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Kuzmin et al., Supplemental Table S1 Mouse mutations causing male infertility.

Process	**Gene**	Testis phenotype	Female Fertility	Other pathology	Reference
PGC Depletion	*Bcl2l1*	Sertoli cell only	infertile		[1]
	Bmp4	PGC depletion		Embryonic lethal	[2]
	Bub1	PGC depletion		Embryonic lethal	[3]
	Dazl	Sertoli cell only	infertile	no	[4]
	Espl1	Sertoli cell only	infertile	no	[5]
	Kit	Sertoli cell only	infertile	Anemia, coat colour spotting	[6]
	Kitl	Sertoli cell only	infertile	Anemia, coat colour dilution	[7]
	Pik3r1	Sertoli cell only	fertile	no	[5]
	Smad1			lethal	[8]
	Smad5			lethal	[9]
	Sohlh1	Sertoli cell only	infertile	no	[10]
	Sohlh2	Sertoli cell only		no	[11]
	Tia1	Sertoli cell only	infertile	Small stature, partial embryonic lethality	[12]
Meiosis	*Aurkb*	Reduced fertility	fertile	no	[13]
	Atm	Meiotic arrest	infertile	Ataxia telangiectasia	[14]
	Bat3	azoospermia	fertile	impaired development of multiple tissues	[15]
	Bsg	Meiotic arrest	infertile	Partial lethality	[16, 17]
	Ccna2	Meiotic arrest	fertile	no	[18]
	Dazap1	Meiotic arrest	infertile	perinatal lethality, growth retardation, progressive loss of renal function	[19]
	Ddx4	Meiotic arrest	fertile	no	[20]
	Dmc1	Meiotic arrest	infertile	no	[21, 22]
	Dmrtc2	Meiotic arrest	fertile	no	[23]
	Ehmt2	Meiotic arrest	fertile	no	[24]
	H2afx	Meiotic arrest	fertile		[25]
	Hsf1	Meiotic arrest	fertile	no	[26]
	Hspa2	Meiotic arrest	fertile	no	[27]
	Mlh1	Meiotic arrest	infertile	Increased susceptibility to tumour formation	[28]
	Msh5	Meiotic arrest	infertile	no	[29]
	Msh4	Meiotic arrest	infertile	no	[30]
	Mybl1	Meiotic arrest	fertile	Small stature,	[31]

				defective breast development	
	Oprl1	Meiotic arrest			[32]
	Rad51c	Meiotic arrest	infertile	no	[33]
	Sgol2	Meiotic arrest	infertile	hypogonadism	[34]
	Siah1a	Meiotic arrest	fertile	Partially lethal	[35]
	Spo11	Meiotic arrest	infertile		[36, 37]
	Stx2	Meiotic arrest	fertile	no	[38]
	Sycp2	Meiotic arrest	reduced	no	[39]
	Sycp3	Meiotic arrest	infertile		[40, 41]
	Trip13	Meiotic arrest	infertile	no	[42]
	Ubb	Meiotic arrest	infertile	hypogonadism	[43]
	Uchl1	Meiotic arrest	infertile	no	[44]
Spermiogenesis	*Apaf1*	Azoospermia	fertile	High perinatal lethality	[45]
	Aurkc	Teratozoospermia	fertile	no	[13]
	Bax	Azoospermia	fertile	Enlarged spleen	[1]
	Bbs1	Asthenoteratozoospermia	fertile	Retinal degeneration, obesity, neuroanatomical defects, cilia defects	[46]
	Bcl2l2	Azoospermia	fertile	no	[47]
	Bmp8b	Hypospermatogenesis		no	[48]
	Brdt	OAT*	fertile	no	[49]
	Brwd1	OAT	infertile	no	[50]
	Cadm1	OAT*	fertile		[51, 52]
	Camk4	Hypospermatogenesis	fertile	no	[53]
	Cdk4	Hypospermatogenesis	infertile	diabetes	[54]
	Cib1	Azoospermaia	fertile	no	[55]
	Cpeb1	azoospermia	infertile		[56]
	Crem	Azoospermia	fertile	no	[57, 58]
	Cstf2t	OAT*	fertile	no	[59]
	Cugbp1	Azoospermia	infertile	Growth retardation	[60]
	Cyld	Azoospermia	fertile	no	[61]
	Dhh	Azoospermia	fertile	no	[62]
	Dmrt1	Azoospermia	fertile		[63]
	Egr4	Hypospermatogenesis	fertile	no	[64]
	Esr1	Hypospermatogen	infertile	no	[65]

		esis			
	Foxa3	Sertoli cell only	fertile		[66]
	Fshr	Hypospermatogenesis	infertile	Low testosterone	[67]
	Fus	Azoospermia	Reduced fertility	Some perinatal lethality	[68]
	Gba2	Globozoospermia	fertile	Glycolipid storage disease	[69]
	Gdi1	Azoospermia	infertile		[70]
	Gdnf	Hypospermatogenesis	fertile	Testicular tumours	[71]
	Gja1	Azoospermia	infertile	Impaired mammary gland development, cardiac defects, embryonic lethality	[72]
	Gpx4	Oligozoospermia	fertile	no	[73]
	Hmgb2	Azoospermia	fertile		[74]
	Hspa4l	Oligoasthenozoospermia	fertile	Hydronephrosis	[75]
	Igf1	Hypospermatogenesis	infertile	Reduced mating behaviour	[76]
	Insl3	Azoospermia	Reduced fertility	cryptorchidism	[77]
	Jund	OAT*	fertile	Small stature	[78]
	Lhcgr	azoospermia	infertile		[79, 80]
	Limk2	azoospermia	fertile		[81]
	Lipe	Azoospermia	fertile	no	[82]
	Lmtk2	Azoospermia	fertle	no	[83]
	Man2a2	azoospermia	fertile		[84]
	Mll5	azoospermia	Reduced fertility	post-natal lethality, retarded growth, abnormalities in hematopoeitic lineages	[85]
	Mtap7	Azoospermia	fertile	no	[86]
	Mtr	Hypospermatogenesis	infertile	Multiple defects	[87]
	Nek1		fertile	Polycystic kidney, facial dysmorphy	[88]
	Nhlh2	Azoospermia	fertile	Hypogonadism, obesity	[89]
	Nphp1	oligoteratozoospermia	fertile	no	[90]
	P2rx1	oligospermia	fertile		[91]
	Pms2	Teratozoospermia	fertile	Increased incidence of lymphomas,	[92]

				sarcomas	
	Ppp1cc	Azoospermia	fertile	no	[93]
	Prkar1a	OAT*	fertile	Neoplasia, cardiac and cutaneous myxomas, increased incidence of endocrine tumors.	[94]
	Prm1	Azoospermia	fertile		[95]
	Prm2	Azoospermia	fertile		[95]
	Psme4	OAT*	fertile	no	[96]
	Rara	Azoospermia		High perinatal lethality	[97]
	Rpgr	OAT	fertile	none	[98]
	Rxrb	OAT*		Pituitary enlarged	[99]
	Sbf1	Azoospermia	fertile	no	[100]
	Slc12a2	Azoospermia	fertile	Inner ear defects	[101]
	Spag16	Oligoasthenozoospermia	fertile	no	[102]
	Spem1	Asthenoteratozoospermia	fertile	no	[103]
	Spz1	oligozoospermia	fertile	no	[104]
	Strbp	oligozoospermia	fertile	Neurologic defects?	[105]
	Styx	azoospermia	fertile		[106]
	Tarbp2	OAT*	Fertile?	Small stature, some perinatal lethality	[107]
	Tbpl1	azoospermia	fertile		[108, 109]
	Tex18	Asthenoteratozoospermia	fertile	no	[110]
	Tnp1	Oligozoospermia	fertile		[111]
	Tnp2	Oligozoospermia	fertile		[112]
	Ube2b	Azoospermia	fertile	no	[113]
	Wipf3	Teratozoospermia	fertile	no	[114]
Sperm Function	*Ace*	Infertile sperm	fertile	no	[115]
	Adam2	Infertile sperm	fertile		[116, 117]
	Adam3	Infertile sperm	fertile		[116, 118]
	Agfg1	Globozoospermia	fertile		[119]
	Apob	Reduced fertilty	fertile		[120]
	B4galt1	Infertile sperm	infertile	Variable embryonic lethality	[121, 122]
	Catsper3	Asthenozoospermia	fertile	no	[123]
	Catsper4	Asthenozoospermi	fertile	no	[123]

