DSD-POI-HH panel

versie	V5 (130 genen)	Centrum voor Medische Genetica Gent

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
AARS2	612035	Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive; Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive
AKR1C2	600450	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, Somatic mutation; 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), X-linked; Premature ovarian failure 4, 300510 (3), X-linked
BMP4	112262	Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant; Orofacial cleft 11, 600625 (3)
BNC1	601930	No OMIM phenotype
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
CLPP	601119	Perrault syndrome 3, 614129 (3), Autosomal recessive

CREBBP	600140	Menke-Hennekam syndrome 1, 618332 (3); Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CYB5A	613218	Methemoglobinemia and ambiguous genitalia, 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
DHX37	617362	No OMIM phenotype
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
EIF4ENIF1	607445	No OMIM phenotype
EP300	602700	Colorectal cancer, somatic, 114500 (3); Menke-Hennekam syndrome 2, 618333 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
ESR1	133430	Breast cancer, somatic, 114480 (3); Estrogen resistance, 615363 (3), Autosomal recessive; {Migraine, susceptibility to}, 157300 (3), Autosomal dominant; {Myocardial infarction, susceptibility to}, 608446 (3)
ESR2	601663	?Ovarian dysgenesis 8, 618187 (3), Autosomal dominant
FANCM	609644	?Premature ovarian failure 15, 618096 (3), Autosomal recessive; Spermatogenic failure 28, 618086 (3), Autosomal recessive
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, somatic mosaic, 613001 (3); 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 dominant; 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)
FIGLA	608697	Premature ovarian failure 6, 612310 (3), Autosomal dominant
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
GDF9	601918	?Premature ovarian failure 14, 618014 (3), Autosomal recessive
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
HFM1	615684	Premature ovarian failure 9, 615724 (3), Autosomal recessive
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 recessive, Autosomal dominant
INSL3	146738	Cryptorchidism, 219050 (3), Autosomal dominant
KHDRBS1	602489	No OMIM phenotype
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
LARS2	604544	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive; Perrault syndrome 4, 615300 (3), Autosomal recessive
LEP	164160	Obesity, morbid, due to leptin deficiency, 614962 (3), Autosomal recessive
LEPR	601007	Obesity, morbid, due to leptin receptor deficiency, 614963 (3), Autosomal recessive
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

LHX4	602146	Pituitary hormone deficiency, combined, 4, 262700 (3), Autosomal dominant
MAMLD1	300120	Hypospadias 2, X-linked, 300758 (3), X-linked recessive
MAP3K1	600982	46XY sex reversal 6, 613762 (3), Autosomal dominant
MCM8	608187	?Premature ovarian failure 10, 612885 (3), Autosomal recessive
MCM9	610098	Ovarian dysgenesis 4, 616185 (3), Autosomal recessive
MEIOB	617670	?Spermatogenic failure 22, 617706 (3), Autosomal recessive
MID1	300552	Opitz GBBB syndrome, type I, 300000 (3), X-linked recessive
MRPS22	605810	Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive; Ovarian dysgenesis 7, 618117 (3), Autosomal recessive
MSH4	602105	No OMIM phenotype
MSH5	603382	?Premature ovarian failure 13, 617442 (3), Autosomal recessive
NANOS3	608229	No OMIM phenotype
NBN	602667	Aplastic anemia, 609135 (3); Leukemia, acute lymphoblastic, 613065 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
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	Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive; ?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive
OTUD4	611744	No OMIM phenotype
PATL2	614661	Oocyte maturation defect 4, 617743 (3), Autosomal recessive
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
POF1B	300603	?Premature ovarian failure 2B, 300604 (3), X-linked recessive
POLR2C	180663	No OMIM phenotype
POLR3A	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive; Wiedemann-Rautenstrauch syndrome, 264090 (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)
SOHLH1	610224	Ovarian dysgenesis 5, 617690 (3), Autosomal recessive; Spermatogenic failure 32, 618115 (3), Autosomal dominant
SOHLH2	616066	No OMIM phenotype
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
SPIDR	615384	No OMIM phenotype
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 (4), X-linked dominant; 46XY sex reversal 1, 400044 (3), Y-linked

STAG3	608489	Premature ovarian failure 8, 615723 (3), Autosomal recessive
STAR	600617	Lipoid adrenal hyperplasia, 201710 (3), Autosomal recessive
SYCE1	611486	?Premature ovarian failure 12, 616947 (3), Autosomal recessive; ?Spermatogenic failure 15, 616950 (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
TP63	603273	ADULT syndrome, 103285 (3), Autosomal dominant; Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Limb-mammary syndrome, 603543 (3), Autosomal dominant; Orofacial cleft 8, 618149 (3); Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; Split-hand/foot malformation 4, 605289 (3), Autosomal dominant
TSPYL1	604714	Sudden infant death with dysgenesis of the testes syndrome, 608800 (3), Autosomal recessive
TWNK	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Perrault syndrome 5, 616138 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant
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: July 04, 2018

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.