Panel de Infertilidad

Síndromes y trastornos comunes cubiertos

- Infertilidad femenina
- Infertilidad masculina

Nº de genes:	270
Entrega:	25 días
Cobertura:	≥99% ≥20x Cobertura media con profundidad ≥ 150 x
Detalles:	Análisis CNV incluido
	Repita el análisis de expansión: *AR, FMR1*
	MLPA: Aneuploidía, región AZF

Relación de genes y enfermedades asociadas

Gene	OMIM (Gene)	Associated diseases (OMIM)	Inheritance
AKR1C4	600451	46XY sex reversal 8, modifier of	AR
AMH	600957	Persistent Mullerian duct syndrome, type I	AR
AMHR2	600956	Persistent Mullerian duct syndrome, type II	AR
ANOS1	300836	Hypogonadotropic hypogonadism 1 with or without anosmia (Kallmann syndrome 1)	XLR
AR	313700	Androgen insensitivity;Androgen insensitivity, partial, with or without breast cancer;Prostate cancer, susceptibility to;Spinal and bulbar muscular atrophy of Kennedy;Hypospadias 1, XL	XLR, AD, SM
ARL13B	608922	Joubert syndrome 8	AR
ARL6	608845	Retinitis pigmentosa 55;Bardet-Biedl syndrome 1, modifier of;Bardet-Biedl syndrome 3	AR, AR, DR

ARX	300382	Developmental and epileptic encephalopathy 1;Mental retardation, XL 29 and others;Hydranencephaly with abnormal genitalia;Partington syndrome;Lissencephaly, XL 2;Proud syndrome	XLR, XL
ATP6V0A4	605239	Distal renal tubular acidosis 3, with or without sensorineural hearing loss	AR
ATRX	300032	Mental retardation-hypotonic facies syndrome, XL;Alpha-thalassemia/mental retardation syndrome;Alpha-thalassemia myelodysplasia syndrome, somatic	XLR, XLD
AURKC	603495	Spermatogenic failure 5	AR
B3GLCT	610308	Peters-plus syndrome	AR
BBS1	209901	Bardet-Biedl syndrome 1	AR, DR
BBS10	610148	Bardet-Biedl syndrome 10	AR
BBS12	610683	Bardet-Biedl syndrome 12	AR
BBS2	606151	Bardet-Biedl syndrome 2;Retinitis pigmentosa 74	AR
BBS4	600374	Bardet-Biedl syndrome 4	AR
BBS5	603650	Bardet-Biedl syndrome 5	AR
BBS7	607590	Bardet-Biedl syndrome 7	AR
BBS9	607968	Bardet-Biedl syndrome 9	AR
BCOR	300485	Microphthalmia, syndromic 2	XLD
BMP15	300247	Premature ovarian failure 4;Ovarian dysgenesis 2	XL
BMP4	112262	Microphthalmia, syndromic 6;Orofacial cleft 11	AD
BNC2	608669	Lower urinary tract obstruction, congenital	AD
CATSPER1	606389	Spermatogenic failure 7	AR
CC2D2A	612013	Meckel syndrome 6;Joubert syndrome 9;COACH syndrome 2	AR
CCDC103	614677	Ciliary dyskinesia, primary, 17	AR
CCDC28B	610162	Bardet-Biedl syndrome 1, modifier of	AR, DR
CCDC39	613798	Ciliary dyskinesia, primary, 14	AR