		a			
	Clgn	Infertile sperm	fertile	no	[124]
	Csnk2a2	Globozoospermia	fertile	no	[125]
	Cyp19a1	Reduced fertility	infertile	Bone defects, internal organ hyperplasia	[126, 127]
	Dnahc1	immotile	fertile		[128]
	Fanca	Reduced fertility	Reduced fertility	Fanconi's anemia	[129]
	Fancc	Reduced fertility	Reduced fertility	Fanconi's anemia, hypogonadism	[130]
	Il1rn	Reduced fertility	fertile		[131]
	Inpp5b	Reduced motility	fertile		[132]
	Nr0b1	Reduced fertility	fertile	no (X linked)	[133]
	Nsun7	Asthenozoospermia	fertile	no	[134]
	Ovol1	Reduced fertility		Coat anomalies, kidney cysts, Bardet-Biedl syndrome in humans?	[135]
	Pcsk4	Infertile sperm	fertile	no	[136]
	Pgap1	Infertile Sperm		Otocephaly, growth retardation, perinatal lethality	[137]
	Poll	immotile	fertile	Some embryonic lethality	[138]
	Prkaca	immotile		Mostly lethal	[139]
	Prlr	Reduced fertility	infertile	no	[140]
	Ros1	immotile	fertile		[141, 142]
	Smcp	immotile	fertile		[143]
	Tal	Infertile sperm	fertile	no	[144]
	Tekt4	Asthenozoospermia	fertile	no	[145]
	Tpst2	Infertile sperm	fertile	Growth retardation	[146]
	Vdac3	immotile	fertile		[147]
	Zpbp	Globozoospermia	fertile	no	[148]
	Zpbp2	Infertile sperm	fertile	no	[148]
Endocrine	*Adra1b*	Reduced fertility	fertile	no	[149]
	Ar	Feminized genitalia	fertile	cryptorchidism	[150]
	Egr1	Reduced LH	infertile		[151]
	Hsd17b2	Sertoli cell only	fertile	Growth retardation, delayed eye opening	[107, 152, 153]

	Nr2f2	azoospermia		hypogonadism, leydig cell hypoplasia	[154]
	Vdr	Defective estrogen	infertile		[152, 155]
Genital Tract/Other	*Acvr1*	delayed/reduced	fertile	Small testes	[156]
	Adamts2	oligozoospermia	fertile	Fragile skin	[157]
	Amh	obstruction	infertile	Developmental obstruction	[158, 159]
	Amhr2	obstruction	fertile	Developmental obstruction	[160]
	Bcl6	Stem cell apoptosis	fertile	Reduced fertility	[161]
	Bmp8a	Progressive infertility	fertile		[162]
	Cenpb	hypogonadal	subfertile		[163, 164]
	Cga	hypogonadal	infertile	hypothyroidism	[165]
	Cldn11	Testis architecture	fertile		[166]
	Csf1	Reduced testosterone	subfertile		[167]
	Cux1	oligozoospermia	fertile		[168]
	Cyp11a1	azoospermia		Embryonic lethal	[169]
	Emx2	No gonads		Embryonic lethal	[170]
	Etv4	Impaired sperm release	fertile		[171]
	Fancg	hypogonadism	infertile		[172]
	Fgf9	Sex reversal		lethal	[173]
	Fmr1	macrorchidism			[174]
	Gdf7	Seminal vesicle defects	fertile		[175]
	Ggt1	hypogonadism	infertile		[176, 177]
	H3f3a	Reduced fertility	fertile		[178]
	Hnf1a	Vas deferens defects	infertile	Dwarfism, diabetes	[179]
	Hoxa10	Reduced fertility	subfertile		[180]
	Hoxa11	Malformed vas	infertile		[181]
	Inha	Secondary infertility	infertile	Gonadal tumours	[182, 183]
	Lep	hypogonadism	infertile	obesity	[184, 185]
	Lepr	hypogonadism	infertile	Obesity, diabetes	[186]
	Mark2		infertile	dwarfism	[187]
	Noc2l	hypogonadism	infertile	Obesity	[188]
	Nr5a1	Gonadal agenesis		lethal	[189]
	Pou1f1	hypogonadism	infertile	dwarfism	[190]
	Ppm1d	Testicular atrophy	Fertile		[191]

	Rad23b	Secondary		Mostly lethal, multiple defects	[192]
	Rarg	Seminal vesicle defects	fertile	tumours	[193]
	Rxfp2	cryptorchidism	fertile		[194]
	Serpine2	Seminal vesicle defects	fertile		[195]
	Serpina5	Sertoli cell destruction	fertile		[196]
	Sh2b1	subfertility	infertile		[197]
	Sp4	Mating behaviour	fertile		[198]
	Star	Feminization of genitalia		lethal	[199]
	Tert	Progressive germ cell loss	infertile		[200]
	Ube3a	Testicular hypoplasia	infertile		[201]
	Wt1	Gonadal agenesis	infertile	Lethal, kidney defects	[202]
	Wnt7a	Mullerian duct regression	infertile		[203]

REFERENCES USED IN SUPPLEMENTAL DATA

1. Rucker EB,3rd, Dierisseau P, Wagner KU, Garrett L, Wynshaw-Boris A, Flaws JA, Hennighausen L. Bcl-x and Bax regulate mouse primordial germ cell survival and apoptosis during embryogenesis. Mol Endocrinol 2000; 14:1038-1052.

2. Lawson KA, Dunn NR, Roelen BA, Zeinstra LM, Davis AM, Wright CV, Korving JP, Hogan BL. Bmp4 is required for the generation of primordial germ cells in the mouse embryo. Genes Dev 1999; 13:424-436.

3. Perera D, Tilston V, Hopwood JA, Barchi M, Boot-Handford RP, Taylor SS. Bub1 maintains centromeric cohesion by activation of the spindle checkpoint. Dev Cell 2007; 13:566-579.

4. Ruggiu M, Speed R, Taggart M, McKay SJ, Kilanowski F, Saunders P, Dorin J, Cooke HJ. The mouse Dazla gene encodes a cytoplasmic protein essential for gametogenesis. Nature 1997; 389:73-77.

5. Huang X, Andreu-Vieyra CV, York JP, Hatcher R, Lu T, Matzuk MM, Zhang P. Inhibitory phosphorylation of separase is essential for genome stability and viability of murine embryonic germ cells. PLoS Biol 2008; 6:e15.

6. Blume-Jensen P, Jiang G, Hyman R, Lee KF, O'Gorman S, Hunter T. Kit/stem cell factor receptor-induced activation of phosphatidylinositol 3'-kinase is essential for male fertility. Nat Genet 2000; 24:157-162.

7. Kissel H, Timokhina I, Hardy MP, Rothschild G, Tajima Y, Soares V, Angeles M, Whitlow SR, Manova K, Besmer P. Point mutation in kit receptor tyrosine kinase reveals essential roles for kit signaling in spermatogenesis and oogenesis without affecting other kit responses. EMBO J 2000; 19:1312-1326.

8. Tremblay KD, Dunn NR, Robertson EJ. Mouse embryos lacking Smad1 signals display defects in extra-embryonic tissues and germ cell formation. Development 2001; 128:3609-3621.

9. Chang H, Matzuk MM. Smad5 is required for mouse primordial germ cell development. Mech Dev 2001; 104:61-67.

10. Ballow D, Meistrich ML, Matzuk M, Rajkovic A. Sohlh1 is essential for spermatogonial differentiation. Dev Biol 2006; 294:161-167.

11. Hao J, Yamamoto M, Richardson TE, Chapman KM, Denard BS, Hammer RE, Zhao GQ, Hamra FK. Sohlh2 Knockout Mice Are Male-Sterile Because of Degeneration of Differentiating Type A Spermatogonia. Stem Cells 2008; 26:1587-1597.

12. Beck AR, Miller IJ, Anderson P, Streuli M. RNA-binding protein TIAR is essential for primordial germ cell development. Proc Natl Acad Sci U S A 1998; 95:2331-2336.

13. Kimmins S, Crosio C, Kotaja N, Hirayama J, Monaco L, Hoog C, van Duin M, Gossen JA, Sassone-Corsi P. Differential functions of the Aurora-B and Aurora-C kinases in mammalian spermatogenesis. Mol Endocrinol 2007; 21:726-739.

14. Xu Y, Ashley T, Brainerd EE, Bronson RT, Meyn MS, Baltimore D. Targeted disruption of ATM leads to growth retardation, chromosomal fragmentation during meiosis, immune defects, and thymic lymphoma. Genes Dev 1996; 10:2411-2422.

