

Identifying reciprocal translocations in pigs through LD analysis of offspring genotypes

Anne Boshove*^{1,2}, Martijn F.L. Derks^{1,2}

¹Topigs Norsvin Research Center, 's-Hertogenbosch, The Netherlands

²Wageningen University & Research, Animal Breeding and Genomics, Wageningen, The Netherlands

Reciprocal translocations (RTs) are chromosomal rearrangements where DNA segments are exchanged between two non-homologous chromosomes. RTs are one of the most common structural chromosomal abnormalities in humans and are also commonly observed in livestock, with an estimated prevalence of 0.47% in pigs. Due to no genetic information being lost, individuals carrying a RT are often not directly affected, unless the RTs breakpoints truncate an (important) gene. However, RTs can lead to reduced reproductive performance, in pigs often observed through a reduction in litter size. Consequently, RTs can lead to severe economic losses especially in the case of influential boars. A recent study showed they were able to successfully detect RTs using offspring genotype data to search for abnormal linkage disequilibrium (LD) patterns (Jourdain et al., 2023). If an RT-carrier produces offspring, approximately half of them will inherit a balanced copy of the RT, increasing the LD between the two chromosomes involved in the RT. In this study we developed a method based on this same principle to identify RTs in pigs using only offspring genotypes. Our dataset consisted of offspring genotypes (50K) from seven boars carrying an RT, previously identified by karyotyping. Additionally, we performed Nanopore long read sequencing on one of the RT-positive boars, allowing us to identify the exact breakpoints of the RT. We used PLINK to perform extensive LD analysis on the offspring populations and compared the results to reference values of offspring populations from 100 RT-negative boars. We found that we can confidently detect RTs when at least 30 genotyped offspring are available. In addition to developing this method, we inspected the impact of RTs on reproductive performance using data of 16 RT-positive boars with a total of 320 litters, which we compared to 200+ RT-negative animals. We found that each of the 16 RT-positive boars had a reduced litter size between 15% and 42%, with a total average reduction of 35%. To conclude, this study presents a method to screen animals for RTs without the use of cytogenic testing, provided offspring genotype data is available. In addition we highlight the importance of identifying RTs in breeding populations by showing the significant impact they have on fertility.