CCDC40	613799	Ciliary dyskinesia, primary, 15	AR
CDKN1C	600856	IMAGE syndrome;Beckwith-Wiedemann syndrome	AD
CEP164	614848	Nephronophthisis 15	AR
CEP290	610142	Leber congenital amaurosis 10;Meckel syndrome 4;?Bardet-Biedl syndrome 14;Senior-Loken syndrome 6;Joubert syndrome 5	AR
CEP41	610523	Joubert syndrome 15	AR
CFAP298	615494	Ciliary dyskinesia, primary, 26	AR
CFAP300	618058	Ciliary dyskinesia, primary, 38	AR
CFAP418	614477	Retinitis pigmentosa 64;Cone-rod dystrophy 16;Bardet-Biedl syndrome 21	AR
CFTR	602421	Congenital bilateral absence of vas deferens;Pancreatitis, hereditary;Bronchiectasis with or without elevated sweat chloride 1, modifier of;Cystic fibrosis	AR, AD
CHD4	603277	Sifrim-Hitz-Weiss syndrome	AD
CHD7	608892	CHARGE syndrome;Hypogonadotropic hypogonadism 5 with or without anosmia	AD
CREBBP	600140	Menke-Hennekam syndrome 1;Rubinstein-Taybi syndrome 1	AD
CUL4B	300304	Mental retardation, XL, syndromic 15 (Cabezas type)	XLR
CUL7	609577	3-M syndrome 1	AR
CYB5A	613218	Methemoglobinemia and ambiguous genitalia	AR
CYP11A1	118485	Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete	
CYP11B1	610613	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency;Aldosteronism, glucocorticoid-remediable	AR, AD
CYP17A1	609300	17,20-lyase deficiency, isolated;17-alpha-hydroxylase/17,20-lyase deficiency	AR

CYP19A1	107910	Aromatase excess syndrome;Aromatase deficiency	AD
CYP21A2	613815	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency;Hyperandrogenism, nonclassic type, due to 21-hydroxylase deficiency	AR
DAZ2	400026		
DHCR24	606418	Desmosterolosis	AR
DHCR7	602858	Smith-Lemli-Opitz syndrome	AR
DHH	605423	46XY gonadal dysgenesis with minifascicular neuropathy;46XY sex reversal 7	AR
DMRT1	602424		
DNAAF1	613190	Ciliary dyskinesia, primary, 13	AR
DNAAF2	612517	Ciliary dyskinesia, primary, 10	AR
DNAAF3	614566	Ciliary dyskinesia, primary, 2	AR
DNAAF4	608706	Dyslexia, susceptibility to, 1;Ciliary dyskinesia, primary, 25	AD, AR
DNAAF5	614864	Ciliary dyskinesia, primary, 18	AR
DNAH11	603339	Ciliary dyskinesia, primary, 7, with or without situs inversus	AR
DNAH5	603335	Ciliary dyskinesia, primary, 3, with or without situs inversus	AR
DNAH9	603330	Ciliary dyskinesia, primary, 40	AR
DNAI1	604366	Ciliary dyskinesia, primary, 1, with or without situs inversus	AR
DNAI2	605483	Ciliary dyskinesia, primary, 9, with or without situs inversus	AR
DNAL1	610062	Ciliary dyskinesia, primary, 16	AR
DUSP6	602748	Hypogonadotropic hypogonadism 19 with or without anosmia	AD
DYNC2H1	603297	Short-rib thoracic dysplasia 3 with or without polydactyly	AR, DR
EFNB1	300035	Craniofrontonasal dysplasia	XLD
ENPP1	173335	Arterial calcification, generalized, of infancy, 1;Obesity, susceptibility to;Hypophosphatemic rickets, AR, 2;Cole disease;Diabetes mellitus, non-insulin-dependent, susceptibility to	AR, AD, AR, mi, AD

EPG5	615068	Vici syndrome	AR
ERAL1	607435	Perrault syndrome 6	AR
ESCO2	609353	Roberts syndrome;Juberg-Hayward syndrome;SC phocomelia syndrome	AR
EVC	604831	Ellis-van Creveld syndrome;?Weyers acrofacial dysostosis	AR, AD
EVC2	607261	Ellis-van Creveld syndrome;Weyers acrofacial dysostosis	AR, AD
FAT4	612411	Hennekam lymphangiectasia-lymphedema syndrome 2;Van Maldergem syndrome 2	AR
FBXL4	605654	Mi DNA depletion syndrome 13 (encephalomyopathic type)	AR
FEZF1	613301	Hypogonadotropic hypogonadism 22, with or without anosmia	AR
FGF10	602115	Aplasia of lacrimal and salivary glands;LADD syndrome	AD
FGF17	603725	Hypogonadotropic hypogonadism 20 with or without anosmia	AD
FGF8	600483	Hypogonadotropic hypogonadism 6 with or without anosmia	AD
FGFR1	136350	Osteoglophonic dysplasia;Trigonocephaly 1;Pfeiffer syndrome;Encephalocraniocutaneous lipomatosis, somatic mosaic;Hypogonadotropic hypogonadism 2 with or without anosmia;Jackson-Weiss syndrome;Hartsfield syndrome	AD