15. Sasaki T, Marcon E, McQuire T, Arai Y, Moens PB, Okada H. Bat3 deficiency accelerates the degradation of Hsp70-2/HspA2 during spermatogenesis. J Cell Biol 2008; 182:449-458.

16. Kuno N, Kadomatsu K, Fan QW, Hagihara M, Senda T, Mizutani S, Muramatsu T. Female sterility in mice lacking the basigin gene, which encodes a transmembrane glycoprotein belonging to the immunoglobulin superfamily. FEBS Lett 1998; 425:191-194.

17. Igakura T, Kadomatsu K, Kaname T, Muramatsu H, Fan QW, Miyauchi T, Toyama Y, Kuno N, Yuasa S, Takahashi M, Senda T, Taguchi O, Yamamura K, Arimura K, Muramatsu T. A null mutation in basigin, an immunoglobulin superfamily member, indicates its important roles in peri-implantation development and spermatogenesis. Dev Biol 1998; 194:152-165.

18. Liu D, Matzuk MM, Sung WK, Guo Q, Wang P, Wolgemuth DJ. Cyclin A1 is required for meiosis in the male mouse. Nat Genet 1998; 20:377-380.

19. Hsu LC, Chen H, Lin Y, Chu W, Lin M, Yan Y, Yen PH. DAZAP1, an hnRNP protein, is required for normal growth and spermatogenesis in mice. RNA 2008; 14:1814-1822.

20. Tanaka SS, Toyooka Y, Akasu R, Katoh-Fukui Y, Nakahara Y, Suzuki R, Yokoyama M, Noce T. The mouse homolog of Drosophila Vasa is required for the development of male germ cells. Genes Dev 2000; 14:841-853.

21. Pittman DL, Cobb J, Schimenti KJ, Wilson LA, Cooper DM, Brignull E, Handel MA, Schimenti JC. Meiotic prophase arrest with failure of chromosome synapsis in mice deficient for Dmc1, a germline-specific RecA homolog. Mol Cell 1998; 1:697-705.

22. Yoshida K, Kondoh G, Matsuda Y, Habu T, Nishimune Y, Morita T. The mouse RecA-like gene Dmc1 is required for homologous chromosome synapsis during meiosis. Mol Cell 1998; 1:707-718.

23. Kawamata M, Nishimori K. Mice deficient in Dmrt7 show infertility with spermatogenic arrest at pachytene stage. FEBS Lett 2006; 580:6442-6446.

24. Tachibana M, Nozaki M, Takeda N, Shinkai Y. Functional dynamics of H3K9 methylation during meiotic prophase progression. EMBO J 2007; 26:3346-3359.

25. Celeste A, Petersen S, Romanienko PJ, Fernandez-Capetillo O, Chen HT, Sedelnikova OA, Reina-San-Martin B, Coppola V, Meffre E, Difilippantonio MJ, Redon C, Pilch DR, Olaru A, Eckhaus M, Camerini-Otero RD, Tessarollo L, Livak F, Manova K, Bonner WM, Nussenzweig MC, Nussenzweig A. Genomic instability in mice lacking histone H2AX. Science 2002; 296:922-927.

26. Nakai A, Suzuki M, Tanabe M. Arrest of spermatogenesis in mice expressing an active heat shock transcription factor 1. EMBO J 2000; 19:1545-1554.

27. Dix DJ, Allen JW, Collins BW, Poorman-Allen P, Mori C, Blizard DR, Brown PR, Goulding EH, Strong BD, Eddy EM. HSP70-2 is required for desynapsis of synaptonemal complexes during meiotic prophase in juvenile and adult mouse spermatocytes. Development 1997; 124:4595-4603.

28. Edelmann W, Cohen PE, Kane M, Lau K, Morrow B, Bennett S, Umar A, Kunkel T, Cattoretti G, Chaganti R, Pollard JW, Kolodner RD, Kucherlapati R. Meiotic pachytene arrest in MLH1-deficient mice. Cell 1996; 85:1125-1134.

29. Edelmann W, Cohen PE, Kneitz B, Winand N, Lia M, Heyer J, Kolodner R, Pollard JW, Kucherlapati R. Mammalian MutS homologue 5 is required for chromosome pairing in meiosis. Nat Genet 1999; 21:123-127.

30. Kneitz B, Cohen PE, Avdievich E, Zhu L, Kane MF, Hou H,Jr, Kolodner RD, Kucherlapati R, Pollard JW, Edelmann W. MutS homolog 4 localization to meiotic chromosomes is required for chromosome pairing during meiosis in male and female mice. Genes Dev 2000; 14:1085-1097.

31. Toscani A, Mettus RV, Coupland R, Simpkins H, Litvin J, Orth J, Hatton KS, Reddy EP. Arrest of spermatogenesis and defective breast development in mice lacking A-myb. Nature 1997; 386:713-717.

32. Watson ML, Zinn AR, Inoue N, Hess KD, Cobb J, Handel MA, Halaban R, Duchene CC, Albright GM, Moreadith RW. Identification of morc (microrchidia), a mutation that results in arrest of spermatogenesis at an early meiotic stage in the mouse. Proc Natl Acad Sci U S A 1998; 95:14361-14366.

33. Kuznetsov S, Pellegrini M, Shuda K, Fernandez-Capetillo O, Liu Y, Martin BK, Burkett S, Southon E, Pati D, Tessarollo L, West SC, Donovan PJ, Nussenzweig A, Sharan SK. RAD51C deficiency in mice results in early prophase I arrest in males and sister chromatid separation at metaphase II in females. J Cell Biol 2007; 176:581-592.

34. Llano E, Gómez R, Gutiérrez-Caballero C, Herrán Y, Sánchez-Martín M, Vázquez-Quiñones L, Hernández T, de Álava E, Cuadrado A, Barbero JL, Suja JA, Pendás AM. Shugoshin-2 is essential for the completion of meiosis but not for mitotic cell division in mice. Genes & Development 2008; 22:2400-2413.

35. Dickins RA, Frew IJ, House CM, O'Bryan MK, Holloway AJ, Haviv I, Traficante N, de Kretser DM, Bowtell DD. The ubiquitin ligase component Siah1a is required for completion of meiosis I in male mice. Mol Cell Biol 2002; 22:2294-2303.

36. Baudat F, Manova K, Yuen JP, Jasin M, Keeney S. Chromosome synapsis defects and sexually dimorphic meiotic progression in mice lacking Spo11. Mol Cell 2000; 6:989-998.

37. Romanienko PJ, Camerini-Otero RD. The mouse Spo11 gene is required for meiotic chromosome synapsis. Mol Cell 2000; 6:975-987.

38. Akiyama K, Akimaru S, Asano Y, Khalaj M, Kiyosu C, Masoudi AA, Takahashi S, Katayama K, Tsuji T, Noguchi J, Kunieda T. A new ENU-induced mutant mouse with defective spermatogenesis caused by a nonsense mutation of the syntaxin 2/epimorphin (Stx2/Epim) gene. J Reprod Dev 2008; 54:122-128.

39. Yang F, De La Fuente R, Leu NA, Baumann C, McLaughlin KJ, Wang PJ. Mouse SYCP2 is required for synaptonemal complex assembly and chromosomal synapsis during male meiosis. J Cell Biol 2006; 173:497-507.

40. Yuan L, Liu JG, Zhao J, Brundell E, Daneholt B, Hoog C. The murine SCP3 gene is required for synaptonemal complex assembly, chromosome synapsis, and male fertility. Mol Cell 2000; 5:73-83.

41. Yuan L, Liu JG, Hoja MR, Wilbertz J, Nordqvist K, Hoog C. Female germ cell aneuploidy and embryo death in mice lacking the meiosis-specific protein SCP3. Science 2002; 296:1115-1118.

42. Li XC, Schimenti JC. Mouse pachytene checkpoint 2 (trip13) is required for completing meiotic recombination but not synapsis. PLoS Genet 2007; 3:e130.

43. Ryu KY, Sinnar SA, Reinholdt LG, Vaccari S, Hall S, Garcia MA, Zaitseva TS, Bouley DM, Boekelheide K, Handel MA, Conti M, Kopito RR. The mouse polyubiquitin gene Ubb is essential for meiotic progression. Mol Cell Biol 2008; 28:1136-1146.

44. Wang YL, Liu W, Sun YJ, Kwon J, Setsuie R, Osaka H, Noda M, Aoki S, Yoshikawa Y, Wada K. Overexpression of ubiquitin carboxyl-terminal hydrolase L1 arrests spermatogenesis in transgenic mice. Mol Reprod Dev 2006; 73:40-49.

