

Gene name	OMIM	phenotype	Inheritance	Comments
ENST00000244101	219000	Fraser syndrome	AR	Fraser syndrome
ENST00000244102	201810	3-beta-hydroxysteroid dehydrogenase, type II, deficiency	AR	3-beta-hydroxysteroid dehydrogenase, type II, deficiency
ENST00000244103	233420	46XY sex reversal 7	AR	46XY sex reversal 7
ENST00000244104	140000	Hand-foot-uterus syndrome	AD	Hand-foot-uterus syndrome
ENST00000244105	201710	Lipoid adrenal hyperplasia	AR	Lipoid adrenal hyperplasia
ENST00000244106	202010	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency	AR	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
ENST00000244107	219000	Fraser syndrome	AR	Fraser syndrome
ENST00000244108	614209	Meckel syndrome 9	AR	Meckel syndrome 9
ENST00000244109	263520	Short-rib thoracic dysplasia 6 with or without polydactyly	AR	Short-rib thoracic dysplasia 6 with or without polydactyly
ENST00000244110	300833	46XX sex reversal 3	XL	46XX sex reversal 3
ENST00000244111	238320	Leydig cell hypoplasia with pseudohermaphroditism	AR	Leydig cell hypoplasia with pseudohermaphroditism
ENST00000244112	615503	Short-rib thoracic dysplasia 8 with or without polydactyly	AR	Short-rib thoracic dysplasia 8 with or without polydactyly
ENST00000244113	613080	46XY sex reversal 5	AR	46XY sex reversal 5
ENST00000244114	615962	Glucocorticoid resistance, Glucocorticoid receptor deficiency	AD	Glucocorticoid resistance, Glucocorticoid receptor deficiency
ENST00000244115	114290	Campomelic dysplasia with autosomal sex reversal	AD	Campomelic dysplasia with autosomal sex reversal
ENST00000244116	194080;130400	Denys-Drash syndrome; Fraser syndrome	AD	Denys-Drash syndrome; Fraser syndrome
ENST00000244117	300215	Hydranencephaly with abnormal genitalia	XL	Hydranencephaly with abnormal genitalia
ENST00000244118	608800	Sudden infant death with dysgenesis of the testes syndrome	AR	Sudden infant death with dysgenesis of the testes syndrome
ENST00000244119	400044;400045	46XX sex reversal 1	XL	46XX sex reversal 1
ENST00000244120	614279	46XY sex reversal 8	AR	46XY sex reversal 8
ENST00000244121	611812	SERKAL syndrome	AR	SERKAL syndrome
ENST00000244122	610644	Palmoplantar hyperkeratosis and true hermaphroditism	AR	Palmoplantar hyperkeratosis and true hermaphroditism
ENST00000244123	270400	Smith-Lemli-Opitz syndrome	AR	Smith-Lemli-Opitz syndrome
ENST00000244124	613762	46XY sex reversal 6	AD	46XY sex reversal 6
ENST00000244125	615542	Testicular anomalies with or without congenital heart disease	AD	Testicular anomalies with or without congenital heart disease
ENST00000244126	614815	Joubert syndrome 18	AR	Joubert syndrome 18
ENST00000244127	613091	Short-rib thoracic dysplasia 3 with or without polydactyly	AR	Short-rib thoracic dysplasia 3 with or without polydactyly
ENST00000244128	236700	McKusick-Kaufman syndrome	AR	McKusick-Kaufman syndrome
ENST00000244129	264300	Pseudohermaphroditism, male, with gynecomastia	AR	Pseudohermaphroditism, male, with gynecomastia
ENST00000244130	264600	Pseudovaginal perineoscrotal hypospadias	AR	Pseudovaginal perineoscrotal hypospadias
ENST00000244131	613546	Aromatase deficiency	AR	Aromatase deficiency
ENST00000244132	261550	Persistent Mullerian duct syndrome, type II	AR	Persistent Mullerian duct syndrome, type II
ENST00000244133	300068	Androgen insensitivity	XL	Androgen insensitivity
ENST00000244134	613743	Adrenal insufficiency, congenital, with 46XY sex reversal	AR	Adrenal insufficiency, congenital, with 46XY sex reversal
ENST00000244135	154230	46XY sex reversal 4	AD	46XY sex reversal 4
ENST00000244136	614464	Joubert syndrome 15	AR	Joubert syndrome 15
ENST00000244137	250790	Methemoglobinemia, type IV	AR	Methemoglobinemia, type IV
ENST00000244138	300018	46XY sex reversal 2, dosage-sensitive	XL	46XY sex reversal 2, dosage-sensitive
ENST00000244139	201750	Antley-Bixler syndrome with genital anomalies and disordered sexual development	AR	Antley-Bixler syndrome with genital anomalies and disordered sexual development
ENST00000244140	309580;309581	Alpha-thalassemia/mental retardation syndrome; Mental retardation, alpha-thalassemic	XL	Alpha-thalassemia/mental retardation syndrome; Mental retardation, alpha-thalassemic
ENST00000244141	261550	Persistent Mullerian duct syndrome, type I	AR	Persistent Mullerian duct syndrome, type I
ENST00000244142	202110	17,20-lyase deficiency, isolated; 17-alpha-hydroxylase/17-beta-HSD deficiency	AR	17,20-lyase deficiency, isolated; 17-alpha-hydroxylase/17-beta-HSD deficiency
ENST00000244143	219000	Fraser syndrome	AR	Fraser syndrome
ENST00000244144	1	46,XY disorder of sex development, 2012 Kalfa et al (geer)	XL	46,XY disorder of sex development, 2012 Kalfa et al (geer)
ENST00000244145	268310	Robinow syndrome, autosomal recessive	AR	Robinow syndrome, autosomal recessive
ENST00000244146	263650	Popliteal pterygium syndrome 2, lethal type	AR	Popliteal pterygium syndrome 2, lethal type
ENST00000244147	612965	46XY sex reversal 3	AR	46XY sex reversal 3

retardation-hypotonic facies syndrome, X-linked