FGFR2	176943	Crouzon syndrome;Saethre-Chotzen syndrome;Craniofacial-skeletal-dermatologic dysplasia;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis;Gastric cancer, somatic;LADD syndrome;Beare-Stevenson cutis gyrata syndrome;Scaphocephaly, maxillary retrusion, and mental retardation;Apert syndrome;Bent bone dysplasia syndrome;Pfeiffer syndrome;Jackson-Weiss syndrome	AD
FGFR3	134934	Achondroplasia;CATSHL syndrome;Thanatophoric dysplasia, type I;Bladder cancer, somatic;Hypochondroplasia;SADDAN;Colorectal cancer, somatic;Crouzon syndrome with acanthosis nigricans;Cervical cancer, somatic;Nevus, epidermal, somatic;Thanatophoric dysplasia, type II;Spermatocytic seminoma, somatic;Muenke syndrome;LADD syndrome	AD, AD, AR
FIG4	609390	Amyotrophic lateral sclerosis 11;Yunis-Varon syndrome;Charcot-Marie-Tooth disease, type 4J;?Polymicrogyria, bilateral temporooccipital	AD, AR
FLNA	300017	Frontometaphyseal dysplasia 1;Heterotopia, periventricular, 1;Terminal osseous dysplasia;Congenital short bowel syndrome;Otopalatodigital syndrome, type II;Melnick-Needles syndrome;Cardiac valvular dysplasia, XL;Intestinal pseudoobstruction, neuronal;?FG syndrome 2;Otopalatodigital syndrome, type I	XLR, XLD, XL
FLRT3	604808	Hypogonadotropic hypogonadism 21 with anosmia	AD

FMR1	309550	Premature ovarian failure 1;Fragile X syndrome;Fragile X tremor/ataxia syndrome	XL, XLD
FOXJ1	602291	Ciliary dyskinesia, primary, 43	AD
FOXL2	605597	Blepharophimosis, epicanthus inversus, and ptosis, type 1;Blepharophimosis, epicanthus inversus, and ptosis, type 2;Premature ovarian failure 3	AD, AR, AD
FRAS1	607830	Fraser syndrome 1	AR
FREM2	608945	Fraser syndrome 2;Cryptophthalmos, unilateral or bilateral, isolated	AR
FSHB	136530	Hypogonadotropic hypogonadism 24 without anosmia	AR
FSHR	136435	Ovarian dysgenesis 1;Ovarian response to FSH stimulation;Ovarian hyperstimulation syndrome	AR, AD
GAS8	605178	Ciliary dyskinesia, primary, 33	AR
GATA4	600576	Atrioventricular septal defect 4;?Testicular anomalies with or without congenital heart disease;Ventricular septal defect 1;Tetralogy of Fallot;Atrial septal defect 2	AD
GLI3	165240	Pallister-Hall syndrome;Polydactyly, preaxial, type IV;Polydactyly, postaxial, types A1 and B;Greig cephalopolysyndactyly syndrome	AD
GNRH1	152760	?Hypogonadotropic hypogonadism 12 with or without anosmia	AR
GNRHR	138850	Hypogonadotropic hypogonadism 7 without anosmia	AR
GPC3	300037	Simpson-Golabi-Behmel syndrome, type 1;Wilms tumor, somatic	XLR
GRIP1	604597	Fraser syndrome 3	AR
HBA1	141800	Methemoglobinemia, alpha type;Heinz body anemias, alpha-;Erythrocytosis 7;Thalassemias, alpha-;Hemoglobin H disease, nondeletional	AD
HCCS	300056	Linear skin defects with multiple congenital anomalies 1	XLD

