VETERINARY TECHNICAL DATASHEET

Autosomal Recessive Severe Combined Immunodeficiency, (ARSCID)

Mutation Found In :Russell Terrier, Parson Russell Terrier

Disorder Type

- Immune system

Disease Severity

- Severe

Background

Autosomal recessive severe combined immunodeficiency (ARSCID) is a severe immunodeficiency disorder discovered in Jack Russell Terriers. Affected dogs are highly susceptible to recurrent infections and usually die at a young age.

Key Signs

- Immunodeficiency
- Lymphopenia
- Lymphoid hypoplasia

Clinical Description

ARSCID causes severe immunodeficiency because of the low number of white blood cells (lymphopenia) involved in the body's immune defenses. There is a complete absence of the IgM antibodies. Affected puppies show incomplete development of the lymphoid tissue so they are highly susceptible to recurrent infections and usually die at a young age due to infection, after the maternal antibodies they received while nursing decline. Necropsy results show that all the lymphoid tissues fail to develop.

Mode of Inheritance

- autosomal recessive

Gene Name

- PRKDC

Next Steps

Therapy is targeted at treating the secondary infections and affected puppies should not be treated with modified-live vaccines. Humane euthanasia for affected puppies is often elected.

References

Bell TG, Butler KL, Sill HB, Stickle JE, Ramos-Vara JA, Dark MJ. Autosomal recessive severe combined immunodeficiency of Jack Russell Terriers. J Vet Diagn Invest 14:194-204, 2002.

Ding, Q., Bramble, L., Yuzbasiyan-Gurkan, V., Bell, T., Meek, K. DNA-PKcs mutations in dogs and horses: allele frequency and association with neoplasia. Gene 283:263-269, 2002.

Meek K, Kienker L, Dallas C, Wang W, Dark MJ, Venta PJ, Huie ML, Hirschhorn R, Bell T. SCID in Jack Russell terriers: a new animal model of DNA-PKcs deficiency. J Immunol 167:2142-50, 2001