

VETERINARY TECHNICAL DATASHEET

Severe Combined Immunodeficiency, (SCID); mutation originally found in Frisian Water Dogs



Mutation Found In :Frisian Water Dog

Disorder Type

- Immune system

Disease Severity

- Severe

Background

Severe combined immunodeficiency (X-SCID) is a severe dysfunction of the immune system; this variant is associated with the Frisian Water Dogs or Wetterhouns. The mode of inheritance is autosomal recessive.

Key Signs

- Poor growth
- Diarrhea
- Vomiting
- Seizures
- Ataxia
- Blindness
- Depletion of lymphoid tissues
- Reduced level of immunoglobulin
- Reduced levels of lymphocytes

Clinical Description

Severe combined immunodeficiency is characterized by T- and B-lymphocyte dysfunction resulting in a non-functional immune system. Affected dogs are predisposed to infections. The onset of signs typically occurs at 7 to 8 weeks of age and includes lethargy, poor growth, vomiting, diarrhea, ataxia, seizures, and blindness. Affected dogs rarely survive over 4 months of age. There is no treatment available for SCID.

Mode of Inheritance

- autosomal recessive

Gene Name

- RAG1

Next Steps

Therapy is limited to symptomatic treatments for resulting infections and general supportive care. There is no cure.

References

Verfuurden B, Wempe F, Reinink P, van Kooten PJ, Martens E, Gerritsen R, Vos JH., Rutten VP, Leegwater PA. Severe combined immunodeficiency in Frisian Water Dogs caused by a RAG1 mutation. Genes Immun 12:310-3, 2011.