HESX1	601802	Septooptic dysplasia;Pituitary hormone deficiency, combined, 5;Growth hormone deficiency with pituitary anomalies	AD, AR
HEXA	606869	Tay-Sachs disease;[Hex A pseudodeficiency];GM2-gangliosidosis, several forms	AR
HFE	613609	Porphyria variegata, susceptibility to;Alzheimer disease, susceptibility to;Hemochromatosis;[Transferrin serum level QTL2];Porphyria cutanea tarda, susceptibility to;Microvascular complications of diabetes 7	AD, AR, AD, AR
HNF1B	189907	Renal cysts and diabetes syndrome;Diabetes mellitus, noninsulin-dependent;Renal cell carcinoma	AD
HOXA13	142959	?Guttmacher syndrome;Hand-foot-uterus syndrome	AD
HS6ST1	604846	Hypogonadotropic hypogonadism 15 with or without anosmia	AD
HSD17B3	605573	Pseudohermaphroditism, male, with gynecomastia	AR
HSD17B4	601860	Perrault syndrome 1;D-bifunctional protein deficiency	AR
HSD3B2	613890	Adrenal hyperplasia, congenital, due to 3-beta-hydroxysteroid dehydrogenase 2 deficiency	AR
HUWE1	300697	Mental retardation, XL syndromic, Turner type	XL
IFT172	607386	Retinitis pigmentosa 71;Bardet-Biedl syndrome 20;Short-rib thoracic dysplasia 10 with or without polydactyly	AR
IFT27	615870	?Bardet-Biedl syndrome 19	AR
IL17RD	606807	Hypogonadotropic hypogonadism 18 with or without anosmia	AD, AR, DD
INPP5E	613037	Mental retardation, truncal obesity, retinal dystrophy, and micropenis;Joubert syndrome 1	AR
INSL3	146738	Cryptorchidism	AD

IRF6	607199	Orofacial cleft 6;van der Woude syndrome;Popliteal pterygium syndrome 1	AD
KHDC3L	611687	Hydatidiform mole, recurrent, 2	AR
KIF7	611254	Joubert syndrome 12;?Hydrolethalus syndrome 2;?Al-Gazali-Bakalinova syndrome;Acrocallosal syndrome	AR
KISS1	603286	?Hypogonadotropic hypogonadism 13 with or without anosmia	AR
KISS1R	604161	?Precocious puberty, central, 1;Hypogonadotropic hypogonadism 8 with or without anosmia	AD, AR
LEP	164160	Obesity, morbid, due to leptin deficiency	AR
LEPR	601007	Obesity, morbid, due to leptin receptor deficiency	AR
LHB	152780	Hypogonadotropic hypogonadism 23 with or without anosmia	AR
LHCGR	152790	Leydig cell hypoplasia with pseudohermaphroditism;Leydig cell adenoma, somatic, with precocious puberty;Precocious puberty, male;Leydig cell hypoplasia with hypergonadotropic hypogonadism;Luteinizing hormone resistance, female	AR, AD
LHX3	600577	Pituitary hormone deficiency, combined, 3	AR
LHX4	602146	Pituitary hormone deficiency, combined, 4	AD
LMNA	150330	Malouf syndrome;Emery-Dreifuss muscular dystrophy 3, AR;Emery-Dreifuss muscular dystrophy 2, AD;Hutchinson-Gilford progeria;Muscular dystrophy, congenital;Restrictive dermopathy, lethal;Lipodystrophy, familial partial, type 2;Charcot-Marie-Tooth disease, type 2B1;Mandibuloacral dysplasia;Cardiomyopathy, dilated, 1A;Heart-hand syndrome, Slovenian type	AD, AR
LRRC56	618227	Ciliary dyskinesia, primary, 39	AR
LZTFL1	606568	Bardet-Biedl syndrome 17	AR
MAMLD1	300120	Hypospadias 2, XL	XLR