45. Honarpour N, Du C, Richardson JA, Hammer RE, Wang X, Herz J. Adult Apaf-1-deficient mice exhibit male infertility. Dev Biol 2000; 218:248-258.

46. Davis RE, Swiderski RE, Rahmouni K, Nishimura DY, Mullins RF, Agassandian K, Philp AR, Searby CC, Andrews MP, Thompson S, Berry CJ, Thedens DR, Yang B, Weiss RM, Cassell MD, Stone EM, Sheffield VC. A knockin mouse model of the Bardet-Biedl syndrome 1 M390R mutation has cilia defects, ventriculomegaly, retinopathy, and obesity. Proc Natl Acad Sci U S A 2007; 104:19422-19427.

47. Print CG, Loveland KL, Gibson L, Meehan T, Stylianou A, Wreford N, de Kretser D, Metcalf D, Kontgen F, Adams JM, Cory S. Apoptosis regulator bcl-w is essential for spermatogenesis but appears otherwise redundant. Proc Natl Acad Sci U S A 1998; 95:12424-12431.

48. Ying Y, Liu XM, Marble A, Lawson KA, Zhao GQ. Requirement of Bmp8b for the generation of primordial germ cells in the mouse. Mol Endocrinol 2000; 14:1053-1063.

49. Shang E, Nickerson HD, Wen D, Wang X, Wolgemuth DJ. The first bromodomain of Brdt, a testis-specific member of the BET sub-family of double-bromodomain-containing proteins, is essential for male germ cell differentiation. Development 2007; 134:3507-3515.

50. Philipps DL, Wigglesworth K, Hartford SA, Sun F, Pattabiraman S, Schimenti K, Handel M, Eppig JJ, Schimenti JC. The dual bromodomain and WD repeat-containing mouse protein BRWD1 is required for normal spermiogenesis and the oocyte–embryo transition. Developmental Biology 2008; 317:72-82.

51. Yamada D, Yoshida M, Williams YN, Fukami T, Kikuchi S, Masuda M, Maruyama T, Ohta T, Nakae D, Maekawa A, Kitamura T, Murakami Y. Disruption of spermatogenic cell adhesion and male infertility in mice lacking TSLC1/IGSF4, an immunoglobulin superfamily cell adhesion molecule. Mol Cell Biol 2006; 26:3610-3624.

52. Fujita E, Kouroku Y, Ozeki S, Tanabe Y, Toyama Y, Maekawa M, Kojima N, Senoo H, Toshimori K, Momoi T. Oligo-astheno-teratozoospermia in mice lacking RA175/TSLC1/SynCAM/IGSF4A, a cell adhesion molecule in the immunoglobulin superfamily. Mol Cell Biol 2006; 26:718-726.

53. Wu JY, Ribar TJ, Cummings DE, Burton KA, McKnight GS, Means AR. Spermiogenesis and exchange of basic nuclear proteins are impaired in male germ cells lacking Camk4. Nat Genet 2000; 25:448-452.

54. Rane SG, Dubus P, Mettus RV, Galbreath EJ, Boden G, Reddy EP, Barbacid M. Loss of Cdk4 expression causes insulin-deficient diabetes and Cdk4 activation results in beta-islet cell hyperplasia. Nat Genet 1999; 22:44-52.

55. Yuan W, Leisner TM, McFadden AW, Clark S, Hiller S, Maeda N, O'Brien DA, Parise LV. CIB1 is essential for mouse spermatogenesis. Mol Cell Biol 2006; 26:8507-8514.

56. Tay J, Richter JD. Germ cell differentiation and synaptonemal complex formation are disrupted in CPEB knockout mice. Dev Cell 2001; 1:201-213.

57. Blendy JA, Kaestner KH, Weinbauer GF, Nieschlag E, Schutz G. Severe impairment of spermatogenesis in mice lacking the CREM gene. Nature 1996; 380:162-165.

58. Nantel F, Monaco L, Foulkes NS, Masquilier D, LeMeur M, Henriksen K, Dierich A, Parvinen M, Sassone-Corsi P. Spermiogenesis deficiency and germ-cell apoptosis in CREM-mutant mice. Nature 1996; 380:159-162.

59. Dass B, Tardif S, Park JY, Tian B, Weitlauf HM, Hess RA, Carnes K, Griswold MD, Small CL, Macdonald CC. Loss of polyadenylation protein tauCstF-64 causes spermatogenic defects and male infertility. Proc Natl Acad Sci U S A 2007; 104:20374-20379.

60. Kress C, Gautier-Courteille C, Osborne HB, Babinet C, Paillard L. Inactivation of CUG-BP1/CELF1 causes growth, viability, and spermatogenesis defects in mice. Mol Cell Biol 2007; 27:1146-1157.

61. Wright A, Reiley WW, Chang M, Jin W, Lee AJ, Zhang M, Sun SC. Regulation of early wave of germ cell apoptosis and spermatogenesis by deubiquitinating enzyme CYLD. Dev Cell 2007; 13:705-716.

62. Bitgood MJ, Shen L, McMahon AP. Sertoli cell signaling by Desert hedgehog regulates the male germline. Curr Biol 1996; 6:298-304.

63. Raymond CS, Murphy MW, O'Sullivan MG, Bardwell VJ, Zarkower D. Dmrt1, a gene related to worm and fly sexual regulators, is required for mammalian testis differentiation. Genes Dev 2000; 14:2587-2595.

64. Tourtellotte WG, Nagarajan R, Bartke A, Milbrandt J. Functional compensation by Egr4 in Egr1-dependent luteinizing hormone regulation and Leydig cell steroidogenesis. Mol Cell Biol 2000; 20:5261-5268.

65. Eddy EM, Washburn TF, Bunch DO, Goulding EH, Gladen BC, Lubahn DB, Korach KS. Targeted disruption of the estrogen receptor gene in male mice causes alteration of spermatogenesis and infertility. Endocrinology 1996; 137:4796-4805.

66. Behr R, Sackett SD, Bochkis IM, Le PP, Kaestner KH. Impaired male fertility and atrophy of seminiferous tubules caused by haploinsufficiency for Foxa3. Dev Biol 2007; 306:636-645.

67. Krishnamurthy H, Danilovich N, Morales CR, Sairam MR. Qualitative and quantitative decline in spermatogenesis of the follicle-stimulating hormone receptor knockout (FORKO) mouse. Biol Reprod 2000; 62:1146-1159.

68. Kuroda M, Sok J, Webb L, Baechtold H, Urano F, Yin Y, Chung P, de Rooij DG, Akhmedov A, Ashley T, Ron D. Male sterility and enhanced radiation sensitivity in TLS(-/-) mice. EMBO J 2000; 19:453-462.

69. Yildiz Y, Matern H, Thompson B, Allegood JC, Warren RL, Ramirez DM, Hammer RE, Hamra FK, Matern S, Russell DW. Mutation of beta-glucosidase 2 causes glycolipid storage disease and impaired male fertility. J Clin Invest 2006; 116:2985-2994.

70. Togawa A, Miyoshi J, Ishizaki H, Tanaka M, Takakura A, Nishioka H, Yoshida H, Doi T, Mizoguchi A, Matsuura N, Niho Y, Nishimune Y, Nishikawa S, Takai Y. Progressive impairment of kidneys and reproductive organs in mice lacking Rho GDIalpha. Oncogene 1999; 18:5373-5380.

71. Meng X, Lindahl M, Hyvonen ME, Parvinen M, de Rooij DG, Hess MW, Raatikainen-Ahokas A, Sainio K, Rauvala H, Lakso M, Pichel JG, Westphal H, Saarma M, Sariola H. Regulation of cell fate decision of undifferentiated spermatogonia by GDNF. Science 2000; 287:1489-1493.

72. Juneja SC, Barr KJ, Enders GC, Kidder GM. Defects in the germ line and gonads of mice lacking connexin43. Biol Reprod 1999; 60:1263-1270.

73. Puglisi R, Bevilacqua A, Carlomagno G, Lenzi A, Gandini L, Stefanini M, Mangia F, Boitani C. Mice overexpressing the mitochondrial phospholipid hydroperoxide glutathione peroxidase in male germ cells show abnormal spermatogenesis and reduced fertility. Endocrinology 2007; 148:4302-4309.

74. Ronfani L, Ferraguti M, Croci L, Ovitt CE, Scholer HR, Consalez GG, Bianchi ME. Reduced fertility and spermatogenesis defects in mice lacking chromosomal protein Hmgb2. Development 2001; 128:1265-1273.

