

Allele Specific Expression analysis and visualization in a unique dilated cardiomyopathy cohort

Daan van Beek¹, Job Verdonschot², Kasper Derks², Han Brunner², Theo de Kok^{1,3}, Ilja Arts¹, Stephane Heymans⁴, Martina Kutmon^{1,5}, Michiel Adriaens¹

Background

Allele specific expression offers a way to **use RNA-seq data** to look at **cis-regulatory variation** and study regulatory genetics in complex genetic phenotypes such as **dilated cardiomyopathy**. Causes of variation include upstream promotor and enhancer mutations, alternative splicing, and RNA silencing.

Here, we present a pipeline that offers:

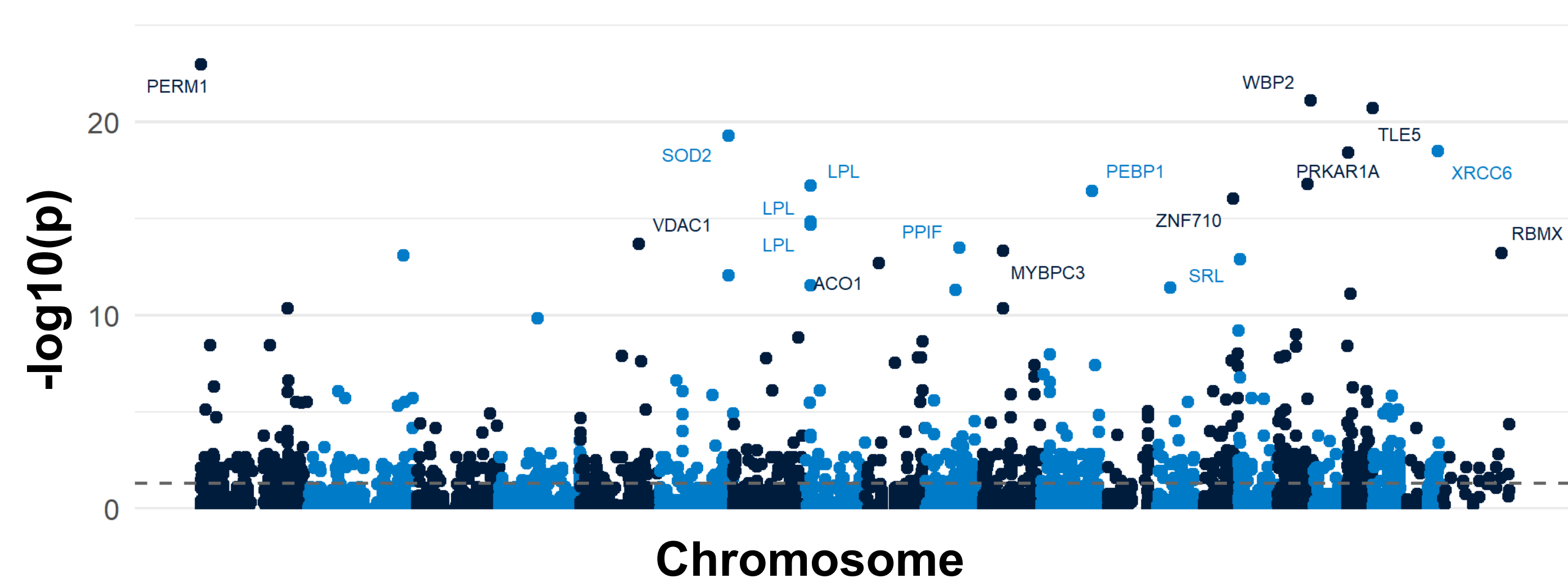
- Easy allele specific expression analysis
- Individual sample results
- Group comparisons
- Intuitive visualizations

Allele specific expression (ASE)

