

Abstracts - 37th Annual Meeting of the Association of Embryo Technology in Europe (AETE) Physiology of reproduction in male and semen technology Genomic analysis of bovine beta-defensin genes implicated in sperm function fertility

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Beta-defensins are short secreted peptides that have traditionally been characterised as antimicrobial. However, it is known that some beta-defensins have important functions in fertility. For example, beta-defensin 126 is an important component of the sperm glycocalyx and is involved in capacitation of sperm prior to fertilisation in macaques, and promotes sperm motility in cattle. Genes encoding beta-defensins are known to show extensive deletion and duplication (known as copy number variation (CNV)) in cattle and other mammals, including humans. However this has not been characterised in detail in cattle. In this study we mapped publicly available sequencing data (1000 Bulls Consortium) from 100 bulls of different breeds to the bovine genome (ARS-UCD1.2-bosTau9) assembly based on long read PacBio sequencing, which is likely to have a more accurate assembly of complex repeated regions of the genome, such as the beta-defensin regions. We show that the chromosome 13 beta-defensins, including beta-defensin 126, are commonly duplicated in Holstein bulls. Other beta-defensins on chromosome 27, in particular DEFB103, show extensive copy number variation across breeds, including complete loss of the gene. In addition we selected positive control bulls target that high and low copy numbers associated with beta-defensin by the mapping. We are currently testing an extended fertility panel of Holstein bulls and the positive control bulls for beta-defensin genes showing CNV using digital droplet PCR, with the aim of investigating the relationship between CNV at bovine beta-defensin genes and fertility.

Our results may improve our understanding of CNV, which is known as an important genomic structural variation in cattle. We also believe that beta-defensin will provide important information about potential CNV effects on reproductive performance, which forms the basis for its inclusion in a future dairy cattle breeding programme.

Keywords: CNV, Beta-defensin, Fertility, Cattle