75. Held T, Paprotta I, Khulan J, Hemmerlein B, Binder L, Wolf S, Schubert S, Meinhardt A, Engel W, Adham IM. Hspa4l-deficient mice display increased incidence of male infertility and hydronephrosis development. Mol Cell Biol 2006; 26:8099-8108.

76. Baker SM, Plug AW, Prolla TA, Bronner CE, Harris AC, Yao X, Christie DM, Monell C, Arnheim N, Bradley A, Ashley T, Liskay RM. Involvement of mouse Mlh1 in DNA mismatch repair and meiotic crossing over. Nat Genet 1996; 13:336-342.

77. Zimmermann S, Steding G, Emmen JM, Brinkmann AO, Nayernia K, Holstein AF, Engel W, Adham IM. Targeted disruption of the Insl3 gene causes bilateral cryptorchidism. Mol Endocrinol 1999; 13:681-691.

78. Thepot D, Weitzman JB, Barra J, Segretain D, Stinnakre MG, Babinet C, Yaniv M. Targeted disruption of the murine junD gene results in multiple defects in male reproductive function. Development 2000; 127:143-153.

79. Zhang FP, Poutanen M, Wilbertz J, Huhtaniemi I. Normal prenatal but arrested postnatal sexual development of luteinizing hormone receptor knockout (LuRKO) mice. Mol Endocrinol 2001; 15:172-183.

80. Lei ZM, Mishra S, Zou W, Xu B, Foltz M, Li X, Rao CV. Targeted disruption of luteinizing hormone/human chorionic gonadotropin receptor gene. Mol Endocrinol 2001; 15:184-200.

81. Takahashi H, Koshimizu U, Miyazaki J, Nakamura T. Impaired spermatogenic ability of testicular germ cells in mice deficient in the LIM-kinase 2 gene. Dev Biol 2002; 241:259-272.

82. Chung S, Wang SP, Pan L, Mitchell G, Trasler J, Hermo L. Infertility and testicular defects in hormone-sensitive lipase-deficient mice. Endocrinology 2001; 142:4272-4281.

83. Kawa S, Ito C, Toyama Y, Maekawa M, Tezuka T, Nakamura T, Nakazawa T, Yokoyama K, Yoshida N, Toshimori K, Yamamoto T. Azoospermia in mice with targeted disruption of the Brek/Lmtk2 (brain-enriched kinase/lemur tyrosine kinase 2) gene. Proc Natl Acad Sci U S A 2006; 103:19344-19349.

84. Akama TO, Nakagawa H, Sugihara K, Narisawa S, Ohyama C, Nishimura S, O'Brien DA, Moremen KW, Millan JL, Fukuda MN. Germ cell survival through carbohydrate-mediated interaction with Sertoli cells. Science 2002; 295:124-127.

85. Madan V, Madan B, Brykczynska U, Zilbermann F, Hogeveen K, Dohner K, Dohner H, Weber O, Blum C, Rodewald H, Sassone-Corsi P, Peters AHFM, Fehling HJ. Impaired function of primitive hematopoietic cells in mice lacking the Mixed-Lineage-Leukemia homolog Mll5. Blood 2008;

86. Komada M, McLean DJ, Griswold MD, Russell LD, Soriano P. E-MAP-115, encoding a microtubule-associated protein, is a retinoic acid-inducible gene required for spermatogenesis. Genes Dev 2000; 14:1332-1342.

87. Rudolph KL, Chang S, Lee HW, Blasco M, Gottlieb GJ, Greider C, DePinho RA. Longevity, stress response, and cancer in aging telomerase-deficient mice. Cell 1999; 96:701-712.

88. Upadhya P, Birkenmeier EH, Birkenmeier CS, Barker JE. Mutations in a NIMA-related kinase gene, Nek1, cause pleiotropic effects including a progressive polycystic kidney disease in mice. Proc Natl Acad Sci U S A 2000; 97:217-221.

89. Good DJ, Porter FD, Mahon KA, Parlow AF, Westphal H, Kirsch IR. Hypogonadism and obesity in mice with a targeted deletion of the Nhlh2 gene. Nat Genet 1997; 15:397-401.

90. Jiang S, Chiou Y, Wang E, Lin H, Lee S, Lu H, Wang CL, Tang M, Li H. Targeted disruption of Nphp1 causes male infertility due to defects in the later steps of sperm morphogenesis in mice. Hum Mol Genet 2008; 17:3368-3379.

91. Mulryan K, Gitterman DP, Lewis CJ, Vial C, Leckie BJ, Cobb AL, Brown JE, Conley EC, Buell G, Pritchard CA, Evans RJ. Reduced vas deferens contraction and male infertility in mice lacking P2X1 receptors. Nature 2000; 403:86-89.

92. Baker SM, Bronner CE, Zhang L, Plug AW, Robatzek M, Warren G, Elliott EA, Yu J, Ashley T, Arnheim N, Flavell RA, Liskay RM. Male mice defective in the DNA mismatch repair gene PMS2 exhibit abnormal chromosome synapsis in meiosis. Cell 1995; 82:309-319.

93. Varmuza S, Jurisicova A, Okano K, Hudson J, Boekelheide K, Shipp EB. Spermiogenesis is impaired in mice bearing a targeted mutation in the protein phosphatase 1cgamma gene. Dev Biol 1999; 205:98-110.

94. Burton KA, McDermott DA, Wilkes D, Poulsen MN, Nolan MA, Goldstein M, Basson CT, McKnight GS. Haploinsufficiency at the protein kinase A RI alpha gene locus leads to fertility defects in male mice and men. Mol Endocrinol 2006; 20:2504-2513.

95. Cho C, Willis WD, Goulding EH, Jung-Ha H, Choi YC, Hecht NB, Eddy EM. Haploinsufficiency of protamine-1 or -2 causes infertility in mice. Nat Genet 2001; 28:82-86.

96. Khor B, Bredemeyer AL, Huang CY, Turnbull IR, Evans R, Maggi LB,Jr, White JM, Walker LM, Carnes K, Hess RA, Sleckman BP. Proteasome activator PA200 is required for normal spermatogenesis. Mol Cell Biol 2006; 26:2999-3007.

97. Lufkin T, Lohnes D, Mark M, Dierich A, Gorry P, Gaub MP, LeMeur M, Chambon P. High postnatal lethality and testis degeneration in retinoic acid receptor alpha mutant mice. Proc Natl Acad Sci U S A 1993; 90:7225-7229.

98. Brunner S, Colman D, Travis AJ, Luhmann UFO, Shi W, Feil S, Imsand C, Nelson J, Grimm C, Rulicke T, Fundele R, Neidhardt J, Berger W. Overexpression of RPGR Leads to Male Infertility in Mice Due to Defects in Flagellar Assembly. Biol Reprod 2008; 79:608-617.

99. Kastner P, Mark M, Leid M, Gansmuller A, Chin W, Grondona JM, Decimo D, Krezel W, Dierich A, Chambon P. Abnormal spermatogenesis in RXR beta mutant mice. Genes Dev 1996; 10:80-92.

100. Firestein R, Nagy PL, Daly M, Huie P, Conti M, Cleary ML. Male infertility, impaired spermatogenesis, and azoospermia in mice deficient for the pseudophosphatase Sbf1. J Clin Invest 2002; 109:1165-1172.

101. Pace AJ, Lee E, Athirakul K, Coffman TM, O'Brien DA, Koller BH. Failure of spermatogenesis in mouse lines deficient in the Na(+)-K(+)-2Cl(-) cotransporter. J Clin Invest 2000; 105:441-450.

102. Zhang Z, Kostetskii I, Tang W, Haig-Ladewig L, Sapiro R, Wei Z, Patel AM, Bennett J, Gerton GL, Moss SB, Radice GL, Strauss JF,3rd. Deficiency of SPAG16L causes male infertility associated with impaired sperm motility. Biol Reprod 2006; 74:751-759.

103. Zheng H, Stratton CJ, Morozumi K, Jin J, Yanagimachi R, Yan W. Lack of Spem1 causes aberrant cytoplasm removal, sperm deformation, and male infertility. Proc Natl Acad Sci U S A 2007; 104:6852-6857.

104. Hsu S, Hsieh-Li H, Li H. Dysfunctional spermatogenesis in transgenic mice overexpressing bHLH-Zip transcription factor, Spz1. Experimental Cell Research 2004; 294:185-198.

105. Pires-daSilva A, Nayernia K, Engel W, Torres M, Stoykova A, Chowdhury K, Gruss P. Mice deficient for spermatid perinuclear RNA-binding protein show neurologic, spermatogenic, and sperm morphological abnormalities. Dev Biol 2001; 233:319-328.

