

Public Abstract

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

Beckwith-Wiedemann syndrome (BWS) is a loss-of-imprinting pediatric overgrowth syndrome that has an incidence of 1 in 13,700. Imprinted genes are expressed from only one of the two inherited parental chromosomes. These genes regulate the growth and development of the fetus and the placenta. The primary features of BWS include general overgrowth, enlarged tongue, abdominal wall defects, hypoglycemia, cardiac malformations, and embryonic tumors. Currently there are no animal models that recapitulate BWS. However, there is a similar overgrowth phenotype observed in ruminants as the result of embryo culture called large offspring syndrome (LOS). The phenotypes associated with LOS are increased birth weight, enlargement of internal organs, skeletal defects, hypoglycemia, and breathing difficulties. We propose that BWS and LOS are the result of similar loss-of-imprinting. In order to determine if the bovine will serve as a good model to study BWS we must first establish what the baseline gene expression and DNA methylation is in the bovine for the imprinted loci associated with BWS. Our study shows that genomic imprinting is conserved between human and bovine at the imprinted regions known to be misregulated in BWS. Future work will determine if LOS and BWS are epigenetically similar. If so, the bovine will serve as an appropriate animal model for studying the pediatric overgrowth syndrome BWS. Using the bovine as a model will allow researchers to understand the mechanisms that result in BWS and may provide therapeutic treatment options for these patients.