

Online Resource 1

Quality control of the Project MinE samples and variants

Samples included in Project MinE underwent whole-genome sequencing, genotyping on the Illumina 2.5M, and methylation data collection. Samples underwent QC based on the whole-genome sequencing data.

Samples were checked for the following metrics (with exclusion criteria in parentheses): total depth (no exclusion necessary), transition/transversion ratio (>2.09), concordance with genotype data ($<95\%$), phenotype/genotype sex concordance (discordant samples are removed), overall missingness ($>5\%$), inbreeding (>3 standard deviations from the distribution), relatedness (duplicates are removed; related samples with kinship > 0.0625 are dropped for analyses requiring independent samples), total single nucleotide variants ($> 4M$), total indels ($>900k$), total singletons (>6 standard deviations from cohort distribution), and ratio of heterozygotes to homozygous non-reference genotypes (>3).

Variants are checked for the following metrics (with exclusion criteria in parentheses): depth of coverage ($<1.5x/sample$ or $>34.8x/sample$), differential missingness ($p < 10^{-3}$), Hardy-Weinberg equilibrium violations ($p < 10^{-3}$), missingness ($>5\%$), and overall "PASSing rate" (according to Illumina genotype calling pipeline which is performed on a sample-level basis, $<70\%$ considering all samples).