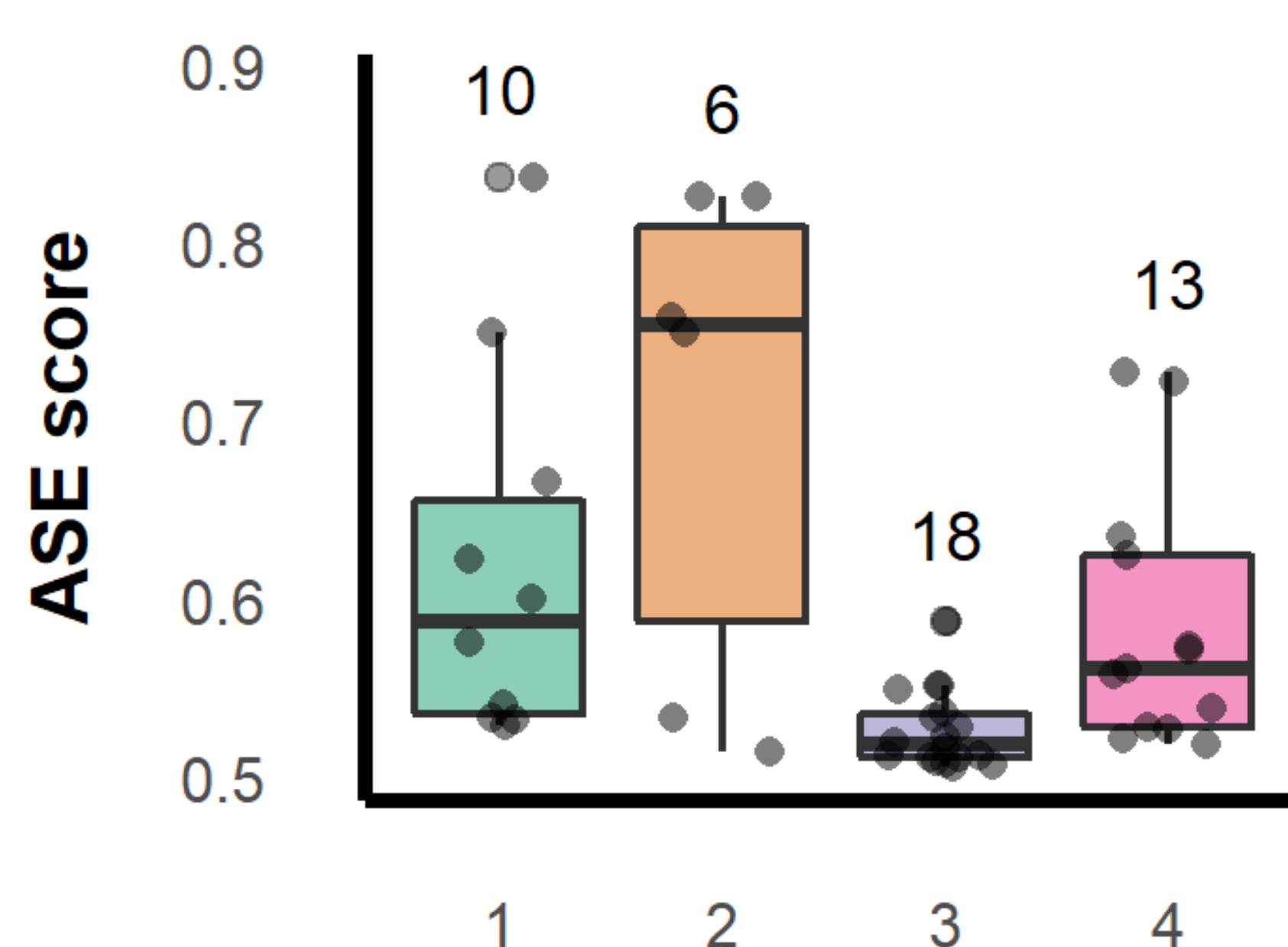
Expression of alleles on the same locus



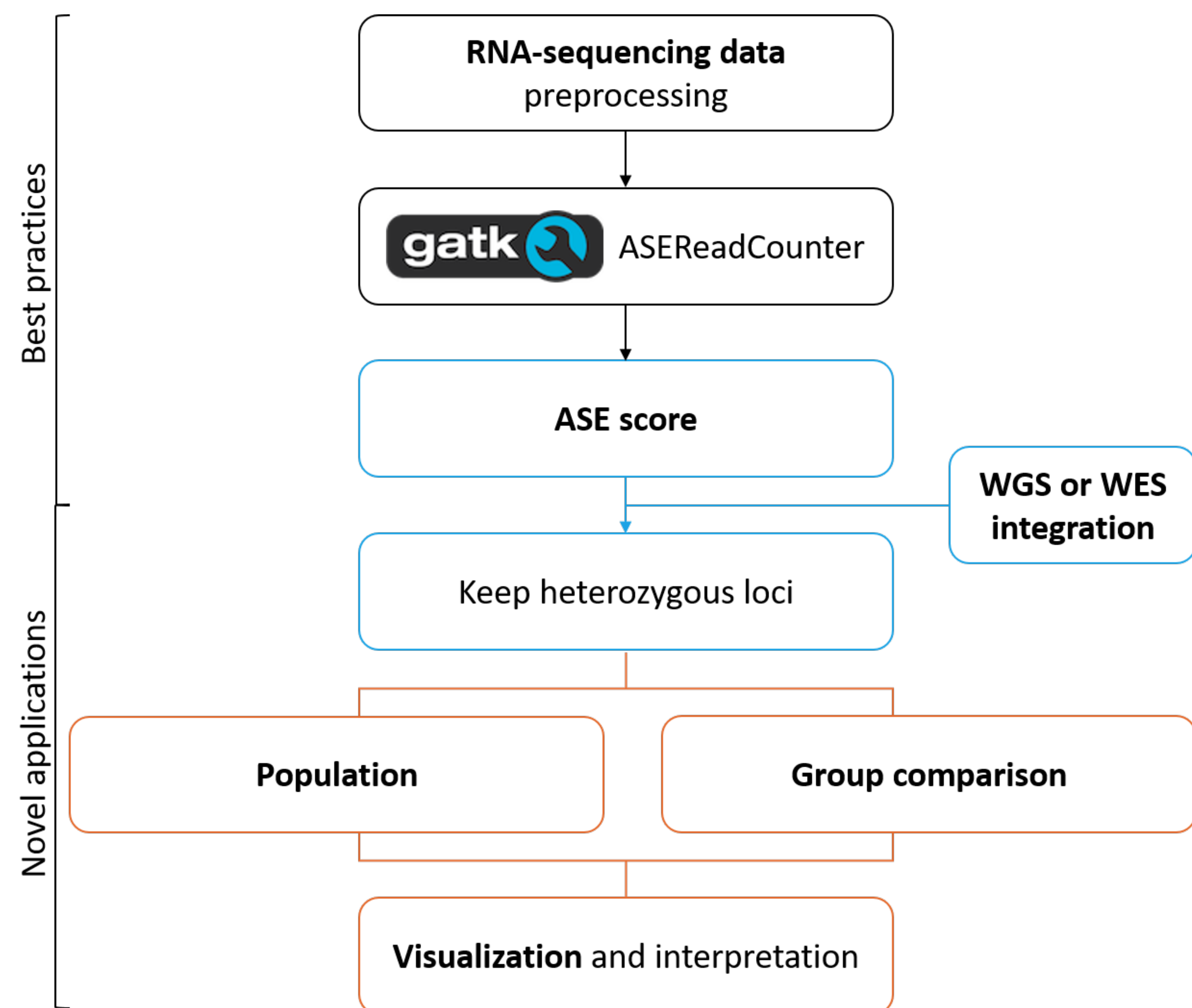
Individual ASE profiles



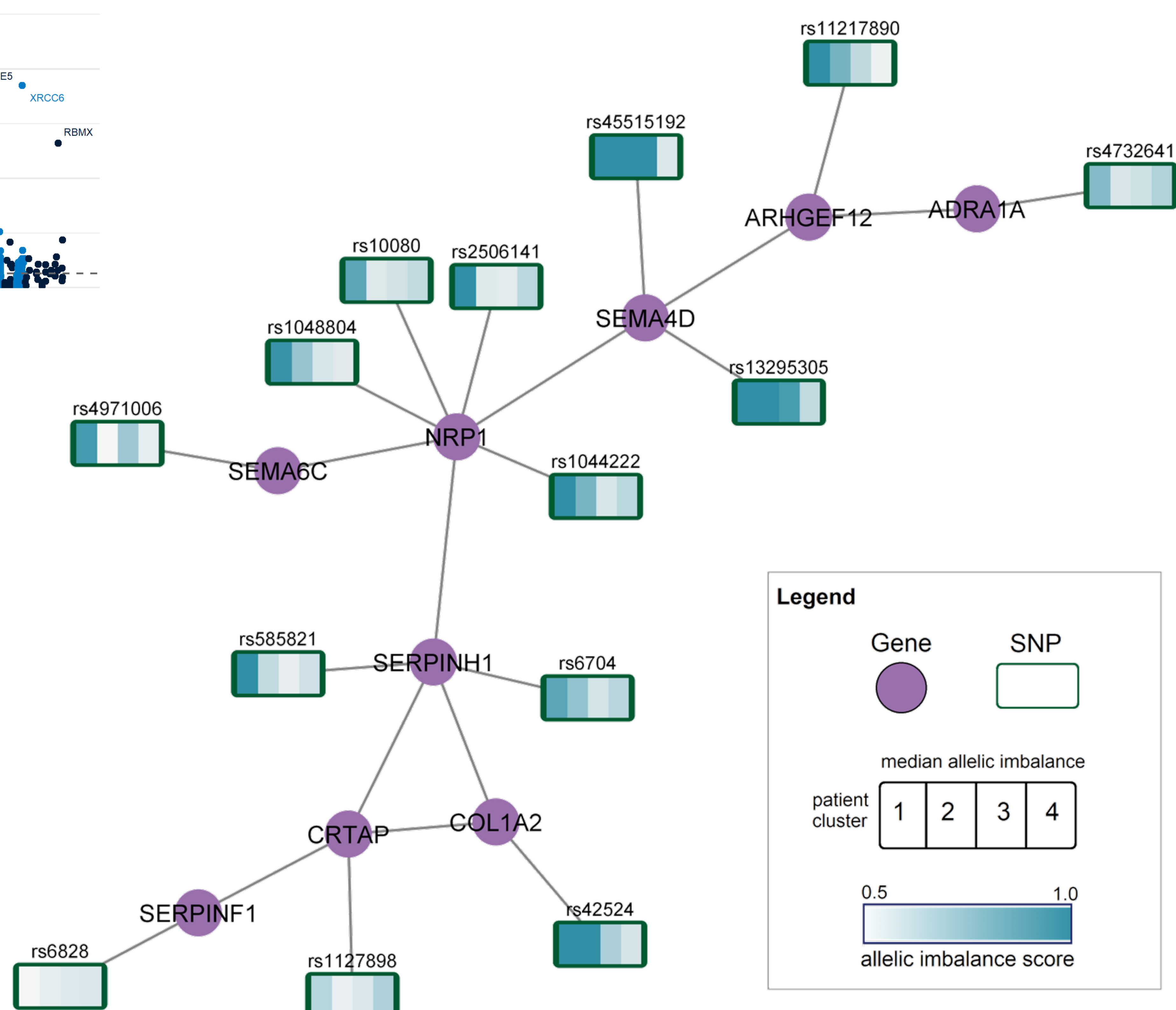
Group comparisons



Pipeline



Functional analysis



¹ Maastricht Centre for Systems Biology – MaCSBio, Maastricht University – UM, Maastricht, 6229 EN, Netherlands

² Department of Clinical Genetics, Maastricht University Medical Centre – MUMC+, UM, Maastricht, 6229 ER, Netherlands

³ Department of Toxicogenomics – TGX, UM, Maastricht, 6229 ER, Netherlands

⁴ Department of Cardiology, School for Cardiovascular Diseases, Cardiovascular Research Institute Maastricht - CARIM, MUMC+, UM, Maastricht, 6229 ER, Netherlands

⁵ Department of Bioinformatics – BiGCaT, School of Nutrition Toxicology and Metabolism – NUTRIM, MUMC+, UM, Maastricht, 6229 ER, Netherlands