Holstein bull fertility linked to more copies of a testis-specific protein in the genome

**What is this research about?**

Some segments of DNA are repeated multiple times in a plant or animal genome (all the genetic material in an individual). The number of copies of these segments can vary from individual to individual. Genes with copy number variants (CNVs) are an important source of genetic variation. Studies have shown that differences in gene copy number can affect the cell (through activation of other genes) and the physical characteristics of the organism as a whole. More importantly, these changes in gene activation or physical characteristics can influence the risk for several different diseases. The TSPY protein (testis-specific protein y-encoded) is an example of a CNV that is found on the Y (male) sex chromosome in many different species, including humans and cows. The role of TSPY in sperm production and fertility is not known, but it may speed up the life cycle of sperm-producing cells, or regulate hormone receptors in the testes. While some studies have found a link between TSPY copy number and sperm counts, the results have been conflicting and no research has been done in cows.

**What did the researchers do?**

Using DNA analysis techniques on blood samples taken from 54 Holstein bulls (male dairy cows used for breeding), the researchers determined the number of copies of TSPY in each bull’s genome. For some of these bulls, tissue samples were also taken from the testes and other organs, and then used to determine the level of activity of the TSPY gene (how much of the RNA was being produced). Bull fertility was measured by the proportion of cows bred to the bull that were pregnant after 56 days. The semen quality was also assessed using records of ejaculate volume, sperm concentration, total sperm count, and sperm motility, averaged over the bull’s breeding history.

**What you need to know:**

In Holstein bulls, higher fertility was linked to having more copies of TSPY (testis-specific protein found on the Y chromosome) in the genome. Bulls with more TSPY copies showed decreased TSPY gene activity and no change in semen characteristics, however, suggesting that copy number indirectly affects fertility.

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What did the researchers find?
TSPY gene activity varied a great deal among bulls, but was limited to the testes in all cases. Overall, TSPY copy numbers in this study ranged from 61 to 281 copies per genome, with an average of 151 copies in the first experiment and 182 in the second. In both experiments, higher copy numbers were associated with greater fertility but, surprisingly, lower TSPY gene activity in the testes. There was no link between TSPY copy number and any of the semen characteristics studied.

How can you use this research?
Cattle breeders and dairy producers can use this research to better understand how bull fertility may be affected by the number of copies of a testis-specific protein in their genome.

Animal and veterinary scientists can further this research by investigating how TSPY copy number influences fertility without affecting gene activity or semen characteristics like sperm count or sperm motility. They can also further this research by identifying if infertility in bulls is associated with abnormally low TSPY copy numbers.

Keywords:
Cows, dairy bulls, field fertility, semen, copy number variation, testis-specific protein y-encoded

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