refgene ID	gene name	omim disease ID	inheritance	comments
NM_000176	NR3C1	800	AD	Glucocorticoid resistance; Glucocorticoid receptor deficiency (geen OMIM disease ID)
NM_001018074	NR3C1	800	AD	Glucocorticoid resistance; Glucocorticoid receptor deficiency (geen OMIM disease ID)
NM_001018075	NR3C1	800	AD	Glucocorticoid resistance; Glucocorticoid receptor deficiency (geen OMIM disease ID)
NM_001018076	NR3C1	800	AD	Glucocorticoid resistance; Glucocorticoid receptor deficiency (geen OMIM disease ID)
NM_001018077	NR3C1	800	AD	Glucocorticoid resistance; Glucocorticoid receptor deficiency (geen OMIM disease ID)
NM_001024094	NR3C1	800	AD	Glucocorticoid resistance; Glucocorticoid receptor deficiency (geen OMIM disease ID)
NM_001020825	NR3C1	800	AD	Glucocorticoid resistance; Glucocorticoid receptor deficiency (geen OMIM disease ID)
NM_005921	MAP3K1	613762	AD	46XY sex reversal 6
NM_003309	TSPYL1	608800	AR	Sudden infant death with dysgenesis of the testes syndrome
NM_002052	GATA4	615542	AD	Testicular anomalies with or without congenital heart disease
NM_000497	CYP11B1	202010	AR	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
NM_001026213	CYP11B1	202010	AR	Adrenal hyperplasia, congenital, due to 11-beta-hydroxylase deficiency
NM_001354	AKR1C2	614279	AR	46XY sex reversal 8
NM_205845	AKR1C2	614279	AR	46XY sex reversal 8
NM_001135241	AKR1C2	614279	AR	46XY sex reversal 8
NM_025074	FRAS1	219000	AR	Fraser syndrome
NM_001166133	FRAS1	219000	AR	Fraser syndrome
NM_018718	CEP41	614464	AR	Joubert syndrome 15
NM_001257159	CEP41	614464	AR	Joubert syndrome 15
NM_001257158	CEP41	614464	AR	Joubert syndrome 15
NM_001257160	CEP41	614464	AR	Joubert syndrome 15
NM_018051	WDR60	615503	AR	Short-rib thoracic dysplasia 8 with or without polydactyly
NM_004560	ROR2	268310	AR	Robinow syndrome, autosomal recessive
NM_015631	TCTN3	614815	AR	Joubert syndrome 18
NM_001143973	TCTN3	614815	AR	Joubert syndrome 18
NM_001163817	DHCR7	270400	AR	Smith-Lemli-Opitz syndrome
NM_001360	DHCR7	270400	AR	Smith-Lemli-Opitz syndrome
NM_001080463	DYNC2H1	613091	AR	Short-rib thoracic dysplasia 3 with or without polydactyly
NM_001377	DYNC2H1	613091	AR	Short-rib thoracic dysplasia 3 with or without polydactyly
NM_021150	GRIP1	219000	AR	Fraser syndrome
NM_207361	FREM2	219000	AR	Fraser syndrome
NM_148923	CYB5A	250790	AR	Methemoglobinemia, type IV
NM_001914	CYB5A	250790	AR	Methemoglobinemia, type IV
NM_001190807	CYB5A	250790	AR	Methemoglobinemia, type IV
NM_020639	RIPK4	263650	AR	Popliteal pterygium syndrome 2, lethal type
NM_005634	SOX3	300833	XL	46XX sex reversal 3
NM_030761	WNT4	611812	AR	SERKAL syndrome
NM_001038633	RSPO1	610644	AR	Palmoplantar hyperkeratosis and true hermaphroditism
NM_001242908	RSPO1	610644	AR	Palmoplantar hyperkeratosis and true

hermaphroditism
NM_001242910 RSPO1 610644 AR Palmoplantar hyperkeratosis and true hermaphroditism
NM_001242909 RSPO1 610644 AR Palmoplantar hyperkeratosis and true hermaphroditism
NM_000198 HSD3B2 201810 AR 3-beta-hydroxysteroid dehydrogenase, type II, deficiency
NM_001166120 HSD3B2 201810 AR 3-beta-hydroxysteroid dehydrogenase, type II, deficiency
NM_000348 SRD5A2 264600 AR Pseudovaginal perineoscrotal hypospadias
