

DSD-POI-HH panel (v4)

versie 29-11-2018
(122 genen)

Centrum voor Medische Genetica Gent

Github commit: 053b2b9ab0c87deb7778cc9b1eeb160c044bc9dc

Gene	OMIM gene ID	Associated phenotype, OMIM phenotype ID, phenotype mapping key and inheritance pattern
<i>AKR1C2</i>	600450	46XY sex reversal 8, 614279 (3), Autosomal recessive
<i>AKR1C4</i>	600451	{46XY sex reversal 8, modifier of}, 614279 (3), Autosomal recessive
<i>AMH</i>	600957	Persistent Mullerian duct syndrome, type I, 261550 (3), Autosomal recessive
<i>AMHR2</i>	600956	Persistent Mullerian duct syndrome, type II, 261550 (3), Autosomal recessive
<i>ANOS1</i>	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive
<i>AR</i>	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
<i>ARX</i>	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
<i>ATRX</i>	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
<i>AXL</i>	109135	No OMIM phenotype
<i>BMP15</i>	300247	Ovarian dysgenesis 2, 300510 (3), X-linked; Premature ovarian failure 4, 300510 (3), X-linked
<i>BMP4</i>	112262	Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant; Orofacial cleft 11, 600625 (3)
<i>BNC1</i>	601930	No OMIM phenotype
<i>CBX2</i>	602770	?46XY sex reversal 5, 613080 (3), Autosomal recessive
<i>CHD7</i>	608892	CHARGE syndrome, 214800 (3), Autosomal dominant; Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant
<i>CREBBP</i>	600140	Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
<i>CYB5A</i>	613218	Methemoglobinemia and ambiguous genitalia, 250790 (3), Autosomal recessive

<i>CYP11A1</i>	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3)
<i>CYP11B1</i>	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive; Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant
<i>CYP17A1</i>	609300	17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3), Autosomal recessive; 17,20-lyase deficiency, isolated, 202110 (3), Autosomal recessive
<i>CYP19A1</i>	107910	Aromatase deficiency, 613546 (3); Aromatase excess syndrome, 139300 (3), Autosomal dominant
<i>CYP21A2</i>	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
<i>DHCR7</i>	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
<i>DHH</i>	605423	46XY partial gonadal dysgenesis, with minifascicular neuropathy, 607080 (3); 46XY sex reversal 7, 233420 (3), Autosomal recessive
<i>DMRT1</i>	602424	No OMIM phenotype
<i>DMXL2</i>	612186	?Deafness, autosomal dominant 71, 617605 (3), Autosomal dominant; ?Polyendocrine-polyneuropathy syndrome, 616113 (3), Autosomal recessive
<i>DUSP6</i>	602748	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269 (3), Autosomal dominant
<i>EIF4ENIF1</i>	607445	No OMIM phenotype
<i>EP300</i>	602700	Colorectal cancer, somatic, 114500 (3); Rubinstein-Taybi syndrome 2, 613684 (3), Autosomal dominant
<i>ESR1</i>	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)
<i>ESR2</i>	601663	No OMIM phenotype
<i>FANCM</i>	609644	No OMIM phenotype
<i>FEZF1</i>	613301	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030 (3), Autosomal recessive
<i>FGF17</i>	603725	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270 (3), Autosomal dominant
<i>FGF8</i>	600483	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant
<i>FGF9</i>	600921	Multiple synostoses syndrome 3, 612961 (3), Autosomal dominant