106. Wishart MJ, Dixon JE. The archetype STYX/dead-phosphatase complexes with a spermatid mRNA-binding protein and is essential for normal sperm production. Proc Natl Acad Sci U S A 2002; 99:2112-2117.

107. Zhong J, Peters AH, Lee K, Braun RE. A double-stranded RNA binding protein required for activation of repressed messages in mammalian germ cells. Nat Genet 1999; 22:171-174.

108. Zhang D, Penttila TL, Morris PL, Teichmann M, Roeder RG. Spermiogenesis deficiency in mice lacking the Trf2 gene. Science 2001; 292:1153-1155.

109. Martianov I, Fimia GM, Dierich A, Parvinen M, Sassone-Corsi P, Davidson I. Late arrest of spermiogenesis and germ cell apoptosis in mice lacking the TBP-like TLF/TRF2 gene. Mol Cell 2001; 7:509-515.

110. Jaroszynski L, Dev A, Li M, Meinhardt A, de Rooij DG, Mueller C, Böhm D, Wolf S, Adham IM, Wulf G, Engel W, Nayernia K. Asthenoteratozoospermia in mice lacking testis expressed gene 18 (Tex18). Mol Hum Reprod 2007; 13:155-163.

111. Yu YE, Zhang Y, Unni E, Shirley CR, Deng JM, Russell LD, Weil MM, Behringer RR, Meistrich ML. Abnormal spermatogenesis and reduced fertility in transition nuclear protein 1-deficient mice. Proc Natl Acad Sci U S A 2000; 97:4683-4688.

112. Zhao M, Shirley CR, Yu YE, Mohapatra B, Zhang Y, Unni E, Deng JM, Arango NA, Terry NH, Weil MM, Russell LD, Behringer RR, Meistrich ML. Targeted disruption of the transition protein 2 gene affects sperm chromatin structure and reduces fertility in mice. Mol Cell Biol 2001; 21:7243-7255.

113. Roest HP, van Klaveren J, de Wit J, van Gurp CG, Koken MH, Vermey M, van Roijen JH, Hoogerbrugge JW, Vreeburg JT, Baarends WM, Bootsma D, Grootegoed JA, Hoeijmakers JH. Inactivation of the HR6B ubiquitin-conjugating DNA repair enzyme in mice causes male sterility associated with chromatin modification. Cell 1996; 86:799-810.

114. Suetsugu S, Banzai Y, Kato M, Fukami K, Kataoka Y, Takai Y, Yoshida N, Takenawa T. Male-specific sterility caused by the loss of CR16. Genes Cells 2007; 12:721-733.

115. Krege JH, John SW, Langenbach LL, Hodgin JB, Hagaman JR, Bachman ES, Jennette JC, O'Brien DA, Smithies O. Male-female differences in fertility and blood pressure in ACE-deficient mice. Nature 1995; 375:146-148.

116. Nishimura H, Cho C, Branciforte DR, Myles DG, Primakoff P. Analysis of loss of adhesive function in sperm lacking cyritestin or fertilin beta. Dev Biol 2001; 233:204-213.

117. Cho C, Bunch DO, Faure JE, Goulding EH, Eddy EM, Primakoff P, Myles DG. Fertilization defects in sperm from mice lacking fertilin beta. Science 1998; 281:1857-1859.

118. Shamsadin R, Adham IM, Nayernia K, Heinlein UA, Oberwinkler H, Engel W. Male mice deficient for germ-cell cyritestin are infertile. Biol Reprod 1999; 61:1445-1451.

119. Kang-Decker N, Mantchev GT, Juneja SC, McNiven MA, van Deursen JM. Lack of acrosome formation in Hrb-deficient mice. Science 2001; 294:1531-1533.

120. Huang LS, Voyiaziakis E, Chen HL, Rubin EM, Gordon JW. A novel functional role for apolipoprotein B in male infertility in heterozygous apolipoprotein B knockout mice. Proc Natl Acad Sci U S A 1996; 93:10903-10907.

121. Lu Q, Hasty P, Shur BD. Targeted mutation in beta1,4-galactosyltransferase leads to pituitary insufficiency and neonatal lethality. Dev Biol 1997; 181:257-267.

122. Lu Q, Shur BD. Sperm from beta 1,4-galactosyltransferase-null mice are refractory to ZP3-induced acrosome reactions and penetrate the zona pellucida poorly. Development 1997; 124:4121-4131.

123. Jin J, Jin N, Zheng H, Ro S, Tafolla D, Sanders KM, Yan W. Catsper3 and Catsper4 are essential for sperm hyperactivated motility and male fertility in the mouse. Biol Reprod 2007; 77:37-44.

124. Ikawa M, Wada I, Kominami K, Watanabe D, Toshimori K, Nishimune Y, Okabe M. The putative chaperone calmegin is required for sperm fertility. Nature 1997; 387:607-611.

125. Xu X, Toselli PA, Russell LD, Seldin DC. Globozoospermia in mice lacking the casein kinase II alpha' catalytic subunit. Nat Genet 1999; 23:118-121.

126. Fisher CR, Graves KH, Parlow AF, Simpson ER. Characterization of mice deficient in aromatase (ArKO) because of targeted disruption of the cyp19 gene. Proc Natl Acad Sci U S A 1998; 95:6965-6970.

127. Robertson KM, O'Donnell L, Jones ME, Meachem SJ, Boon WC, Fisher CR, Graves KH, McLachlan RI, Simpson ER. Impairment of spermatogenesis in mice lacking a functional aromatase (cyp 19) gene. Proc Natl Acad Sci U S A 1999; 96:7986-7991.

128. Neesen J, Kirschner R, Ochs M, Schmiedl A, Habermann B, Mueller C, Holstein AF, Nuesslein T, Adham I, Engel W. Disruption of an inner arm dynein heavy chain gene results in asthenozoospermia and reduced ciliary beat frequency. Hum Mol Genet 2001; 10:1117-1128.

129. Cheng NC, van de Vrugt HJ, van der Valk MA, Oostra AB, Krimpenfort P, de Vries Y, Joenje H, Berns A, Arwert F. Mice with a targeted disruption of the Fanconi anemia homolog Fanca. Hum Mol Genet 2000; 9:1805-1811.

130. Whitney MA, Royle G, Low MJ, Kelly MA, Axthelm MK, Reifsteck C, Olson S, Braun RE, Heinrich MC, Rathbun RK, Bagby GC, Grompe M. Germ cell defects and hematopoietic hypersensitivity to gamma-interferon in mice with a targeted disruption of the Fanconi anemia C gene. Blood 1996; 88:49-58.

131. Ganaiem M, AbuElhija M, Lunenfeld E, Cherniy N, Weisze N, Itach SB, Breitbart H, Apte R, Huleihel M. Effect of IL-1 Receptor Antagonist Gene Deletion on Mouse Male Fertility. Endocrinology 2008;

132. Hellsten E, Evans JP, Bernard DJ, Janne PA, Nussbaum RL. Disrupted sperm function and fertilin beta processing in mice deficient in the inositol polyphosphate 5-phosphatase Inpp5b. Dev Biol 2001; 240:641-653.

133. Yu RN, Ito M, Saunders TL, Camper SA, Jameson JL. Role of Ahch in gonadal development and gametogenesis. Nat Genet 1998; 20:353-357.

134. Harris T, Marquez B, Suarez S, Schimenti J. Sperm motility defects and infertility in male mice with a mutation in Nsun7, a member of the Sun domain-containing family of putative RNA methyltransferases. Biol Reprod 2007; 77:376-382.

135. Dai X, Schonbaum C, Degenstein L, Bai W, Mahowald A, Fuchs E. The ovo gene required for cuticle formation and oogenesis in flies is involved in hair formation and spermatogenesis in mice. Genes Dev 1998; 12:3452-3463.

136. Mbikay M, Tadros H, Ishida N, Lerner CP, De Lamirande E, Chen A, El-Alfy M, Clermont Y, Seidah NG, Chretien M, Gagnon C, Simpson EM. Impaired fertility in mice deficient for the testicular germ-cell protease PC4. Proc Natl Acad Sci U S A 1997; 94:6842-6846.

137. Ueda Y, Yamaguchi R, Ikawa M, Okabe M, Morii E, Maeda Y, Kinoshita T. PGAP1 knock-out mice show otocephaly and male infertility. J Biol Chem 2007; 282:30373-30380.

138. Kobayashi Y, Watanabe M, Okada Y, Sawa H, Takai H, Nakanishi M, Kawase Y, Suzuki H, Nagashima K, Ikeda K, Motoyama N. Hydrocephalus, situs inversus, chronic sinusitis, and male infertility in DNA polymerase lambda-deficient mice: possible implication for the pathogenesis of immotile cilia syndrome. Mol Cell Biol 2002; 22:2769-2776.

