

Table 6. Gene null mutations that display an epididymal phenotype

Gene of Interest	Phenotype	Reference
Estrogen receptor α	Disruption of fluid reabsorption in efferent ducts. No alteration in seminal vesicles, coagulating gland, and prostate. Recent studies show dilatation and displacement of initial segment tubules, some epithelial cell types (apical, narrow and clear cells) are abnormal in some epididymal region. Sperm granulomas seen in corpus and cauda	Eddy et al., 1996; Hess et al., 1997a; Hess et al., 2000
Apolipoprotein B	Male heterozygotes for mutation show severely reduced fertility. Sperm do not fertilize eggs in vivo nor in vitro, but do fertilize eggs if zona pellucida is removed. Sperm count, motility, and survival time reduced. Apolipoprotein B transcripts are expressed in testis and epididymis	Huang et al., 1996
Mononuclear phagocytic growth factor colony-stimulating factor 1 (CSF-1)	Lower density of macrophages in testis, caput, and cauda epididymidis, seminal vesicles and vas deferens. Macrophages failed to take up normal positions lining the caput tubules	Pollard et al., 1997
Somatic and testis-specific angiotensin-converting enzyme (ACE)	Male mice have reduced fertility. Sperm show defects in transport within oviducts and in binding to zonae pellucidae. Males are fertile in mice that lack somatic ACE but retain testis specific ACE	Hagaman et al., 1998
C-ros tyrosine kinase receptor	Failure of initial segment to develop. Male mice are infertile. Minor motility defects, but sperm display various degrees of flagellar angulation which may be responsible for reduction of sperm numbers observed in reproductive tract of mated females	Sonnenberg-Riethmacher et al., 1996
Bone morphogenic protein 8A	Degeneration of epididymal epithelium	Zhao et al., 1998
β -Hexosaminidase A and B	All epididymal epithelial cell types, myoid cells and macrophages of the testis in Hex β $-/-$ mice show increase in the size and number of lysosomes. Abnormalities restricted to initial segment and intermediate zone with increase in size and number of lysosomes in narrow and halo cells	Adamali et al., 1999a,b
γ -Glutamyl transpeptidase	Hypoplasia of the epididymis, testis, and seminal vesicles	Lieberman et al., 1996
ATP-gated ion channel: P2X1 receptor	Reduction in contraction of vas deferens. Reduction in fertility due to reduction of sperm in the ejaculate	Mulryan et al., 2000
Hoxa-10/11	Anterior homeotic transformation of cauda epididymidis and ductus deferens. Bilateral cryptorchidism	Satokata et al., 1995; Benson et al., 1996; Hsieh-Li et al., 1995; Davis et al., 1995
Pax 2	Absence of kidneys, uterus, and genital tracts	Torres et al., 1995
HE6 (G-Coupled Protein Receptor 64):	Male infertility due to dysregulation of fluid reabsorption in efferent ducts leading to accumulation of sperm in efferent ducts and blockage. Phenotype similar to ERKO mice.	Davies et al., 2004

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