DSD panel

versie	12-Mar-2018 (103 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
AKR1C2	600450	Obesity, hyperphagia, and developmental delay (3); 46XY sex reversal 8, 614279 (3), Autosomal recessive
AKR1C4	600451	{46XY sex reversal 8, modifier of}, 614279 (3), Autosomal recessive
AMH	600957	Persistent Mullerian duct syndrome, type I, 261550 (3), Autosomal recessive
AMHR2	600956	Persistent Mullerian duct syndrome, type II, 261550 (3), Autosomal recessive
ANOS1	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive
AR	313700	Androgen insensitivity, 300068 (3), X-linked recessive; Androgen insensitivity, partial, with or without breast cancer, 312300 (3), X-linked recessive; Hypospadias 1, X-linked, 300633 (3), X-linked recessive; {Prostate cancer, susceptibility to}, 176807 (3), Autosomal dominant; Spinal and bulbar muscular atrophy of Kennedy, 313200 (3), X-linked recessive
ARX	300382	Epileptic encephalopathy, early infantile, 1, 308350 (3), X-linked recessive; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Lissencephaly, X-linked 2, 300215 (3), X-linked; Mental retardation, X-linked 29 and others, 300419 (3), X-linked recessive; Partington syndrome, 309510 (3), X-linked recessive; Proud syndrome, 300004 (3), X-linked
ATRX	300032	Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Alpha-thalassemia/mental retardation syndrome, 301040 (3), X-linked dominant; Mental retardation-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive
AXL	109135	No OMIM phenotype
BMP15	300247	Ovarian dysgenesis 2, 300510 (3), Autosomal dominant; Premature ovarian failure 4, 300510 (3), Autosomal dominant
BMP4	112262	Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant; Orofacial cleft 11, 600625 (3)
CBX2	602770	?46XY sex reversal 5, 613080 (3), Autosomal recessive
CHD7	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant
CREBBP	600140	Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant

CYB5A	613218	?Methemoglobinemia, type IV, 250790 (3), Autosomal recessive
CYP11A1	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3)
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive; Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant
CYP17A1	609300	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3), Autosomal recessive; 17,20-lyase deficiency, isolated, 202110 (3), Autosomal recessive
CYP19A1	107910	Aromatase deficiency, 613546 (3); Aromatase excess syndrome, 139300 (3), Autosomal dominant
CYP21A2	613815	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive; Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency, 201910 (3), Autosomal recessive
DHCR7	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DHH	605423	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 (3); 46XY sex reversal 7, 233420 (3), Autosomal recessive
DMRT1	602424	No OMIM phenotype
DMXL2	612186	?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive
DUSP6	602748	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269 (3), Autosomal dominant
EP300	602700	Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
ESR1	133430	{Atherosclerosis, susceptibility to} (3); {Breast cancer}, 114480 (1), Autosomal dominant; Estrogen resistance, 615363 (3), Autosomal recessive; {HDL response to hormone replacement, augmented} (3); {Migraine, susceptibility to}, 157300 (3), Autosomal dominant; {Myocardial infarction, susceptibility to}, 608446 (3)
ESR2	601663	No OMIM phenotype
FANCM	609644	No OMIM phenotype
FEZF1	613301	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030 (3), Autosomal recessive
FGF17	603725	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270 (3), Autosomal dominant
FGF8	600483	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant
FGF9	600921	?Multiple synostoses syndrome 3, 612961 (3), Autosomal dominant

