Title: Exploring the intersection between structural variants and functional regulatory elements in pigs

Tittel: Undersøke sammenhengen mellom strukturelle varianter og funksjonelle regulatoriske elementer i gris

Keywords/Stikkord: genomics, breeding, structural variation, genome diversity

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Description:
Structural variants (SVs) are an essential source of genome diversity and can, because of their size (>50 bp), contribute substantially to phenotypic diversity and disease. SVs do not only disrupt protein-coding genes, but they can also disrupt the expression and regulation of these genes. The impact of SVs on gene regulatory elements in pigs is not well studied, and the aim of this thesis is to use structural variants detected from long-read sequencing data in pigs to characterize overlap with functional regulatory elements. A catalogue of SVs in Norwegian pigs is currently being generated by the Causative project led from BIOVIT. At the same time regulatory elements are being catalogued in different tissues by an ongoing EU project, Gene-Switch. By overlapping SVs with the identified regulatory elements, the work of this thesis can identify potential disruptors that may contribute to disease or other traits. The work will also contribute to the understanding of gene regulation, as SVs might bring regulatory elements closer to or further from target genes, thereby altering gene expression.

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