Overview

This spontaneous mutation in the lipoma HMGIC fusion partner-like 2 gene disrupts the development of the distal reproductive tract in both females and males and causes corneal opacity, exophthalmos, and possibly hearing impairment.
This spontaneous mutation causes ocular defects and developmental defects in both the male and female distal reproductive systems. Homozygotes are reported to have corneal hazing, exophthalmos, and increased pupil movement, and auditory brainstem response testing of two homozygotes indicated elevated thresholds. Homozygous females can be identified between four to five weeks of age by complete closure of the vagina and a soft, reducible swelling of the perineum. Viscous fluid builds up in the uteri and females do not reproduce, but do live a normal lifespan. The Mullerian duct tips fail to merge or enter the urogenital sinus at E15.5, which results in an unmerged upper vagina and vaginal atresia. Although the ovaries have normal histology and function and the uterus seems to develop normally too, during the estrus stage the reproductive tracts are larger than normal with 30% having a dark appearance in the uterus and upper vagina. The distended uterine horns have an enlarged lumen filled with viscous fluid, the uterine and vaginal walls are compressed, and the upper vagina has enlarged, fluid-filled double lumens. Approximately 70% of male homozygotes are sterile due to failed development in the distal vas deferens. The tips of the Wolffian ducts fail to merge with the urogenital sinus, which results in convolutions at the junctions of the vas deferens and urethra. In the majority of males these knots in the distal vas deferens do not permit the passage of sperm. Some homozygotes have abnormal seminal vesicles on one or both sides. The sperm counts from cauda epididymis were found to be normal, and the lumen of the adult vas deferens was found to be dense with sperm.
Genetics

Lhfpl2<sup>vgim</sup>

Disease/Phenotype

Disease Terms

Research Areas By Phenotype

Mammalian Phenotype Terms by Genotype

References

Technical Support

Genotyping Protocols
Genotyping resources and troubleshooting

Mating System
Ovarian Transplant-Cross-Intercross

Citation
When using the B6(129S4)-Lhfpl2<sup>vgim</sup>/GrsrJ mouse strain in a publication, please cite the originating article(s) and include JAX stock #013716 in your Materials and Methods section.

Animal Health Reports
Facility Barrier Level Descriptions

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