139. Skalhegg BS, Huang Y, Su T, Idzerda RL, McKnight GS, Burton KA. Mutation of the Calpha subunit of PKA leads to growth retardation and sperm dysfunction. Mol Endocrinol 2002; 16:630-639.

140. Goffin V, Bouchard B, Ormandy CJ, Weimann E, Ferrag F, Touraine P, Bole-Feysot C, Maaskant RA, Clement-Lacroix P, Edery M, Binart N, Kelly PA. Prolactin: a hormone at the crossroads of neuroimmunoendocrinology. Ann N Y Acad Sci 1998; 840:498-509.

141. Yeung CH, Sonnenberg-Riethmacher E, Cooper TG. Infertile spermatozoa of c-ros tyrosine kinase receptor knockout mice show flagellar angulation and maturational defects in cell volume regulatory mechanisms. Biol Reprod 1999; 61:1062-1069.

142. Yeung CH, Wagenfeld A, Nieschlag E, Cooper TG. The cause of infertility of male c-ros tyrosine kinase receptor knockout mice. Biol Reprod 2000; 63:612-618.

143. Nayernia K, Adham IM, Burkhardt-Gottges E, Neesen J, Rieche M, Wolf S, Sancken U, Kleene K, Engel W. Asthenozoospermia in mice with targeted deletion of the sperm mitochondrion-associated cysteine-rich protein (Smcp) gene. Mol Cell Biol 2002; 22:3046-3052.

144. Perl A, Qian Y, Chohan KR, Shirley CR, Amidon W, Banerjee S, Middleton FA, Conkrite KL, Barcza M, Gonchoroff N, Suarez SS, Banki K. Transaldolase is essential for maintenance of the mitochondrial transmembrane potential and fertility of spermatozoa. Proc Natl Acad Sci U S A 2006; 103:14813-14818.

145. Roy A, Lin YN, Agno JE, DeMayo FJ, Matzuk MM. Absence of tektin4 causes asthenozoospermia and subfertility in male mice. FASEB J 2007; 21:1013-1025.

146. Borghei A, Ouyang YB, Westmuckett AD, Marcello MR, Landel CP, Evans JP, Moore KL. Targeted disruption of tyrosylprotein sulfotransferase-2, an enzyme that catalyzes post-translational protein tyrosine O-sulfation, causes male infertility. J Biol Chem 2006; 281:9423-9431.

147. Sampson MJ, Decker WK, Beaudet AL, Ruitenbeek W, Armstrong D, Hicks MJ, Craigen WJ. Immotile sperm and infertility in mice lacking mitochondrial voltage-dependent anion channel type 3. J Biol Chem 2001; 276:39206-39212.

148. Lin YN, Roy A, Yan W, Burns KH, Matzuk MM. Loss of zona pellucida binding proteins in the acrosomal matrix disrupts acrosome biogenesis and sperm morphogenesis. Mol Cell Biol 2007; 27:6794-6805.

149. Mhaouty-Kodja S, Lozach A, Habert R, Tanneux M, Guigon C, Brailly-Tabard S, Maltier JP, Legrand-Maltier C. Fertility and spermatogenesis are altered in {alpha}1b-adrenergic receptor knockout male mice. J Endocrinol 2007; 195:281-292.

150. Charest NJ, Zhou ZX, Lubahn DB, Olsen KL, Wilson EM, French FS. A frameshift mutation destabilizes androgen receptor messenger RNA in the Tfm mouse. Mol Endocrinol 1991; 5:573-581.

151. Topilko P, Schneider-Maunoury S, Levi G, Trembleau A, Gourdji D, Driancourt MA, Rao CV, Charnay P. Multiple pituitary and ovarian defects in Krox-24 (NGFI-A, Egr-1)-targeted mice. Mol Endocrinol 1998; 12:107-122.

152. Kinuta K, Tanaka H, Moriwake T, Aya K, Kato S, Seino Y. Vitamin D is an important factor in estrogen biosynthesis of both female and male gonads. Endocrinology 2000; 141:1317-1324.

153. Zhongyi S, Rantakari P, Lamminen T, Toppari J, Poutanen M. Transgenic male mice expressing human hydroxysteroid dehydrogenase 2 indicate a role for the enzyme independent of its action on sex steroids. Endocrinology 2007; 148:3827-3836.

154. Qin J, Tsai M, Tsai SY. Essential Roles of COUP-TFII in Leydig Cell Differentiation and Male Fertility. PLoS ONE 2008; 3:e3285.

155. Johnson LE, DeLuca HF. Vitamin D receptor null mutant mice fed high levels of calcium are fertile. J Nutr 2001; 131:1787-1791.

156. Matzuk MM, Kumar TR, Bradley A. Different phenotypes for mice deficient in either activins or activin receptor type II. Nature 1995; 374:356-360.

157. Li SW, Arita M, Fertala A, Bao Y, Kopen GC, Langsjo TK, Hyttinen MM, Helminen HJ, Prockop DJ. Transgenic mice with inactive alleles for procollagen N-proteinase (ADAMTS-2) develop fragile skin and male sterility. Biochem J 2001; 355:271-278.

158. Behringer RR, Finegold MJ, Cate RL. Mullerian-inhibiting substance function during mammalian sexual development. Cell 1994; 79:415-425.

159. Durlinger AL, Kramer P, Karels B, de Jong FH, Uilenbroek JT, Grootegoed JA, Themmen AP. Control of primordial follicle recruitment by anti-Mullerian hormone in the mouse ovary. Endocrinology 1999; 140:5789-5796.

160. Mishina Y, Rey R, Finegold MJ, Matzuk MM, Josso N, Cate RL, Behringer RR. Genetic analysis of the Mullerian-inhibiting substance signal transduction pathway in mammalian sexual differentiation. Genes Dev 1996; 10:2577-2587.

161. Kojima S, Hatano M, Okada S, Fukuda T, Toyama Y, Yuasa S, Ito H, Tokuhisa T. Testicular germ cell apoptosis in Bcl6-deficient mice. Development 2001; 128:57-65.

162. Zhao GQ, Liaw L, Hogan BL. Bone morphogenetic protein 8A plays a role in the maintenance of spermatogenesis and the integrity of the epididymis. Development 1998; 125:1103-1112.

163. Hudson DF, Fowler KJ, Earle E, Saffery R, Kalitsis P, Trowell H, Hill J, Wreford NG, de Kretser DM, Cancilla MR, Howman E, Hii L, Cutts SM, Irvine DV, Choo KH. Centromere protein B null mice are mitotically and meiotically normal but have lower body and testis weights. J Cell Biol 1998; 141:309-319.

164. Fowler KJ, Hudson DF, Salamonsen LA, Edmondson SR, Earle E, Sibson MC, Choo KH. Uterine dysfunction and genetic modifiers in centromere protein B-deficient mice. Genome Res 2000; 10:30-41.

165. Kendall SK, Samuelson LC, Saunders TL, Wood RI, Camper SA. Targeted disruption of the pituitary glycoprotein hormone alpha-subunit produces hypogonadal and hypothyroid mice. Genes Dev 1995; 9:2007-2019.

166. Gow A, Southwood CM, Li JS, Pariali M, Riordan GP, Brodie SE, Danias J, Bronstein JM, Kachar B, Lazzarini RA. CNS myelin and sertoli cell tight junction strands are absent in Osp/claudin-11 null mice. Cell 1999; 99:649-659.

167. Cohen PE, Zhu L, Pollard JW. Absence of colony stimulating factor-1 in osteopetrotic (csfmop/csfmop) mice disrupts estrous cycles and ovulation. Biol Reprod 1997; 56:110-118.

168. Luong MX, van der Meijden CM, Xing D, Hesselton R, Monuki ES, Jones SN, Lian JB, Stein JL, Stein GS, Neufeld EJ, van Wijnen AJ. Genetic ablation of the CDP/Cux protein C terminus results in hair cycle defects and reduced male fertility. Mol Cell Biol 2002; 22:1424-1437.

169. Hu MC, Hsu NC, El Hadj NB, Pai CI, Chu HP, Wang CK, Chung BC. Steroid deficiency syndromes in mice with targeted disruption of Cyp11a1. Mol Endocrinol 2002; 16:1943-1950.

170. Miyamoto N, Yoshida M, Kuratani S, Matsuo I, Aizawa S. Defects of urogenital development in mice lacking Emx2. Development 1997; 124:1653-1664.

