



Figure 5-figure supplement 3 Example of evidence for a segmental duplication in donor NC17, chromosome 6. (A) For a segment of the chromosome enriched for low uncorrected p-values ($p\text{-value} < 1 \times 10^{-8}$), we plot each hetSNP based on the number of sperm in which it was observed as the reference allele or alternative allele. Points cluster on lines with slope = 0.5 and slope = 2, deviating from the null expectation of clustering on slope = 1 (equal representation of the alternative and reference alleles across the pool of sperm). (B) To compare, we consider SNPs across the rest of the chromosome, outside of this region. Points cluster to the line slope = 1. (C) Across the whole chromosome, we consider the number of sperm cells in which each SNP is observed, based on whether their uncorrected p-value was greater or less than 1×10^{-8} .