MAP3K1	600982	46XY sex reversal 6	AD
MCM9	610098	Ovarian dysgenesis 4	AR
MED12	300188	Opitz-Kaveggia syndrome;Lujan-Fryns syndrome;Ohdo syndrome, XL;Hardikar syndrome	XLR, XLD
MID1	300552	Opitz GBBB syndrome, type I	XLR
MKKS	604896	McKusick-Kaufman syndrome;Bardet-Biedl syndrome 6	AR
MKS1	609883	Bardet-Biedl syndrome 13;Joubert syndrome 28;Meckel syndrome 1	AR
MT-ATP6			
MT-ATP8			
MT-CO1			
MT-CO2			
MT-CO3			
MT-CYB			
MT-ND1			
MT-ND2			
MT-ND3			
MT-ND4			
MT-ND4L			
MT-ND5			
MT-ND6			
MT-RNR1			
MT-RNR2			
MT-TA			
MT-TC			
MT-TD			
MT-TE			
MT-TF			
MT-TG			
MT-TH			
MT-TI			
MT-TK			
MT-TL1			
MT-TL2			

MT-TM			
MT-TN			
MT-TP			
MT-TQ			
MT-TR			
MT-TS1			
MT-TS2			
MT-TT			
MT-TV			
MT-TW			
MT-TY			
MYO7A	276903	Deafness, AD 11;Usher syndrome, type 1B;Deafness, AR 2	AD, AR
MYRF	608329	Encephalitis/encephalopathy, mild, with reversible myelin vacuolization;Cardiac-urogenital syndrome	AD
NEK1	604588	Short-rib thoracic dysplasia 6 with or without polydactyly;Amyotrophic lateral sclerosis, susceptibility to, 24	AR, DR, AD
NEK10	618726	Ciliary dyskinesia, primary, 44	AR
NME8	607421	Ciliary dyskinesia, primary, 6	AR
NPHP1	607100	Joubert syndrome 4;Nephronophthisis 1, juvenile;Senior-Loken syndrome-1	AR
NPHP3	608002	Nephronophthisis 3;Meckel syndrome 7;Renal-hepatic-pancreatic dysplasia 1	AR
NR0B1	300473	Adrenal hypoplasia, congenital;46XY sex reversal 2, dosage-sensitive	XLR, XL
NR0B2	604630	Obesity, mild, early-onset	AD, AR, mi
NR3C1	138040	Glucocorticoid resistance	AD
NR5A1	184757	46, XX sex reversal 4;Adrenocortical insufficiency;46XY sex reversal 3;Premature ovarian failure 7;Spermatogenic failure 8	AD
NSMF	608137	Hypogonadotropic hypogonadism 9 with or without anosmia	AD
ODAD4	617095	Ciliary dyskinesia, primary, 35	AR

OFD1	300170	Joubert syndrome 10;Simpson-Golabi-Behmel syndrome, type 2;?Retinitis pigmentosa 23;Orofaciodigital syndrome I	XLR, XLD
OPHN1	300127	Mental retardation, XL, with cerebellar hypoplasia and distinctive facial appearance	XLR
PANX1	608420	Oocyte maturation defect 7	AD
PATL2	614661	Oocyte maturation defect 4	AR
PAX6	607108	?Coloboma, ocular;Aniridia;?Morning glory disc anomaly;Keratitis;Optic nerve hypoplasia;?Coloboma of optic nerve;Anterior segment dysgenesis 5, multiple subtypes;Cataract with late-onset corneal dystrophy;Foveal hypoplasia 1	AD
PCNT	605925	Microcephalic osteodysplastic primordial dwarfism, type II	AR
PCSK1	162150	Obesity with impaired prohormone processing;Obesity, susceptibility to, BMIQ12	AR
PDE4D	600129	Acrodysostosis 2, with or without hormone resistance	AD
PEX1	602136	Peroxisome biogenesis disorder 1A (Zellweger);Peroxisome biogenesis disorder 1B (NALD/IRD);Heimler syndrome 1	AR
PHF6	300414	Borjeson-Forssman-Lehmann syndrome	XLR
PITX2	601542	Anterior segment dysgenesis 4;Ring dermoid of cornea;Axenfeld-Rieger syndrome, type 1	AD
PLCZ1	608075	Spermatogenic failure 17	AR
PNPLA6	603197	Spastic paraplegia 39, AR;Boucher-Neuhauser syndrome;Oliver-McFarlane syndrome;?Laurence-Moon syndrome	AR
POLR3B	614366	Leukodystrophy, hypomyelinating, 8, with or without oligodontia and/or hypogonadotropic hypogonadism	AR

