

# VETERINARY TECHNICAL DATASHEET

X-linked Severe Combined Immunodeficiency, (XSCID); mutation originally found in Cardigan Welsh Corgi



Mutation Found In :Cardigan Welsh Corgi

## Disorder Type

- Immune system

## Disease Severity

- Severe

## Background

X-linked severe combined immunodeficiency (X-SCID) is a severe dysfunction of the immune system; this variant is associated with the Cardigan Welsh Corgi. The mode of inheritance is X-linked recessive and, therefore, affected dogs tend to be males.

## Key Signs

- Immunodeficiency
- Lymphopenia
- Lack of lymph nodes
- Vomiting
- Diarrhea
- Failure to thrive
- Opportunistic infections

## Clinical Description

X-SCID is a severe immunodeficiency, which results from abnormally low levels of lymphocytes (white blood cells) and their dysfunctionality. Lymphatic tissue of affected dogs is also underdeveloped. Affected dogs are extremely susceptible to opportunistic infections and typically die at a very young age.

## Mode of Inheritance

- X-linked

## Gene Name

- IL2RG

## Next Steps

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

## References

Henthorn PS, Somberg RL, Fimiani VM, Puck JM, Patterson DF, Felsburg PJ. IL-2R gamma gene microdeletion demonstrates that canine X- linked severe combined immunodeficiency is a homologue of the human disease. Genomics 23:69-74, 1994.

Somberg RL, Pullen RP, Casal ML, Patterson DF, Felsburg PJ, Henthorn PS. A single nucleotide insertion in the canine interleukin-2 receptor gamma chain results in X-linked severe combined immunodeficiency disease. Vet Immunol Immunopathol 47:203-213, 1995.

