

0000-29ba-16fd-a78e-a10.txt

ENST00000464104, ENST00000375263, ENST00000375262 HSD17B3 264300 AR
Pseudohermaphroditism, male, with gynecomastia
ENST00000405650 SRD5A2 264600 AR Pseudovaginal perineoscrotal hypospadias
ENST00000396402, ENST00000260433, ENST00000396404, ENST00000559878, ENST00000405913,
ENST00000557858, ENST00000492852 CYP19A1 613546 AR Aromatase deficiency
ENST00000257863, ENST00000550311, ENST00000379791 AMHR2 261550 AR
Persistent Mullerian duct syndrome, type II
ENST00000374690, ENST00000513847, ENST00000504326, ENST00000396044, ENST00000396043
AR 300068 XL Androgen insensitivity
ENST00000358632, ENST00000268053, ENST00000419019, ENST00000541301, ENST00000467407
CYP11A1 613743 AR Adrenal insufficiency, congenital, with 46XY sex
reversal, partial or complete
ENST00000382276, ENST00000569227 DMRT1 154230 AD 46XY sex reversal 4
ENST00000223208, ENST00000541543, ENST00000343969, ENST00000489512, ENST00000495702
CEP41 614464 AR Joubert syndrome 15
ENST00000340533, ENST00000494131, ENST00000397914, ENST00000299438, ENST00000579064
CYB5A 250790 AR Methemoglobinemia, type IV
ENST00000378970, ENST00000378963, ENST00000453287 NROB1 300018 XL 46XY sex
reversal 2, dosage-sensitive
ENST00000461988, ENST00000419840, ENST00000394893, ENST00000475509, ENST00000545601,
ENST00000450476, ENST00000439269 POR 201750 AR Antley-Bixler syndrome
with genital anomalies and disordered steroidogenesis
ENST00000373344, ENST00000395603, ENST00000480283, ENST00000373341 ATRX
309580;301040 XL Alpha-thalassemia/mental retardation syndrome; Mental
retardation-hypotonic facies syndrome, X-linked
ENST00000221496 AMH 261550 AR Persistent Mullerian duct syndrome, type
I
ENST00000369887, ENST00000489268 CYP17A1 202110 AR 17,20-lyase deficiency,
isolated; 17-alpha-hydroxylase/17,20-lyase deficiency
ENST00000280481, ENST00000482551 FREM2 219000 AR Fraser syndrome
ENST00000468306, ENST00000370401, ENST00000432680, ENST00000262858, ENST00000426613,
ENST00000455522 MAMLD1 1 XL 46,XY disorder of sex development, 2012
Kalfa et al (geen OMIM disease ID)
ENST00000375715, ENST00000375708, ENST00000550066 ROR2 268310 AR Robinow
syndrome, autosomal recessive
ENST00000332512, ENST00000352483, ENST00000542057, ENST00000544709 RIPK4 263650
AR Popliteal pterygium syndrome 2, lethal type
ENST00000373588 NR5A1 612965 AR 46XY sex reversal 3