ABSTRACT
In dairy cattle, the widespread use of artificial insemination has resulted in increased selection intensity, which has led to spectacular increase in productivity. However, cow fertility has concomitantly severely declined. It is generally assumed that this reduction is primarily due to the negative energy balance of high-producing cows at the peak of lactation. We herein describe the fine-mapping of a major fertility QTL in Nordic Red cattle, and identify a 660-kb deletion encompassing four genes as the causative variant. We show that the deletion is a recessive embryonically lethal mutation. This probably results from the loss of RNASEH2B, which is known to cause embryonic death in mice. Despite its dramatic effect on fertility, 13%, 23% and 32% of the animals carry the deletion in Danish, Swedish and Finnish Red Cattle, respectively. To explain this, we searched for favorable effects on other traits and found that the deletion has strong positive effects on milk yield. This study demonstrates that embryonic lethal mutations account for a non-negligible fraction of the decline in fertility of domestic cattle, and that associated positive effects on milk yield may account for part of the negative genetic correlation. Our study adds to the evidence that structural variants contribute to animal phenotypic variation, and that balancing selection might be more common in livestock species than previously appreciated.