

## VETERINARY TECHNICAL DATASHEET

Persistent Müllerian Duct Syndrome, (PMDS); mutation originally found in Miniature Schnauzer



Mutation Found In :Schnauzer (Miniature)

### Disorder Type

- Reproductive system

### Disease Severity

- Moderate

### Background

Persistent Müllerian duct syndrome (PMDS, pseudohermaphroditism) is a disorder of sexual development affecting male Miniature Schnauzers. PMDS is characterized by Müllerian duct derivatives (e.g., uterus) developing in otherwise externally normal-appearing males. The condition is inherited in a sex-limited, autosomal recessive manner, i.e., the disease only manifests in males, although it can be inherited via females.

### Key Signs

- Internal female reproductive organs in externally normal males
- Cryptorchidism
- Sterility or subfertility
- Uterine infections
- Sertoli cell tumor

### Clinical Description

The males affected with PMDS appear externally normal, but they have a uterus, cervix, part of the vagina, and fallopian tubes. Approximately 50% of PMDS-affected males are cryptorchid (absence of one or both testes from the scrotum). PMDS is characterized by sterility or subfertility, but affected males that are unilateral cryptorchids have sired litters. Aged PMDS dogs may develop pyometra (uterine infection) or a Sertoli cell tumor.

### Mode of Inheritance

- autosomal recessive

### Gene Name

- AMHR2

### Next Steps

Treatment is surgical removal of any female reproductive organs and retained testes to eliminate the risk of uterine infections or Sertoli cell tumors. It is also recommended that any descended testes be removed to prevent unintended matings that could allow the mutation to be passed on to puppies.

### References

Wu X, Wan S, Pujar S, Haskins ME, Schlafer DH, Lee MM, Meyers-Wallen VN. A single base pair mutation encoding a premature stop codon in the MIS type II receptor is responsible for canine persistent Müllerian duct syndrome. J Androl. 30(1):46-56, 2009.