POMC	176830	Obesity, adrenal insufficiency, and red hair due to POMC deficiency;Obesity, early-onset, susceptibility to	AR, AD, AR, mi
POR	124015	Disordered steroidogenesis due to cytochrome P450 oxidoreductase;Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis	AR
POU1F1	173110	Pituitary hormone deficiency, combined, 1	AD, AR
PPARG	601487	Carotid intimal medial thickness 1;Obesity, severe;Lipodystrophy, familial partial, type 3;Diabetes, type 2;Insulin resistance, severe, digenic	AD, AR, mi, AD
PROK2	607002	Hypogonadotropic hypogonadism 4 with or without anosmia	AD
PROKR2	607123	Hypogonadotropic hypogonadism 3 with or without anosmia	AD
PROM1	604365	Cone-rod dystrophy 12;Macular dystrophy, retinal, 2;Retinitis pigmentosa 41;Stargardt disease 4	AD, AR, AD, AR
PROP1	601538	Pituitary hormone deficiency, combined, 2	AR
PRPH2	179605	Retinitis pigmentosa 7 and digenic form;Retinitis punctata albescens;Choroidal dystrophy, central areolar 2;Macular dystrophy, patterned, 1;Macular dystrophy, vitelliform, 3;Leber congenital amaurosis 18	AD, AR, DD, AD, AR, AD
PSMC3IP	608665	Ovarian dysgenesis 3	AR
PTDSS1	612792	Lenz-Majewski hyperostotic dwarfism	AD
PTPN11	176876	Leukemia, juvenile myelomonocytic, somatic;LEOPARD syndrome 1;Metachondromatosis;Noonan syndrome 1	AD
RBBP8	604124	Seckel syndrome 2;Jawad syndrome	AR
RDH5	601617	Fundus albipunctatus	AD, AR
RHO	180380	Retinitis punctata albescens;Retinitis pigmentosa 4, AD or recessive;Night blindness, congenital stationary, AD 1	AD, AR

RIPK4	605706	Popliteal pterygium syndrome, Bartsocas-Papas type;CHAND syndrome	AR
RLBP1	180090	Newfoundland rod-cone dystrophy;Fundus albipunctatus;Retinitis punctata albescens;Bothnia retinal dystrophy	AD, AR, AR
RNF216	609948	Cerebellar ataxia and hypogonadotropic hypogonadism	AR
ROR2	602337	Brachydactyly, type B1;Robinow syndrome, AR	AD, AR
RPGRIP1L	610937	Joubert syndrome 7;?COACH syndrome 3;Meckel syndrome 5	AR
RPL10	312173	Autism, susceptibility to, XL 5;Mental retardation, XL, syndromic, 35	XLR
RSPO1	609595	Palmoplantar hyperkeratosis with squamous cell carcinoma of skin and sex reversal;Palmoplantar hyperkeratosis and true hermaphroditism	AR
SALL1	602218	Townes-Brocks branchiootorenal-like syndrome;Townes-Brocks syndrome 1	AD
SAMD9	610456	Monosomy 7 myelodysplasia and leukemia syndrome 2;MIRAGE syndrome;Tumoral calcinosis, familial, normophosphatemic	AD, AR
SDCCAG8	613524	Bardet-Biedl syndrome 16;Senior-Loken syndrome 7	AR
SEMA3A	603961	Hypogonadotropic hypogonadism 16 with or without anosmia	AD
SETBP1	611060	Schinzel-Giedion midface retraction syndrome;Mental retardation, AD 29	AD
SGPL1	603729	Nephrotic syndrome, type 14	AR
SOS1	182530	Noonan syndrome 4;?Fibromatosis, gingival, 1	AD
SOX10	602229	Waardenburg syndrome, type 4C;PCWH syndrome;Waardenburg syndrome, type 2E, with or without neurologic involvement	AD
SOX2	184429	Microphthalmia, syndromic 3;Optic nerve hypoplasia and abnormalities of the central nervous system	AD

