

# 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.