FGFR1	136350	Encephalocraniocutaneous lipomatosis, 613001 (3), Somatic mosaicism; Hartsfield syndrome, 615465 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant
FGFR2	176943	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal recessive; Apert syndrome, 101200 (3), Autosomal dominant; Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); Crouzon syndrome, 123500 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; LADD syndrome, 149730 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Scaphocephaly, maxillary retrusion, and mental retardation, 609579 (3)
FLRT3	604808	Hypogonadotropic hypogonadism 21 with anosmia, 615271 (3), Autosomal dominant
FOXL2	605597	Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 (3), Autosomal dominant; Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 (3), Autosomal dominant; Premature ovarian failure 3, 608996 (3), Autosomal dominant
FSHB	136530	Hypogonadotropic hypogonadism 24 without anosmia, 229070 (3), Autosomal recessive
FSHR	136435	Ovarian dysgenesis 1, 233300 (3), Autosomal recessive; Ovarian hyperstimulation syndrome, 608115 (3), Autosomal dominant; Ovarian response to FSH stimulation, 276400 (3), Autosomal recessive
GATA4	600576	Atrial septal defect 2, 607941 (3), Autosomal dominant; Atrioventricular septal defect 4, 614430 (3), Autosomal dominant; ?Testicular anomalies with or without congenital heart disease, 615542 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant; Ventricular septal defect 1, 614429 (3), Autosomal dominant
GNRH1	152760	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841 (3), Autosomal recessive
GNRHR	138850	Hypogonadotropic hypogonadism 7 without anosmia, 146110 (3), Autosomal recessive
HDAC8	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant

HESX1	601802	Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant; Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septooptic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant
HHAT	605743	No OMIM phenotype
HOXA13	142959	Guttmacher syndrome, 176305 (3), Autosomal dominant; Hand-foot-uterus syndrome, 140000 (3), Autosomal dominant
HS6ST1	604846	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880 (3), Autosomal dominant
HSD17B3	605573	Pseudohermaphroditism, male, with gynecomastia, 264300 (3), Autosomal recessive
HSD17B4	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810 (3), Autosomal recessive
IL17RD	606807	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267 (3), Autosomal dominant
INSL3	146738	Cryptorchidism, 219050 (3), Autosomal dominant
KISS1	603286	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842 (3), Autosomal recessive
KISS1R	604161	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 (3), Autosomal recessive; ?Precocious puberty, central, 1, 176400 (3), Autosomal dominant
LEP	164160	Obesity, morbid, due to leptin deficiency, 614962 (3), Autosomal recessive
LEPR	601007	Obesity, morbid, due to leptin receptor deficiency, 614963 (3)
LHB	152780	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300 (3), Autosomal recessive
LHCGR	152790	Leydig cell adenoma, somatic, with precocious puberty, 176410 (3); Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 (3), Autosomal recessive; Leydig cell hypoplasia with pseudohermaphroditism, 238320 (3), Autosomal recessive; Luteinizing hormone resistance, female, 238320 (3), Autosomal recessive; Precocious puberty, male, 176410 (3), Autosomal dominant
LHX1	601999	No OMIM phenotype
LHX3	600577	Pituitary hormone deficiency, combined, 3, 221750 (3), Autosomal recessive
MAMLD1	300120	Hypospadias 2, X-linked, 300758 (3), X-linked recessive
MAP3K1	600982	46XY sex reversal 6, 613762 (3), Autosomal dominant
MID1	300552	Opitz GBBB syndrome, type I, 300000 (3), X-linked recessive
MSH4	602105	No OMIM phenotype
NOBOX	610934	Premature ovarian failure 5, 611548 (3), Autosomal dominant

