VETERINARY TECHNICAL DATASHEET

Pyruvate Kinase Deficiency; mutation originally found in West Highland White Terrier

Mutation Found In :Cairn Terrier, West Highland White Terrier

Disorder Type

- Blood

Disease Severity

- Moderate/severe

Background

Pyruvate kinase deficiency is a disorder causing hemolytic anemia and usually results in the death of affected individuals before they reach 5 years of age. The disease follows an autosomal recessive mode of inheritance.

Key Signs

- Anemia
- Drowsiness
- Pale mucous membranes
- Enlargement of the spleen and liver
- Osteosclerosis
- Myelofibrosis

Clinical Description

Pyruvate kinase (PK) is an enzyme needed for normal energy production by the red blood cells. PK deficiency affects the life span of erythrocytes (red blood cells) that break down particularly easily, which results in hemolytic anemia. Clinical signs include reduced exercise tolerance, weakness, pale mucous membranes, slowed growth, and a heart murmur. Accumulation of iron released from red blood cells damages the liver and bone marrow, resulting in liver failure and abnormal bone density. The disease leads to the death of the affected dog, usually at less than 5 years of age.

Mode of Inheritance

- autosomal recessive

Gene Name

- PKLR

Next Steps

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

References

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