

Delayed-Onset Retinal Degeneration_CLN3

Client Name:	SA Studbook (SAB005)	Report No:	ZO2026/11301/20260625/#125807
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Profile:	ZOCA2026/113900	Species:	Bos taurus / Bovid / Cattle
Breed:	Hereford	Registration #:	96121306
Test:	[CLN3] Delayed-Onset Retinal Degeneration_CLN3		
Results:	c.1106delG	G/-	Carrier

Sample Type: Hair (Cattle and Wildlife only)	Extraction Method: Direct PCR	Test Type: Genetic Health
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[CLN3] Delayed-Onset Retinal Degeneration_CLN3

Delayed blindness (DB) in Hereford cattle is a hereditary retinal disorder characterised by progressive loss of vision beginning in young adulthood. Early signs include difficulty navigating unfamiliar environments and reduced visual responsiveness, progressing to complete blindness. Molecular studies have identified a causative mutation in the CLN3 (Ceroid Lipofuscinosis Neuronal 3) gene.

The mutation is described as **c.1106del (p.Pro369Argfs*8)**.

The disorder is caused by a single base pair deletion in exon 16 of the *CLN3* gene (chromosome 25 in *Bos taurus*) resulting in a frameshift mutation. This frameshift introduces a premature stop codon, producing a truncated battenin protein with an altered C-terminal region. Battenin is involved in lysosomal function and cellular maintenance, particularly within retinal photoreceptor cells.

Delayed blindness in Hereford cattle follows an autosomal recessive pattern of inheritance. Homozygous normal animals (G/G) are unaffected and will not transmit the mutation. Heterozygous carrier animals (G/-) are clinically normal but carry one copy of the mutation and can pass it to offspring. Homozygous affected animals (-/-) will develop delayed blindness.

References: Hughes, A. M., et al. (2024). Identification of a frameshift mutation in CLN3 associated with delayed blindness in Hereford cattle.

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