ESTABLISHMENT OF A PHENOTYPICAL MODEL OF ADVERSE OUTCOMES ASSOCIATED WITH ASSISTED REPRODUCTIVE TECHNOLOGIES

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ABSTRACT

Beckwith-Wiedemann syndrome (BWS) is a loss-of-imprinting pediatric overgrowth syndrome. BWS is speculated to occur primarily as the result of the misregulation of imprinted genes associated with two clusters on chromosome 11p15.5, namely the KvDMR1 and *H19/IGF2*. There is a similar overgrowth phenotype that is observed in ruminants as a result of embryo culture. This syndrome is known as large offspring syndrome (LOS). The genomic region/s associated with LOS have not yet been determined. We hypothesized that BWS and LOS are epigenetically similar. The aim of this research was to ascertain baseline allelic expression and DNA methylation in bovine of imprinted loci known to be misregulated in BWS. We conclude that the imprinted gene expression of *KCNQ1OT1*, *CDKN1C*, *H19*, and *PLAGL1* are conserved between the bovine and human. In addition, the KvDMR1 and *H19/IGF2* imprinting control regions also have conserved DNA methylation patterns between humans and bovine.