171. Laing MA, Coonrod S, Hinton BT, Downie JW, Tozer R, Rudnicki MA, Hassell JA. Male sexual dysfunction in mice bearing targeted mutant alleles of the PEA3 ets gene. Mol Cell Biol 2000; 20:9337-9345.

172. Koomen M, Cheng NC, van de Vrugt HJ, Godthelp BC, van der Valk MA, Oostra AB, Zdzienicka MZ, Joenje H, Arwert F. Reduced fertility and hypersensitivity to mitomycin C characterize Fancg/Xrcc9 null mice. Hum Mol Genet 2002; 11:273-281.

173. Colvin JS, Green RP, Schmahl J, Capel B, Ornitz DM. Male-to-female sex reversal in mice lacking fibroblast growth factor 9. Cell 2001; 104:875-889.

174. Kooy RF, D'Hooge R, Reyniers E, Bakker CE, Nagels G, De Boulle K, Storm K, Clincke G, De Deyn PP, Oostra BA, Willems PJ. Transgenic mouse model for the fragile X syndrome. Am J Med Genet 1996; 64:241-245.

175. Settle S, Marker P, Gurley K, Sinha A, Thacker A, Wang Y, Higgins K, Cunha G, Kingsley DM. The BMP family member Gdf7 is required for seminal vesicle growth, branching morphogenesis, and cytodifferentiation. Dev Biol 2001; 234:138-150.

176. Kumar TR, Wiseman AL, Kala G, Kala SV, Matzuk MM, Lieberman MW. Reproductive defects in gamma-glutamyl transpeptidase-deficient mice. Endocrinology 2000; 141:4270-4277.

177. Lieberman MW, Wiseman AL, Shi ZZ, Carter BZ, Barrios R, Ou CN, Chevez-Barrios P, Wang Y, Habib GM, Goodman JC, Huang SL, Lebovitz RM, Matzuk MM. Growth retardation and cysteine deficiency in gamma-glutamyl transpeptidase-deficient mice. Proc Natl Acad Sci U S A 1996; 93:7923-7926.

178. Couldrey C, Carlton MB, Nolan PM, Colledge WH, Evans MJ. A retroviral gene trap insertion into the histone 3.3A gene causes partial neonatal lethality, stunted growth, neuromuscular deficits and male sub-fertility in transgenic mice. Hum Mol Genet 1999; 8:2489-2495.

179. Lee YH, Sauer B, Gonzalez FJ. Laron dwarfism and non-insulin-dependent diabetes mellitus in the Hnf-1alpha knockout mouse. Mol Cell Biol 1998; 18:3059-3068.

180. Satokata I, Benson G, Maas R. Sexually dimorphic sterility phenotypes in Hoxa10-deficient mice. Nature 1995; 374:460-463.

181. Hsieh-Li HM, Witte DP, Weinstein M, Branford W, Li H, Small K, Potter SS. Hoxa 11 structure, extensive antisense transcription, and function in male and female fertility. Development 1995; 121:1373-1385.

182. Matzuk MM, Finegold MJ, Su JG, Hsueh AJ, Bradley A. Alpha-inhibin is a tumour-suppressor gene with gonadal specificity in mice. Nature 1992; 360:313-319.

183. Kumar TR, Wang Y, Matzuk MM. Gonadotropins are essential modifier factors for gonadal tumor development in inhibin-deficient mice. Endocrinology 1996; 137:4210-4216.

184. Swerdloff RS, Batt RA, Bray GA. Reproductive hormonal function in the genetically obese (ob/ob) mouse. Endocrinology 1976; 98:1359-1364.

185. Swerdloff RS, Peterson M, Vera A, Batt RA, Heber D, Bray GA. The hypothalamic-pituitary axis in genetically obese (ob/ob) mice: response to luteinizing hormone-releasing hormone. Endocrinology 1978; 103:542-547.

186. Chua SC,Jr, Chung WK, Wu-Peng XS, Zhang Y, Liu SM, Tartaglia L, Leibel RL. Phenotypes of mouse diabetes and rat fatty due to mutations in the OB (leptin) receptor. Science 1996; 271:994-996.

187. Bessone S, Vidal F, Le Bouc Y, Epelbaum J, Bluet-Pajot MT, Darmon M. EMK protein kinase-null mice: dwarfism and hypofertility associated with alterations in the somatotrope and prolactin pathways. Dev Biol 1999; 214:87-101.

188. Bruning JC, Gautam D, Burks DJ, Gillette J, Schubert M, Orban PC, Klein R, Krone W, Muller-Wieland D, Kahn CR. Role of brain insulin receptor in control of body weight and reproduction. Science 2000; 289:2122-2125.

189. Luo X, Ikeda Y, Parker KL. A cell-specific nuclear receptor is essential for adrenal and gonadal development and sexual differentiation. Cell 1994; 77:481-490.

190. Li S, Crenshaw EB,3rd, Rawson EJ, Simmons DM, Swanson LW, Rosenfeld MG. Dwarf locus mutants lacking three pituitary cell types result from mutations in the POU-domain gene pit-1. Nature 1990; 347:528-533.

191. Choi J, Nannenga B, Demidov ON, Bulavin DV, Cooney A, Brayton C, Zhang Y, Mbawuike IN, Bradley A, Appella E, Donehower LA. Mice deficient for the wild-type p53-induced phosphatase gene (Wip1) exhibit defects in reproductive organs, immune function, and cell cycle control. Mol Cell Biol 2002; 22:1094-1105.

192. Ng JM, Vrieling H, Sugasawa K, Ooms MP, Grootegoed JA, Vreeburg JT, Visser P, Beems RB, Gorgels TG, Hanaoka F, Hoeijmakers JH, van der Horst GT. Developmental defects and male sterility in mice lacking the ubiquitin-like DNA repair gene mHR23B. Mol Cell Biol 2002; 22:1233-1245.

193. Lohnes D, Kastner P, Dierich A, Mark M, LeMeur M, Chambon P. Function of retinoic acid receptor gamma in the mouse. Cell 1993; 73:643-658.

194. Overbeek PA, Gorlov IP, Sutherland RW, Houston JB, Harrison WR, Boettger-Tong HL, Bishop CE, Agoulnik AI. A transgenic insertion causing cryptorchidism in mice. Genesis 2001; 30:26-35.

195. Murer V, Spetz JF, Hengst U, Altrogge LM, de Agostini A, Monard D. Male fertility defects in mice lacking the serine protease inhibitor protease nexin-1. Proc Natl Acad Sci U S A 2001; 98:3029-3033.

196. Uhrin P, Dewerchin M, Hilpert M, Chrenek P, Schofer C, Zechmeister-Machhart M, Kronke G, Vales A, Carmeliet P, Binder BR, Geiger M. Disruption of the protein C inhibitor gene results in impaired spermatogenesis and male infertility. J Clin Invest 2000; 106:1531-1539.

197. Ohtsuka S, Takaki S, Iseki M, Miyoshi K, Nakagata N, Kataoka Y, Yoshida N, Takatsu K, Yoshimura A. SH2-B is required for both male and female reproduction. Mol Cell Biol 2002; 22:3066-3077.

198. Supp DM, Witte DP, Branford WW, Smith EP, Potter SS. Sp4, a member of the Sp1-family of zinc finger transcription factors, is required for normal murine growth, viability, and male fertility. Dev Biol 1996; 176:284-299.

199. Caron KM, Soo SC, Wetsel WC, Stocco DM, Clark BJ, Parker KL. Targeted disruption of the mouse gene encoding steroidogenic acute regulatory protein provides insights into congenital lipoid adrenal hyperplasia. Proc Natl Acad Sci U S A 1997; 94:11540-11545.

200. Lee HW, Blasco MA, Gottlieb GJ, Horner JW,2nd, Greider CW, DePinho RA. Essential role of mouse telomerase in highly proliferative organs. Nature 1998; 392:569-574.

201. Smith CL, DeVera DG, Lamb DJ, Nawaz Z, Jiang YH, Beaudet AL, O'Malley BW. Genetic ablation of the steroid receptor coactivator-ubiquitin ligase, E6-AP, results in tissue-selective steroid hormone resistance and defects in reproduction. Mol Cell Biol 2002; 22:525-535.

202. Kreidberg JA, Sariola H, Loring JM, Maeda M, Pelletier J, Housman D, Jaenisch R. WT-1 is required for early kidney development. Cell 1993; 74:679-691.

203. Parr BA, McMahon AP. Sexually dimorphic development of the mammalian reproductive tract requires Wnt-7a. Nature 1998; 395:707-710.