NR0B1	300473	Adrenal hypoplasia, congenital, 300200 (3), X-linked recessive; 46XY sex reversal 2, dosage-sensitive, 300018 (3), X-linked
NR2F2	107773	Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant
NR5A1	184757	Adrenocortical insufficiency, 612964 (3), Autosomal dominant; Premature ovarian failure 7, 612964 (3), Autosomal dominant; Spermatogenic failure 8, 613957 (3), Autosomal dominant; 46, XX sex reversal 4, 617480 (3), Autosomal dominant; 46XY sex reversal 3, 612965 (3), Autosomal dominant
NSMF	608137	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838 (3), Autosomal dominant
NUP107	607617	Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive
OTUD4	611744	No OMIM phenotype
PATL2	614661	No OMIM phenotype
PCSK1	162150	Obesity with impaired prohormone processing, 600955 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ12}, 612362 (3)
PNPLA6	603197	Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive
POLR3A	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
POR	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive; Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3)
PROK2	607002	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628 (3), Autosomal dominant
PROKR2	607123	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200 (3), Autosomal dominant
PROP1	601538	Pituitary hormone deficiency, combined, 2, 262600 (3), Autosomal recessive
PSMC3IP	608665	Ovarian dysgenesis 3, 614324 (3), Autosomal recessive
RNF216	609948	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive
RSPO1	609595	Palmoplantar hyperkeratosis and true hermaphroditism, 610644 (3), Autosomal recessive; Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal, 610644 (3), Autosomal recessive
RXFP2	606655	No OMIM phenotype
SEMA3A	603961	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897 (3), Autosomal dominant
SEMA7A	607961	[Blood group, John-Milton-Hagen system], 614745 (3)

SOX10	602229	PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant; Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant
SOX2	184429	Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant; Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant
SOX3	313430	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
SOX8	605923	No OMIM phenotype
SOX9	608160	Acampomelic campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia with autosomal sex reversal, 114290 (3), Autosomal dominant
SPRY4	607984	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266 (3), Autosomal dominant
SRD5A2	607306	Pseudovaginal perineoscrotal hypospadias, 264600 (3), Autosomal recessive
SRY	480000	46XX sex reversal 1, 400045 (3); 46XY sex reversal 1, 400044 (3)
STAR	600617	Lipoid adrenal hyperplasia, 201710 (3), Autosomal recessive
TAC3	162330	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839 (3), Autosomal recessive
TACR3	162332	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840 (3), Autosomal recessive
TSPYL1	604714	Sudden infant death with dysgenesis of the testes syndrome, 608800 (3), Autosomal recessive
WDR11	606417	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858 (3), Autosomal dominant
WNT4	603490	Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant; ?SERKAL syndrome, 611812 (3), Autosomal recessive
WT1	607102	Denys-Drash syndrome, 194080 (3), Autosomal dominant, Somatic mutation; Frasier syndrome, 136680 (3), Autosomal dominant, Somatic mutation; Meacham syndrome, 608978 (3); Mesothelioma, somatic, 156240 (3); Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant; Wilms tumor, type 1, 194070 (3), Autosomal dominant, Somatic mutation
WWOX	605131	Epileptic encephalopathy, early infantile, 28, 616211 (3), Autosomal recessive; Esophageal squamous cell carcinoma, somatic, 133239 (3); Spinocerebellar ataxia, autosomal recessive 12, 614322 (3), Autosomal recessive
ZFPM2	603693	Diaphragmatic hernia 3, 610187 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant; 46XY sex reversal 9, 616067 (3), Autosomal dominant

ZNRF3	612062	No OMIM phenotype

Gene symbols used are according to the HGNC guidelines. For some genes a previously HGNC-approved symbol is in brackets.
Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.
OMIM release used for OMIM disease identifiers and descriptions: June 06, 2017

Possible phenotype mapping keys
(1) the disorder is placed on the map based on its association with a gene, but the underlying defect is not known
(2) the disorder has been placed on the map by linkage; no mutation has been found
(3) the molecular basis for the disorder is known; a mutation has been found in the gene
(4) a contiguous gene deletion or duplication syndrome, multiple genes are deleted or duplicated causing the phenotype

Brackets, "[ ]", indicate "nondiseases," mainly genetic variations that lead to apparently abnormal laboratory test values (e.g., dysalbuminemic euthyroidal hyperthyroxinemia).
Braces, "{ }", indicate mutations that contribute to susceptibility to multifactorial disorders (e.g., diabetes, asthma) or to susceptibility to infection (e.g., malaria).
A question mark, "?", before the phenotype name indicates that the relationship between the phenotype and gene is provisional. More details about this relationship are provided in the comment field of the map and in the gene and phenotype OMIM entries.