NM_000233 LHCGR 238320 AR Leydig cell hypoplasia with pseudohermaphroditism
NM_000941 POR 201750 AR Antley-Bixler syndrome with genital anomalies and disordered steroidogenesis
NM_000349 STAR 201710 AR Lipoid adrenal hyperplasia
NM_021951 DMRT1 154230 AD 46XY sex reversal 4
NM_000197 HSD17B3 264300 AR Pseudohermaphroditism, male, with gynecomastia
NM_004959 NR5A1 612965 AR 46XY sex reversal 3
NM_000102 CYP17A1 202110 AR 17,20-lyase deficiency, isolated; 17-alpha-hydroxylase/17,20-lyase deficiency
NM_001198551 WT1 194080;136680 AD Denys-Drash syndrome; Frasier syndrome
NM_001198552 WT1 194080;136680 AD Denys-Drash syndrome; Frasier syndrome
NM_024424 WT1 194080;136680 AD Denys-Drash syndrome; Frasier syndrome
NM_000378 WT1 194080;136680 AD Denys-Drash syndrome; Frasier syndrome
NM_024426 WT1 194080;136680 AD Denys-Drash syndrome; Frasier syndrome
NM_021044 DHH 233420 AR 46XY sex reversal 7
NM_001164690 AMHR2 261550 AR Persistent Mullerian duct syndrome, type II
NM_001164691 AMHR2 261550 AR Persistent Mullerian duct syndrome, type II
NM_020547 AMHR2 261550 AR Persistent Mullerian duct syndrome, type II
NM_000781 CYP11A1 613743 AR Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete
NM_001099773 CYP11A1 613743 AR Adrenal insufficiency, congenital, with 46XY sex reversal, partial or complete
NM_000346 SOX9 114290 AD Campomelic dysplasia with autosomal sex reversal
NM_032647 CBX2 613080 AR 46XY sex reversal 5
NM_005189 CBX2 613080 AR 46XY sex reversal 5
NM_000479 AMH 261550 AR Persistent Mullerian duct syndrome, type I
NM_139058 ARX 300215 XL Hydranencephaly with abnormal genitalia
NM_000475 DAX1 300018 XL 46XY sex reversal 2, dosage-sensitive
NM_000044 AR 300068 XL Androgen insensitivity
NM_001011645 AR 300068 XL Androgen insensitivity
NM_000489 ATRX 301040;309580 XL Alpha-thalassemia/mental retardation syndrome; Mental retardation-hypotonic facies syndrome, X-linked
NM_138270 ATRX 301040;309580 XL Alpha-thalassemia/mental retardation syndrome; Mental retardation-hypotonic facies syndrome, X-linked

NM_005491	MAMLD1	800	XL	46,XY disorder of sex development, 2012 Kalfa et al (geen OMIM disease ID)
NM_001177466	MAMLD1	800	XL	46,XY disorder of sex development, 2012 Kalfa et al (geen OMIM disease ID)
NM_001177465	MAMLD1	800	XL	46,XY disorder of sex development, 2012 Kalfa et al (geen OMIM disease ID)
NM_003140	SRY	400045	XL	46XX sex reversal 1
NM_003140	SRY	400044	XL	46XY sex reversal 1
NM_012224	NEK1	263520	AR	Short-rib thoracic dysplasia 6 with or without polydactyly
NM_001199399	NEK1	263520	AR	Short-rib thoracic dysplasia 6 with or without polydactyly
NM_001199400	NEK1	263520	AR	Short-rib thoracic dysplasia 6 with or without polydactyly
NM_001199398	NEK1	263520	AR	Short-rib thoracic dysplasia 6 with or without polydactyly
NM_001199397	NEK1	263520	AR	Short-rib thoracic dysplasia 6 with or without polydactyly
NM_015681	B9D1	614209	AR	Meckel syndrome 9
NM_001243475	B9D1	614209	AR	Meckel syndrome 9
NM_000103	CYP19A1	613546	AR	Aromatase deficiency
NM_031226	CYP19A1	613546	AR	Aromatase deficiency
NM_018848	MKKS	236700	AR	McKusick-Kaufman syndrome
NM_170784	MKKS	236700	AR	McKusick-Kaufman syndrome
NM_000522	HOXA13	140000	AD	Hand-foot-uterus syndrome