<i>FGFR1</i>	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; Trignocephaly 1, 190440 (3), Autosomal dominant
<i>FGFR2</i>	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)
<i>FIGLA</i>	608697	Premature ovarian failure 6, 612310 (3), Autosomal dominant
<i>FLRT3</i>	604808	Hypogonadotropic hypogonadism 21 with anosmia, 615271 (3), Autosomal dominant
<i>FOXL2</i>	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
<i>FSHB</i>	136530	Hypogonadotropic hypogonadism 24 without anosmia, 229070 (3), Autosomal recessive
<i>FSHR</i>	136435	Ovarian dysgenesis 1, 233300 (3), Autosomal recessive; Ovarian hyperstimulation syndrome, 608115 (3), Autosomal dominant; Ovarian response to FSH stimulation, 276400 (3), Autosomal recessive
<i>GATA4</i>	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
<i>GDF9</i>	601918	?Premature ovarian failure 14, 618014 (3), Autosomal recessive
<i>GNRH1</i>	152760	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841 (3), Autosomal recessive
<i>GNRHR</i>	138850	Hypogonadotropic hypogonadism 7 without anosmia, 146110 (3), Autosomal recessive
<i>HDAC8</i>	300269	Cornelia de Lange syndrome 5, 300882 (3), X-linked dominant

<i>HESX1</i>	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
<i>HFM1</i>	615684	Premature ovarian failure 9, 615724 (3), Autosomal recessive
<i>HHAT</i>	605743	No OMIM phenotype
<i>HOXA13</i>	142959	Guttmacher syndrome, 176305 (3), Autosomal dominant; Hand-foot-uterus syndrome, 140000 (3), Autosomal dominant
<i>HS6ST1</i>	604846	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880 (3), Autosomal dominant
<i>HSD17B3</i>	605573	Pseudohermaphroditism, male, with gynecomastia, 264300 (3), Autosomal recessive
<i>HSD17B4</i>	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
<i>HSD3B2</i>	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810 (3), Autosomal recessive
<i>IL17RD</i>	606807	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267 (3), Autosomal recessive, Autosomal dominant
<i>INSL3</i>	146738	Cryptorchidism, 219050 (3), Autosomal dominant
<i>KHDRBS1</i>	602489	No OMIM phenotype
<i>KISS1</i>	603286	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842 (3), Autosomal recessive
<i>KISS1R</i>	604161	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 (3), Autosomal recessive; ?Precocious puberty, central, 1, 176400 (3), Autosomal dominant
<i>LARS2</i>	604544	?Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive; Perrault syndrome 4, 615300 (3), Autosomal recessive
<i>LEP</i>	164160	Obesity, morbid, due to leptin deficiency, 614962 (3), Autosomal recessive
<i>LEPR</i>	601007	Obesity, morbid, due to leptin receptor deficiency, 614963 (3)
<i>LHB</i>	152780	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300 (3), Autosomal recessive
<i>LHCGR</i>	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
<i>LHX1</i>	601999	No OMIM phenotype
<i>LHX3</i>	600577	Pituitary hormone deficiency, combined, 3, 221750 (3), Autosomal recessive
<i>MAMLD1</i>	300120	Hypospadias 2, X-linked, 300758 (3), X-linked recessive
<i>MAP3K1</i>	600982	46XY sex reversal 6, 613762 (3), Autosomal dominant

<i>MCM8</i>	608187	?Premature ovarian failure 10, 612885 (3), Autosomal recessive
<i>MCM9</i>	610098	Ovarian dysgenesis 4, 616185 (3), Autosomal recessive
<i>MID1</i>	300552	Opitz GBBB syndrome, type I, 300000 (3), X-linked recessive
<i>MRPS22</i>	605810	Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive
<i>MSH4</i>	602105	No OMIM phenotype
<i>MSH5</i>	603382	?Premature ovarian failure 13, 617442 (3), Autosomal recessive
<i>NANOS3</i>	608229	No OMIM phenotype
<i>NOBOX</i>	610934	Premature ovarian failure 5, 611548 (3), Autosomal dominant
<i>NR0B1</i>	300473	Adrenal hypoplasia, congenital, 300200 (3), X-linked recessive; 46XY sex reversal 2, dosage-sensitive, 300018 (3), X-linked
<i>NR2F2</i>	107773	Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant
<i>NR5A1</i>	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
<i>NSMF</i>	608137	Hypogonadotropic hypogonadism 9 with or without anosmia, 614838 (3), Autosomal dominant
<i>NUP107</i>	607617	Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive
<i>OTUD4</i>	611744	No OMIM phenotype
<i>PATL2</i>	614661	Oocyte maturation defect 4, 617743 (3), Autosomal recessive
<i>PCSK1</i>	162150	Obesity with impaired prohormone processing, 600955 (3), Autosomal recessive; {Obesity, susceptibility to, BMIQ12}, 612362 (3)
<i>PNPLA6</i>	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
<i>POF1B</i>	300603	?Premature ovarian failure 2B, 300604 (3), X-linked recessive
<i>POLR2C</i>	180663	No OMIM phenotype
<i>POLR3A</i>	614258	Leukodystrophy, hypomyelinating, 7, with or without oligodontia and/or hypogonadotropic hypogonadism, 607694 (3), Autosomal recessive
<i>POR</i>	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive; Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3)
<i>PROK2</i>	607002	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628 (3), Autosomal dominant
<i>PROKR2</i>	607123	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200 (3), Autosomal dominant
<i>PROP1</i>	601538	Pituitary hormone deficiency, combined, 2, 262600 (3), Autosomal recessive
<i>PSMC3IP</i>	608665	Ovarian dysgenesis 3, 614324 (3), Autosomal recessive

<i>RNF216</i>	609948	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive
<i>RSPO1</i>	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
<i>RXFP2</i>	606655	No OMIM phenotype
<i>SEMA3A</i>	603961	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897 (3), Autosomal dominant
<i>SEMA7A</i>	607961	[Blood group, John-Milton-Hagen system], 614745 (3)
<i>SOHLH1</i>	610224	Ovarian dysgenesis 5, 617690 (3), Autosomal recessive
<i>SOHLH2</i>	616066	No OMIM phenotype
<i>SOX10</i>	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
<i>SOX2</i>	184429	Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant; Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant
<i>SOX3</i>	313430	Mental retardation, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
<i>SOX8</i>	605923	No OMIM phenotype
<i>SOX9</i>	608160	Acampomelic campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia with autosomal sex reversal, 114290 (3), Autosomal dominant
<i>SPIDR</i>	615384	No OMIM phenotype
<i>SPRY4</i>	607984	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266 (3), Autosomal dominant
<i>SRD5A2</i>	607306	Pseudovaginal perineoscrotal hypospadias, 264600 (3), Autosomal recessive
<i>SRY</i>	480000	46XX sex reversal 1, 400045 (3); 46XY sex reversal 1, 400044 (3)
<i>STAG3</i>	608489	Premature ovarian failure 8, 615723 (3), Autosomal recessive
<i>STAR</i>	600617	Lipoid adrenal hyperplasia, 201710 (3), Autosomal recessive
<i>SYCE1</i>	611486	?Premature ovarian failure 12, 616947 (3), Autosomal recessive; ?Spermatogenic failure 15, 616950 (3), Autosomal recessive
<i>TAC3</i>	162330	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839 (3), Autosomal recessive
<i>TACR3</i>	162332	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840 (3), Autosomal recessive
<i>TSPYL1</i>	604714	Sudden infant death with dysgenesis of the testes syndrome, 608800 (3), Autosomal recessive
<i>WDR11</i>	606417	Hypogonadotropic hypogonadism 14 with or without anosmia, 614858 (3), Autosomal dominant

<i>WNT4</i>	603490	Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant; ?SERKAL syndrome, 611812 (3), Autosomal recessive
<i>WT1</i>	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
<i>WWOX</i>	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
<i>ZFPM2</i>	603693	Diaphragmatic hernia 3, 610187 (3); Tetralogy of Fallot, 187500 (3), Autosomal dominant; 46XY sex reversal 9, 616067 (3), Autosomal dominant
<i>ZNRF3</i>	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.