

refgene ID	gene name	omim	disease ID	inheritance	comments
NM_000176	NR3C1	-67	AD		Glucocorticoid resistance;
	Glucocorticoid receptor deficiency	(geen OMIM disease ID)			
NM_001018074	NR3C1	-68	AD		Glucocorticoid resistance;
	Glucocorticoid receptor deficiency	(geen OMIM disease ID)			
NM_001018075	NR3C1	-69	AD		Glucocorticoid resistance;
	Glucocorticoid receptor deficiency	(geen OMIM disease ID)			
NM_001018076	NR3C1	-70	AD		Glucocorticoid resistance;
	Glucocorticoid receptor deficiency	(geen OMIM disease ID)			
NM_001018077	NR3C1	-71	AD		Glucocorticoid resistance;
	Glucocorticoid receptor deficiency	(geen OMIM disease ID)			
NM_001024094	NR3C1	-72	AD		Glucocorticoid resistance;
	Glucocorticoid receptor deficiency	(geen OMIM disease ID)			
NM_001020825	NR3C1	-73	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	RSP01	610644	AR		Palmoplantar hyperkeratosis and true hermaphroditism
NM_001242908	RSP01	610644	AR		Palmoplantar hyperkeratosis and true

hermaphroditism					
NM_001242910	RSP01	610644	AR		Palmoplantar hyperkeratosis and true
hermaphroditism					
NM_001242909	RSP01	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	-74	XL	46,XY disorder of sex development, 2012
Kalfa et al (geen OMIM disease ID)				
NM_001177466	MAMLD1	-75	XL	46,XY disorder of sex development, 2012
Kalfa et al (geen OMIM disease ID)				
NM_001177465	MAMLD1	-76	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