The TransplantLines Kidney eQTL Resource

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INTRODUCTION AND AIMS: TransplantLines is a Dutch biobank collecting data on organ transplantation. It contains a unique resource: a large number of healthy kidney biopsies of which both genome-wide genotype and expression (Illumina HumanHT-12 v4) measurements are available.

METHODS: The dataset includes both living (N=34) and deceased donors (N=100). In a subset of samples, we also have repeated expression measurements from different time-points: before transplantation (N=134), before (N=17) and after reperfusion (N=49). In total, expression and genotype data are available for 237 kidney biopsies of 134 donors. To our knowledge no larger public healthy kidney eQTL dataset exists.

RESULTS: The TransplantLines Kidney eQTL resource has been used by various studies to follow up on findings of genome-wide association studies. For example, additional to an eQTL analysis in whole-blood datasets, we performed an eQTL analysis of 161 loci associated with blood pressure in the TransplantLines resource. This yielded an association of 9 SNPs with expression levels of 13 genes. Of these, 4 signals were unique to the kidney, and 3 had not been previously reported (Wain et al., 2017). We also performed eQTL analyses for genetic markers associated with kidney function. We looked at 64 markers associated with estimated glomerular filtration rate by the CKDgen consortium (Gorksi et al., 2017) and identified 13 kidney eQTLs related to 11 SNPs (unpublished data). Furthermore, we analysed 29 SNPs associated with serum urea, and found three to be eQTLs (manuscript in preparation).

CONCLUSIONS: We recently performed genotyping in 52 additional samples. Both old and new samples will be re-imputed using data of the Haplotype Reference Consortium (HRC). To date, the TransplantLines Kidney eQTL resource has only been used for cir-eQTL look-ups. However, our main goal is to perform a genome-wide eQTL analysis using the HRC-imputed genotypes of the expanded sample.