DISORDERS OF SEX DEVELOPMENT GENE PANEL DG 2.16 (56 genes)

Releasedate: 07-06-2019

Gene	*Median coverage*	*% covered > 10x*	*% covered > 20x*	*Associated phenotype description and OMIM disease ID*
AKR1C2	135,8	94.9%	87.9%	46XY sex reversal 8, 614279
AMH	82,2	99.9%	98.2%	Persistent Mullerian duct syndrome, type I, 261550
AMHR2	143,2	100.0%	99.4%	Persistent Mullerian duct syndrome, type II, 261550
AR	90,8	98.1%	93.7%	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	49,3	87.3%	79.2%	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	115,7	99.6%	95.8%	No OMIM phenotype
ATRX	89,2	99.1%	95.5%	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 Alpha-thalassemia/mental retardation syndrome, 301040 Mental retardation-hypotonic facies syndrome, X-linked, 309580
B9D1	103,7	92.2%	92.1%	?Meckel syndrome 9, 614209 Joubert syndrome 27, 617120
CBX2	149,7	100.0%	100.0%	?46XY sex reversal 5, 613080
CEP41	78,2	98.9%	94.4%	Joubert syndrome 15, 614464
CYB5A	132,5	100.0%	100.0%	Methemoglobinemia and ambiguous genitalia, 250790
CYP11A1	121,2	99.2%	95.0%	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743
CYP11B1	155,9	100.0%	100.0%	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 Aldosteronism, glucocorticoid-remediable, 103900
CYP17A1	108,5	100.0%	99.6%	17,20-lyase deficiency, isolated, 202110 17-alpha-hydroxylase/17,20-lyase deficiency, 202110
CYP19A1	125,7	99.4%	97.3%	Aromatase deficiency, 613546 Aromatase excess syndrome, 139300

CYP21A2	91,6	99.2%	93.4%	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910
DHCR7	144,9	100.0%	100.0%	Smith-Lemli-Opitz syndrome, 270400
DHH	164,9	100.0%	100.0%	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 46XY sex reversal 7, 233420
DMRT1	100,5	100.0%	99.4%	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	144,4	100.0%	99.6%	No OMIM phenotype
DYNC2H1	102,2	98.8%	95.5%	Short-rib thoracic dysplasia 3 with or without polydactyly, 613091
FAM58A	NC	NC	NC	STAR syndrome, 300707
FGFR2	113,1	97.7%	96.8%	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	117,7	100.0%	98.8%	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 Premature ovarian failure 3, 608996
FRAS1	119,2	99.9%	99.2%	Fraser syndrome 1, 219000
FREM2	152,4	100.0%	99.5%	Cryptophthalmos, unilateral or bilateral, isolated, 123570 Fraser syndrome 2, 617666
GATA4	87,6	95.9%	86.7%	?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	111,1	100.0%	99.3%	Fraser syndrome 3, 617667
HOXA13	77,7	90.9%	79.6%	Guttmacher syndrome, 176305 Hand-foot-uterus syndrome, 140000
HSD17B3	116,4	100.0%	99.9%	Pseudohermaphroditism, male, with gynecomastia, 264300
HSD3B2	131,8	100.0%	99.9%	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810
LHCGR	137	97.9%	94.1%	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	125	99.8%	98.2%	Hypospadias 2, X-linked, 300758
MAP3K1	144	99.4%	97.0%	46XY sex reversal 6, 613762
MKKS	155,7	83.2%	83.2%	Bardet-Biedl syndrome 6, 605231 McKusick-Kaufman syndrome, 236700
NEK1	115,9	99.7%	98.1%	Short-rib thoracic dysplasia 6 with or without polydactyly, 263520 {Amyotrophic lateral sclerosis, susceptibility to, 24}, 617892
NR0B1	138,6	99.9%	99.2%	46XY sex reversal 2, dosage-sensitive, 300018 Adrenal hypoplasia, congenital, 300200
NR3C1	129,3	100.0%	99.9%	Glucocorticoid resistance, 615962
NR5A1	111	100.0%	99.7%	46, XX sex reversal 4, 617480 46XY sex reversal 3, 612965 Adrenocortical insufficiency, 612964 Premature ovarian failure 7, 612964 Spermatogenic failure 8, 613957
POR	175,5	99.2%	97.1%	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571
RIPK4	167,5	100.0%	100.0%	CHAND syndrome, 214350 Popliteal pterygium syndrome, Bartsocas-Papas type, 263650
ROR2	160,6	100.0%	99.7%	Brachydactyly, type B1, 113000 Robinow syndrome, autosomal recessive, 268310
RSPO1	103,8	100.0%	99.9%	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644
SOX3	74	97.7%	92.9%	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 Panhypopituitarism, X-linked, 312000
SOX9	159,9	100.0%	100.0%	Acampomelic campomelic dysplasia, 114290 Campomelic dysplasia, 114290 Campomelic dysplasia with autosomal sex reversal, 114290
SRCAP	153	100.0%	99.6%	Floating-Harbor syndrome, 136140

SRD5A2	85,6	100.0%	98.1%	Pseudovaginal perineoscrotal hypospadias, 264600
SRY	31,6	50.0%	50.0%	46XX sex reversal 1, 400045 46XY sex reversal 1, 400044
STAR	135	100.0%	100.0%	Lipoid adrenal hyperplasia, 201710
TCTN3	116,3	100.0%	99.9%	Joubert syndrome 18, 614815 Orofaciodigital syndrome IV, 258860
TOE1	141,1	100.0%	99.8%	Pontocerebellar hypoplasia, type 7, 614969
TSPYL1	144,9	100.0%	100.0%	Sudden infant death with dysgenesis of the testes syndrome, 608800
WDR60	108,1	99.7%	98.1%	Short-rib thoracic dysplasia 8 with or without polydactyly, 615503
WNT4	226,5	99.5%	97.3%	?SERKAL syndrome, 611812 Mullerian aplasia and hyperandrogenism, 158330
WT1	90,1	100.0%	99.3%	Denys-Drash syndrome, 194080 Frasier syndrome, 136680 Meacham syndrome, 608978 Mesothelioma, somatic, 156240 Nephrotic syndrome, type 4, 256370 Wilms tumor, type 1, 194070
ZFPM2	155,6	100.0%	99.8%	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 : May 8th, 2019.
This list is accurate for panel version DG 2.16
Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors