DSD-POI-HH-PAI

Gene panel

Gene panel information

Gene panel	**DSD-POI-HH-PAI**
Version	6
Total genes	147
Activation date	Thursday 29 februari 2024
Publisher	Center for Medical Genetics, Ghent

Genes

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
AAAS	99.88 %	605378	Achalasia-addisonianism-alacrimia syndrome, 231550 (3), Autosomal recessive
AARS2	99.98 %	612035	Leukoencephalopathy, progressive, with ovarian failure, 615889 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 8, 614096 (3), Autosomal recessive
ABCD1	99.98 %	300371	Adrenoleukodystrophy, 300100 (3), X-linked recessive; Adrenomyeloneuropathy, adult, 300100 (3), X-linked recessive
AIRE	99.95 %	607358	Autoimmune polyendocrinopathy syndrome , type I, with or without reversible metaphyseal dysplasia, 240300 (3), Autosomal recessive, Autosomal dominant
AKR1C2	91.19 %	600450	46XY sex reversal 8, 614279 (3), Autosomal recessive
AKR1C4	100 %	600451	{46XY sex reversal 8, modifier of}, 614279 (3), Autosomal recessive
AMH	100 %	600957	Persistent Mullerian duct syndrome, type I, 261550 (3), Autosomal recessive
AMHR2	99.96 %	600956	Persistent Mullerian duct syndrome, type II, 261550 (3), Autosomal recessive
ANOS1	99.96 %	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1), 308700 (3), X-linked recessive
AR	99.76 %	313700	Androgen insensitivity, partial, with or without breast cancer, 312300 (3), X-linked recessive; {Prostate cancer, susceptibility to}, 176807 (3), Somatic mutation, Autosomal dominant; Androgen insensitivity, 300068 (3), X-linked recessive; Spinal and bulbar muscular atrophy of Kennedy, 313200 (3), X-linked recessive; Hypospadias 1, X-linked, 300633 (3), X-linked recessive
ARX	95.36 %	300382	Proud syndrome, 300004 (3), X-linked; Hydranencephaly with abnormal genitalia, 300215 (3), X-linked; Partington syndrome, 309510 (3), X-linked recessive; Developmental and epileptic encephalopathy 1, 308350 (3), X-linked recessive; Lissencephaly, X-linked 2, 300215 (3), X-linked; Intellectual developmental disorder, X-linked 29, 300419 (3), X-linked recessive
ATRX	99.44 %	300032	Alpha-thalassemia/mental retardation syndrome, 301040 (3), X-linked dominant; Alpha-thalassemia myelodysplasia syndrome, somatic, 300448 (3); Intellectual disability-hypotonic facies syndrome, X-linked, 309580 (3), X-linked recessive
BMP15	99.98 %	300247	Premature ovarian failure 4, 300510 (3), X-linked; Ovarian dysgenesis 2, 300510 (3), X-linked
BMP4	100 %	112262	Orofacial cleft 11, 600625 (3); Microphthalmia, syndromic 6, 607932 (3), Autosomal dominant
BNC1	100 %	601930	?Premature ovarian failure 16, 618723 (3), Autosomal dominant
C14orf39	99.76 %	617307	Spermatogenic failure 52, 619202 (3), Autosomal recessive; ?Premature ovarian failure 18, 619203 (3), Autosomal recessive
CBX2	100 %	602770	?46XY sex reversal 5, 613080 (3), Autosomal recessive
CDKN1C	100 %	600856	IMAGE syndrome, 614732 (3), Autosomal dominant; Beckwith-Wiedemann syndrome, 130650 (3), Autosomal dominant

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CHD7	99.99 %	608892	Hypogonadotropic hypogonadism 5 with or without anosmia, 612370 (3), Autosomal dominant; CHARGE syndrome, 214800 (3), Autosomal dominant
CLPP	99.99 %	601119	Perrault syndrome 3, 614129 (3), Autosomal recessive
CREBBP	99.97 %	600140	Menke-Hennekam syndrome 1, 618332 (3), Autosomal dominant; Rubinstein-Taybi syndrome 1, 180849 (3), Autosomal dominant
CYB5A	99.98 %	613218	Methemoglobinemia and ambiguous genitalia, 250790 (3), Autosomal recessive
CYP11A1	99.99 %	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete, 613743 (3)
CYP11B1	100 %	610613	Aldosteronism, glucocorticoid-remediable, 103900 (3), Autosomal dominant; Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency, 202010 (3), Autosomal recessive
CYP11B2	100 %	124080	Hypoaldosteronism, congenital, due to CMO I deficiency, 203400 (3), Autosomal recessive; Aldosterone to renin ratio raised (3); {Low renin hypertension, susceptibility to} (3); Hypoaldosteronism, congenital, due to CMO II deficiency, 610600 (3), Autosomal recessive
CYP17A1	100 %	609300	17,20-lyase deficiency, isolated, 202110 (3), Autosomal recessive; 17-alpha-hydroxylase/17,20-lyase deficiency, 202110 (3), Autosomal recessive
CYP19A1	99.99 %	107910	Aromatase deficiency, 613546 (3); Aromatase excess syndrome, 139300 (3), Autosomal dominant
DAAM2	99.99 %	606627	Nephrotic syndrome, type 24, 619263 (3), Autosomal recessive
DCAF17	99.84 %	612515	Woodhouse-Sakati syndrome, 241080 (3), Autosomal recessive
DHCR7	99.97 %	602858	Smith-Lemli-Opitz syndrome, 270400 (3), Autosomal recessive
DHH	100 %	605423	46XY gonadal dysgenesis with minifascicular neuropathy, 607080 (3), Autosomal recessive; 46XY sex reversal 7, 233420 (3), Autosomal recessive
DHX37	99.98 %	617362	Neurodevelopmental disorder with brain anomalies and with or without vertebral or cardiac anomalies, 618731 (3), Autosomal recessive; 46XY sex reversal 11, 273250 (3), Autosomal dominant
DMRT1	99.99 %	602424	*No OMIM phenotypes*
DUSP6	99.99 %	602748	Hypogonadotropic hypogonadism 19 with or without anosmia, 615269 (3), Autosomal dominant
EIF4ENIF1	100 %	607445	*No OMIM phenotypes*
ERAL1	99.99 %	607435	Perrault syndrome 6, 617565 (3), Autosomal recessive
ESR1	100 %	133430	Breast cancer, somatic, 114480 (3); {Migraine, susceptibility to}, 157300 (3), Autosomal dominant; Estrogen resistance, 615363 (3), Autosomal recessive; {Myocardial infarction, susceptibility to}, 608446 (3)
ESR2	99.96 %	601663	?Ovarian dysgenesis 8, 618187 (3), Autosomal dominant
FANCM	99.83 %	609644	?Premature ovarian failure 15, 618096 (3), Autosomal recessive; Spermatogenic failure 28, 618086 (3), Autosomal recessive
FEZF1	100 %	613301	Hypogonadotropic hypogonadism 22, with or without anosmia, 616030 (3), Autosomal recessive
FGF17	99.81 %	603725	Hypogonadotropic hypogonadism 20 with or without anosmia, 615270 (3), Autosomal dominant
FGF8	100 %	600483	Hypogonadotropic hypogonadism 6 with or without anosmia, 612702 (3), Autosomal dominant
FGF9	100 %	600921	Multiple synostoses syndrome 3, 612961 (3), Autosomal dominant

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FGFR1	100 %	136350	Pfeiffer syndrome, 101600 (3), Autosomal dominant; Hypogonadotropic hypogonadism 2 with or without anosmia, 147950 (3), Autosomal dominant; Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Hartsfield syndrome, 615465 (3), Autosomal dominant; Trigonocephaly 1, 190440 (3), Autosomal dominant; Osteoglophonic dysplasia, 166250 (3), Autosomal dominant; Encephalocraniocutaneous lipomatosis, somatic mosaic, 613001 (3)
FGFR2	99.99 %	176943	Bent bone dysplasia syndrome, 614592 (3), Autosomal dominant; LADD syndrome 1, 149730 (3), Autosomal dominant; Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis, 207410 (3), Autosomal dominant; Scaphocephaly and Axenfeld-Rieger anomaly (3); Jackson-Weiss syndrome, 123150 (3), Autosomal dominant; Gastric cancer, somatic, 613659 (3); Craniofacial-skeletal-dermatologic dysplasia, 101600 (3), Autosomal dominant; Apert syndrome, 101200 (3), Autosomal dominant; Pfeiffer syndrome, 101600 (3), Autosomal dominant; Craniosynostosis, nonspecific (3); ?Scaphocephaly, maxillary retrusion, and impaired intellectual development, 609579 (3); Beare-Stevenson cutis gyrata syndrome, 123790 (3), Autosomal dominant; Crouzon syndrome, 123500 (3), Autosomal dominant; Saethre-Chotzen syndrome, 101400 (3), Autosomal dominant
FIGLA	99.95 %	608697	Premature ovarian failure 6, 612310 (3), Autosomal dominant
FLRT3	100 %	604808	Hypogonadotropic hypogonadism 21 with anosmia, 615271 (3), Autosomal dominant
FOXL2	99.97 %	605597	Blepharophimosis, epicanthus inversus, and ptosis, type 2, 110100 (3), Autosomal recessive, Autosomal dominant; Blepharophimosis, epicanthus inversus, and ptosis, type 1, 110100 (3), Autosomal recessive, Autosomal dominant; Premature ovarian failure 3, 608996 (3), Autosomal dominant
FSHB	100 %	136530	Hypogonadotropic hypogonadism 24 without anosmia, 229070 (3), Autosomal recessive
FSHR	99.99 %	136435	Ovarian response to FSH stimulation, 276400 (3), Autosomal recessive; Ovarian hyperstimulation syndrome, 608115 (3), Autosomal dominant; Ovarian dysgenesis 1, 233300 (3), Autosomal recessive
GATA4	99.99 %	600576	Tetralogy of Fallot, 187500 (3), Autosomal dominant; Atrial septal defect 2, 607941 (3), Autosomal dominant; Ventricular septal defect 1, 614429 (3), Autosomal dominant; Atrioventricular septal defect 4, 614430 (3), Autosomal dominant; ?Testicular anomalies with or without congenital heart disease, 615542 (3), Autosomal dominant
GDF9	99.98 %	601918	?Premature ovarian failure 14, 618014 (3), Autosomal recessive
GNRH1	99.96 %	152760	?Hypogonadotropic hypogonadism 12 with or without anosmia, 614841 (3), Autosomal recessive
GNRHR	99.94 %	138850	Hypogonadotropic hypogonadism 7 without anosmia, 146110 (3), Autosomal recessive
GREB1L	99.99 %	617782	Deafness, autosomal dominant 80, 619274 (3), Autosomal dominant; Renal hypodysplasia/aplasia 3, 617805 (3), Autosomal dominant
HARS2	99.97 %	600783	Perrault syndrome 2, 614926 (3), Autosomal recessive
HESX1	99.77 %	601802	Pituitary hormone deficiency, combined, 5, 182230 (3), Autosomal recessive, Autosomal dominant; Septooptic dysplasia, 182230 (3), Autosomal recessive, Autosomal dominant; Growth hormone deficiency with pituitary anomalies, 182230 (3), Autosomal recessive, Autosomal dominant
HFM1	89.74 %	615684	Premature ovarian failure 9, 615724 (3), Autosomal recessive
HHAT	99.93 %	605743	Nivelon-Nivelon-Mabille syndrome, 600092 (3), Autosomal recessive
HNF1B	100 %	189907	Type 2 diabetes mellitus, 125853 (3), Autosomal dominant; Renal cysts and diabetes syndrome, 137920 (3), Autosomal dominant; {Renal cell carcinoma}, 144700 (3)

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HOXA13	99.94 %	142959	Hand-foot-uterus syndrome, 140000 (3), Autosomal dominant; ?Guttmacher syndrome, 176305 (3), Autosomal dominant
HS6ST1	99.99 %	604846	{Hypogonadotropic hypogonadism 15 with or without anosmia}, 614880 (3), Autosomal dominant
HSD17B3	99.77 %	605573	Pseudohermaphroditism, male, with gynecomastia, 264300 (3), Autosomal recessive
HSD17B4	99.71 %	601860	D-bifunctional protein deficiency, 261515 (3), Autosomal recessive; Perrault syndrome 1, 233400 (3), Autosomal recessive
HSD3B2	100 %	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency, 201810 (3), Autosomal recessive
IL17RD	99.94 %	606807	Hypogonadotropic hypogonadism 18 with or without anosmia, 615267 (3), Autosomal recessive, Digenic dominant, Autosomal dominant
INSL3	98.02 %	146738	Cryptorchidism, 219050 (3), Autosomal dominant
KHDRBS1	99.35 %	602489	*No OMIM phenotypes*
KISS1	99.97 %	603286	?Hypogonadotropic hypogonadism 13 with or without anosmia, 614842 (3), Autosomal recessive
KISS1R	100 %	604161	Hypogonadotropic hypogonadism 8 with or without anosmia, 614837 (3), Autosomal recessive; ?Precocious puberty, central, 1, 176400 (3), Autosomal dominant
LARS2	99.96 %	604544	Perrault syndrome 4, 615300 (3), Autosomal recessive; Hydrops, lactic acidosis, and sideroblastic anemia, 617021 (3), Autosomal recessive
LEP	100 %	164160	Obesity, morbid, due to leptin deficiency, 614962 (3), Autosomal recessive
LEPR	92.52 %	601007	Obesity, morbid, due to leptin receptor deficiency, 614963 (3), Autosomal recessive
LHB	99.93 %	152780	Hypogonadotropic hypogonadism 23 with or without anosmia, 228300 (3), Autosomal recessive
LHCGR	99.94 %	152790	Leydig cell adenoma, somatic, with precocious puberty, 176410 (3); Leydig cell hypoplasia with pseudohermaphroditism, 238320 (3), Autosomal recessive; Leydig cell hypoplasia with hypergonadotropic hypogonadism, 238320 (3), Autosomal recessive; Luteinizing hormone resistance, female, 238320 (3), Autosomal recessive; Precocious puberty, male, 176410 (3), Autosomal dominant
LHX1	99.96 %	601999	*No OMIM phenotypes*
LHX3	99.99 %	600577	Pituitary hormone deficiency, combined, 3, 221750 (3), Autosomal recessive
LHX4	99.99 %	602146	Pituitary hormone deficiency, combined, 4, 262700 (3), Autosomal dominant
MAMLD1	99.84 %	300120	Hypospadias 2, X-linked, 300758 (3), X-linked recessive
MAP3K1	99.93 %	600982	46XY sex reversal 6, 613762 (3), Autosomal dominant
MC2R	100 %	607397	Glucocorticoid deficiency, due to ACTH unresponsiveness, 202200 (3), Autosomal recessive
MCM8	98.78 %	608187	?Premature ovarian failure 10, 612885 (3), Autosomal recessive
MCM9	99.76 %	610098	Ovarian dysgenesis 4, 616185 (3), Autosomal recessive
MEIOB	99.98 %	617670	?Spermatogenic failure 22, 617706 (3), Autosomal recessive
MKKS	100 %	604896	McKusick-Kaufman syndrome, 236700 (3), Autosomal recessive; Bardet-Biedl syndrome 6, 605231 (3), Autosomal recessive
MKRN3	100 %	603856	Precocious puberty, central, 2, 615346 (3), Autosomal dominant
MRAP	100 %	609196	Glucocorticoid deficiency 2, 607398 (3), Autosomal recessive
MRPS22	99.87 %	605810	Ovarian dysgenesis 7, 618117 (3), Autosomal recessive; Combined oxidative phosphorylation deficiency 5, 611719 (3), Autosomal recessive
MSH4	89.29 %	602105	Premature ovarian failure 20, 619938 (3), Autosomal recessive; Spermatogenic failure 2, 108420 (3), Autosomal recessive

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MSH5	100 %	603382	?Premature ovarian failure 13, 617442 (3), Autosomal recessive; Spermatogenic failure 74, 619937 (3), Autosomal recessive
MYRF	99.98 %	608329	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization, 618113 (3), Autosomal dominant; Cardiac-urogenital syndrome, 618280 (3), Autosomal dominant
NANOS3	100 %	608229	*No OMIM phenotypes*
NBN	99.93 %	602667	Leukemia, acute lymphoblastic, 613065 (3); Aplastic anemia, 609135 (3); Nijmegen breakage syndrome, 251260 (3), Autosomal recessive
NDNF	99.89 %	616506	Hypogonadotropic hypogonadism 25 with anosmia, 618841 (3), Autosomal dominant
NNT	99.87 %	607878	Glucocorticoid deficiency 4, with or without mineralocorticoid deficiency, 614736 (3), Autosomal recessive
NOBOX	99.95 %	610934	Premature ovarian failure 5, 611548 (3), Autosomal dominant
NR0B1	99.98 %	300473	Adrenal hypoplasia, congenital, 300200 (3), X-linked recessive; 46XY sex reversal 2, dosage-sensitive, 300018 (3), X-linked
NR2F2	100 %	107773	46XX sex reversal 5, 618901 (3), Autosomal dominant; Congenital heart defects, multiple types, 4, 615779 (3), Autosomal dominant
NR3C1	99.93 %	138040	Glucocorticoid resistance, 615962 (3), Autosomal dominant
NR5A1	99.92 %	184757	46XX sex reversal 4, 617480 (3), Autosomal dominant; Premature ovarian failure 7, 612964 (3), Autosomal dominant; 46XY sex reversal 3, 612965 (3), Autosomal dominant; Adrenocortical insufficiency, 612964 (3), Autosomal dominant; Spermatogenic failure 8, 613957 (3), Autosomal dominant
NUP107	97.46 %	607617	?Ovarian dysgenesis 6, 618078 (3), Autosomal recessive; Galloway-Mowat syndrome 7, 618348 (3), Autosomal recessive; Nephrotic syndrome, type 11, 616730 (3), Autosomal recessive
PAX8	99.99 %	167415	Hypothyroidism, congenital, due to thyroid dysgenesis or hypoplasia, 218700 (3), Autosomal dominant
PBX1	99.88 %	176310	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay, 617641 (3), Autosomal dominant
PCSK1	99.99 %	162150	{Obesity, susceptibility to, BMIQ12}, 612362 (3); Endocrinopathy due to proprotein convertase 1/3 deficiency, 600955 (3), Autosomal recessive
PNPLA6	99.99 %	603197	Spastic paraplegia 39, autosomal recessive, 612020 (3), Autosomal recessive; Oliver-McFarlane syndrome, 275400 (3), Autosomal recessive; ?Laurence-Moon syndrome, 245800 (3), Autosomal recessive; Boucher-Neuhauser syndrome, 215470 (3), Autosomal recessive
POF1B	99.34 %	300603	?Premature ovarian failure 2B, 300604 (3), X-linked recessive
POLR2C	99.99 %	180663	*No OMIM phenotypes*
POMC	99.99 %	176830	{Obesity, early-onset, susceptibility to}, 601665 (3), Autosomal recessive, Multifactorial, Autosomal dominant; Obesity, adrenal insufficiency, and red hair due to POMC deficiency, 609734 (3), Autosomal recessive
POR	99.98 %	124015	Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis, 201750 (3), Autosomal recessive; Disordered steroidogenesis due to cytochrome P450 oxidoreductase, 613571 (3)
PPP1R12A	99.08 %	602021	Genitourinary and/or/brain malformation syndrome, 618820 (3), Autosomal dominant
PPP2R3C	99.79 %	615902	Spermatogenic failure 36, 618420 (3), Autosomal dominant; Myoectodermal gonadal dysgenesis syndrome, 618419 (3), Autosomal recessive
PROK2	98.65 %	607002	Hypogonadotropic hypogonadism 4 with or without anosmia, 610628 (3), Autosomal dominant
PROKR2	100 %	607123	Hypogonadotropic hypogonadism 3 with or without anosmia, 244200 (3), Autosomal dominant

Gene	% coding region covered*	OMIM gene id	OMIM Phenotypes
PROP1	99.88 %	601538	Pituitary hormone deficiency, combined, 2, 262600 (3), Autosomal recessive
PSMC3IP	99.82 %	608665	Ovarian dysgenesis 3, 614324 (3), Autosomal recessive
RNF216	99.99 %	609948	Cerebellar ataxia and hypogonadotropic hypogonadism, 212840 (3), Autosomal recessive
RSPO1	99.99 %	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	99.99 %	606655	*No OMIM phenotypes*
SAMD9	99.93 %	610456	Tumoral calcinosis, familial, normophosphatemic, 610455 (3), Autosomal recessive; Monosomy 7 myelodysplasia and leukemia syndrome 2, 619041 (3), Autosomal dominant; MIRAGE syndrome, 617053 (3), Autosomal dominant
SEMA3A	99.42 %	603961	{Hypogonadotropic hypogonadism 16 with or without anosmia}, 614897 (3), Autosomal dominant
SEMA7A	99.9 %	607961	?Cholestasis, progressive familial intrahepatic, 11, 619874 (3), Autosomal recessive; [Blood group, John-Milton-Hagen system], 614745 (3)
SGPL1	99.95 %	603729	Nephrotic syndrome, type 14, 617575 (3), Autosomal recessive
SOHLH1	100 %	610224	Ovarian dysgenesis 5, 617690 (3), Autosomal recessive; Spermatogenic failure 32, 618115 (3), Autosomal dominant
SOX10	100 %	602229	Waardenburg syndrome, type 4C, 613266 (3), Autosomal dominant; PCWH syndrome, 609136 (3), Autosomal dominant; Waardenburg syndrome, type 2E, with or without neurologic involvement, 611584 (3), Autosomal dominant
SOX2	100 %	184429	Optic nerve hypoplasia and abnormalities of the central nervous system, 206900 (3), Autosomal dominant; Microphthalmia, syndromic 3, 206900 (3), Autosomal dominant
SOX3	100 %	313430	Intellectual developmental disorder, X-linked, with isolated growth hormone deficiency, 300123 (3); Panhypopituitarism, X-linked, 312000 (3), X-linked
SOX8	99.99 %	605923	*No OMIM phenotypes*
SOX9	100 %	608160	Campomelic dysplasia with autosomal sex reversal, 114290 (3), Autosomal dominant; Acampomelic campomelic dysplasia, 114290 (3), Autosomal dominant; Campomelic dysplasia, 114290 (3), Autosomal dominant
SPIDR	99.96 %	615384	Ovarian dysgenesis 9, 619665 (3), Autosomal recessive
SPRY4	100 %	607984	Hypogonadotropic hypogonadism 17 with or without anosmia, 615266 (3), Autosomal dominant
SRD5A2	100 %	607306	Pseudovaginal perineoscrotal hypospadias, 264600 (3), Autosomal recessive
SRY	51.98 %	480000	46XY sex reversal 1, 400044 (3), Y-linked; 46XX sex reversal 1, 400045 (4), X-linked dominant
STAG3	98.99 %	608489	Spermatogenic failure 61, 619672 (3), Autosomal recessive; Premature ovarian failure 8, 615723 (3), Autosomal recessive
STAR	99.97 %	600617	Lipoid adrenal hyperplasia, 201710 (3), Autosomal recessive
SYCE1	99.99 %	611486	?Spermatogenic failure 15, 616950 (3), Autosomal recessive; ?Premature ovarian failure 12, 616947 (3), Autosomal recessive
TAC3	99.94 %	162330	Hypogonadotropic hypogonadism 10 with or without anosmia, 614839 (3), Autosomal recessive
TACR3	99.99 %	162332	Hypogonadotropic hypogonadism 11 with or without anosmia, 614840 (3), Autosomal recessive
TBX19	99.91 %	604614	Adrenocorticotropic hormone deficiency, 201400 (3), Autosomal recessive
TCF12	99.97 %	600480	Craniosynostosis 3, 615314 (3), Autosomal dominant; Hypogonadotropic hypogonadism 26 with or without anosmia, 619718 (3), Autosomal recessive, Autosomal dominant

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TP63	99.97 %	603273	Premature ovarian failure 21, 620311 (3), Autosomal dominant; Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3, 604292 (3), Autosomal dominant; Hay-Wells syndrome, 106260 (3), Autosomal dominant; Split-hand/foot malformation 4, 605289 (3), Autosomal dominant; Orofacial cleft 8, 618149 (3); Rapp-Hodgkin syndrome, 129400 (3), Autosomal dominant; ADULT syndrome, 103285 (3), Autosomal dominant; Limb-mammary syndrome, 603543 (3), Autosomal dominant
TSPYL1	100 %	604714	Sudden infant death with dysgenesis of the testes syndrome, 608800 (3), Autosomal recessive
TWNK	100 %	606075	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type), 271245 (3), Autosomal recessive; Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3, 609286 (3), Autosomal dominant; Perrault syndrome 5, 616138 (3), Autosomal recessive
WDR11	99.91 %	606417	Intellectual developmental disorder, autosomal recessive 78, 620237 (3), Autosomal recessive; Hypogonadotropic hypogonadism 14 with or without anosmia, 614858 (3), Autosomal dominant
WNT4	99.94 %	603490	?SERKAL syndrome, 611812 (3), Autosomal recessive; Mullerian aplasia and hyperandrogenism, 158330 (3), Autosomal dominant
WT1	99.99 %	607102	Mesothelioma, somatic, 156240 (3); Meacham syndrome, 608978 (3), Autosomal dominant; Frasier syndrome, 136680 (3), Somatic mutation, Autosomal dominant; Nephrotic syndrome, type 4, 256370 (3), Autosomal dominant; Denys-Drash syndrome, 194080 (3), Somatic mutation, Autosomal dominant; Wilms tumor, type 1, 194070 (3), Somatic mutation, Autosomal dominant
ZFPM2	100 %	603693	Diaphragmatic hernia 3, 610187 (3); 46XY sex reversal 9, 616067 (3), Autosomal dominant; Tetralogy of Fallot, 187500 (3), Autosomal dominant
ZNRF3	99.94 %	612062	*No OMIM phenotypes*

Explanation

OMIM release used for OMIM disease identifiers and descriptions: **2023-07-31**

Gene symbols used are according to the HGNC guidelines (corresponding to Ensembl database release 105).

Each Phenotype is followed by its MIM number, phenotype mapping key and inheritance pattern.

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.

* Exome panels: >=20x, HyperCap panels: >=30x