SOX3	313430	Panhypopituitarism, XL;Mental retardation, XL, with isolated growth hormone deficiency	XL
SOX9	608160	Acampomelic campomelic dysplasia;Campomelic dysplasia;Campomelic dysplasia with autosomal sex reversal	AD
SPAG1	603395	Ciliary dyskinesia, primary, 28	AR
SPECC1L	614140	Opitz GBBB syndrome, type II;?Facial clefting, oblique, 1;Hypertelorism, Teebi type	AD
SPRY4	607984	Hypogonadotropic hypogonadism 17 with or without anosmia	AD
SRY	480000	46XX sex reversal 1;46XY sex reversal 1	XLD, Y-linked
STAR	600617	Lipoid adrenal hyperplasia	AR
TAC3	162330	Hypogonadotropic hypogonadism 10 with or without anosmia	AR
TACR3	162332	Hypogonadotropic hypogonadism 11 with or without anosmia	AR
TBX15	604127	Cousin syndrome	AR
TEX11	300311	Spermatogenic failure, XL, 2	XLR
TEX15	605795	Spermatogenic failure 25	AR
TLE6	612399	Preimplantation embryonic lethality	AR
TMEM67	609884	COACH syndrome 1;?RHYNS syndrome;Meckel syndrome 3;Joubert syndrome 6;Bardet-Biedl syndrome 14, modifier of;Nephronophthisis 11	AR
TMEM70	612418	Mi complex V (ATP synthase) deficiency, nuclear type 2	AR
TOE1	613931	Pontocerebellar hypoplasia, type 7	AR
TP63	603273	Rapp-Hodgkin syndrome;Orofacial cleft 8;Limb-mammary syndrome;Split-hand/foot malformation 4;Ectrodactyly, ectodermal dysplasia, and cleft lip/palate syndrome 3;Hay-Wells syndrome;ADULT syndrome	AD
TRAF3IP1	607380	Senior-Loken syndrome 9	AR

TRAIP	605958	Seckel syndrome 9	AR
TRIM32	602290	Muscular dystrophy, limb-girdle, AR 8;?Bardet-Biedl syndrome 11	AR
TSPYL1	604714	Sudden infant death with dysgenesis of the testes syndrome	AR
TTC12	610732	Ciliary dyskinesia, primary, 45	AR
TTC21B	612014	Short-rib thoracic dysplasia 4 with or without polydactyly;Nephronophthisis 12	AR, AD, AR
TTC8	608132	Bardet-Biedl syndrome 8;?Retinitis pigmentosa 51	AR
TUBB8	616768	Oocyte maturation defect 2	AD, AR
TWIST2	607556	Ablepharon-macrostomia syndrome;Focal facial dermal dysplasia 3, Setleis type;Barber-Say syndrome	AD, AR
UBR1	605981	Johanson-Blizzard syndrome	AR
USP9Y	400005	Spermatogenic failure, Y-linked, 2	Y-linked
WDPCP	613580	Congenital heart defects, hamartomas of tongue, and polysyndactyly;?Bardet-Biedl syndrome 15	AR
WDR11	606417	Hypogonadotropic hypogonadism 14 with or without anosmia	AD
WDR35	613602	Cranioectodermal dysplasia 2;Short-rib thoracic dysplasia 7 with or without polydactyly	AR
WEE2	614084	Oocyte maturation defect 5	AR
WNT4	603490	Mullerian aplasia and hyperandrogenism;?SERKAL syndrome	AD, AR
ZMYND10	607070	Ciliary dyskinesia, primary, 22	AR
ZP1	195000	Oocyte maturation defect 1	AR
ZP2	182888	Oocyte maturation defect 6	AR
ZP3	182889	Oocyte maturation defect 3	AD
AR	313700	Androgen insensitivity;Androgen insensitivity, partial, with or without breast cancer;Prostate cancer, susceptibility to;Spinal and bulbar muscular atrophy of Kennedy;Hypospadias 1, XL	XLR, AD, SM

FMR1	309550	Premature ovarian failure 1;Fragile X syndrome;Fragile X tremor/ataxia syndrome	XL, XLD