	Adrenal hypoplasia, congenital, 300200 46XY sex reversal 2, dosage-sensitive, 300018
NR3C1	131.2	100.0%	100.0%	Glucocorticoid resistance, 615962
NR5A1	126.3	100.0%	100.0%	Adrenocortical insufficiency, 612964 46, XX sex reversal 4, 617480 Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957 46XY sex reversal 3, 612965
POR	195.6	99.4%	97.5%	Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750
RIPK4	189.1	100.0%	100.0%	Popliteal pterygium syndrome, Bartsocas-Papas type, 263650 CHAND syndrome, 214350
ROR2	176.7	100.0%	99.9%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RSPO1	115.6	100.0%	100.0%	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 Palmoplantar hyperkeratosis and true hermaphroditism, 610644
SOX3	87.5	99.0%	95.2%	Panhypopituitarism, X-linked, 312000 Mental retardation, X-linked, with isolated growth hormone deficiency, 300123
SOX9	181.5	100.0%	100.0%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SRCAP	166.7	100.0%	99.8%	Floating-Harbor syndrome, 136140
SRD5A2	92.3	100.0%	97.6%	Pseudovaginal perineoscrotal hypospadias, 264600
SRY	31.7	50.0%	50.0%	46XY sex reversal 1, 400044
STAR	146.4	100.0%	100.0%	Lipoid adrenal hyperplasia, 201710
TCTN3	121.0	100.0%	100.0%	Orofaciodigital syndrome IV, 258860 Joubert syndrome 18, 614815
TOE1	153.2	100.0%	100.0%	Pontocerebellar hypoplasia, type 7, 614969

TSPYL1	159.7	100.0%	100.0%	Sudden infant death with dysgenesis of the testes syndrome, 608800
WDR60	111.7	99.8%	97.8%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNT4	254.1	99.9%	99.1%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	96.5	100.0%	99.6%	Mesothelioma, somatic, 156240 Wilms tumor, type 1, 194070 Frasier syndrome, 136680 Denys-Drash syndrome, 194080 Meacham syndrome, 608978 Nephrotic syndrome, type 4, 256370
ZFPM2	160.7	100.0%	99.9%	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 11th , 2019.
This list is accurate for panel version DG 2.17
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors