

## **Complex Vertebral Malformation and Factor XI Deficiency in Friesian-Holstein**

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<b>Judul Thesis</b>	:	Deteksi Dini Kelainan Genetik Complex Vertebral Malformation dan Factor XI Deficiency pada Sapi Perah Friesian-Holstein Early Detection of Genetic Disorder Complex Vertebral Malformation and Factor XI Deficiency in Friesian-Holstein

### **Abstract:**

Complex vertebral malformation (CVM) and factor XI deficiency (F11D) are genetic disorders caused by autosomal recessive genes in cattle. CVM is due to mutation in SLC35A3 gene exon 4 chromosome BTA3 encoding for a UDP-N-acetylglucosamine transporter. The substitution of guanine by thymine at nucleotide position 559 in SLC35A3 gene will change valine by phenylalanine at position 180 (V180F). Calves affected by this mutation have numerous anatomic deformations. While F11D is due to insertion of 76 bp in a Factor XI gene exon 12 at chromosomes 17. Calves affected by this insertion can be asymptomatic or show a number of indicator that may include prolonged bleeding after injection, production of bloody milk and anemia. The aim of research was to develop detecting method of the genetic disorder of CVM and F11D of Holstein-Friesian cattle by polymerase chain reaction (PCR) method. Genotyping of 676 Holstein-Friesian blood sample from breeding center and small-scale husbandaries had been analyzed. Five samples of all successful amplified were detected as F11D carriers but no CVM allele was found. As F11D carrier, animal was in normal performance. The F11D carrier supposed came from old population before last decades. Eventhough the Indonesian's Holstein-Friesian population lack CVM carrier, all stakeholder needs to be aware. The early warning system resulted from this research could be useful to prevent the introduction of mutated alleles in Indonesian cattle population.

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