

VETERINARY TECHNICAL DATASHEET

Factor VIII Deficiency or Hemophilia A; p.Cys548Tyr mutation originally found in German Shepherd Dog



Mutation Found In :German Shepherd Dog

Disorder Type

- Blood

Disease Severity

- Moderate/severe

Background

Hemophilia A is an inherited bleeding disorder encountered in several breeds. Hematomas or abdominal bleeding without apparent reason may be observed in a severely affected dog. If untreated, the disorder can lead to death caused by bleeding. The condition is usually more severe in large, active dogs. The disease follows an X-linked mode of inheritance and is therefore more commonly observed in male dogs as males only have one X chromosome and a single affected copy will cause the condition; females require two copies to exhibit the condition.

Key Signs

- Potentially life threatening bleeding events
- Hematomas
- Abdominal bleeding

Clinical Description

Blood coagulation is a complex process. Factor VIII is one of the proteins necessary for the blood coagulation process and a deficiency of this protein causes hemophilia A in an affected dog. Clinical signs of hemophilia A vary depending on the activity of factor VIII in the blood. Specific factor assay may be measured by a reference laboratory. Hematomas or abdominal bleeding without apparent reason may be observed in a severely affected dog. If untreated, the disorder can lead to death caused by bleeding. The condition is usually more severe in large, active dogs. Prior to surgery or invasive procedures, a prothrombin (PT) and partial thromboplastin time (PTT) should be measured. Additional supportive measures, including transfusions, may be necessary.

Mode of Inheritance

- X-linked

Gene Name

- FVIII

Next Steps

Exceptionally excessive and prolonged bleeding may be observed during shedding of deciduous teeth, routine surgeries, and even minor traumas. Prior to surgery or invasive procedures, a prothrombin (PT) and partial thromboplastin time (PTT) should be measured. Affected dogs should be monitored closely for excessive and prolonged bleeding during and after any required surgical procedures or after any trauma. Transfusions of cryoprecipitate or fresh-frozen plasma should be provided as necessary to ensure proper clotting if other means are unsuccessful.

References

Wilhelm C, Czwalińska A, Hoffmann M, Mischke R, Ganser A, Von Depka M. A mutation leading to a stop codon in the FVIII gene is the cause of severe canine Hemophilia A. J Thromb Haemost 1 Sup P0672, 2013.

