

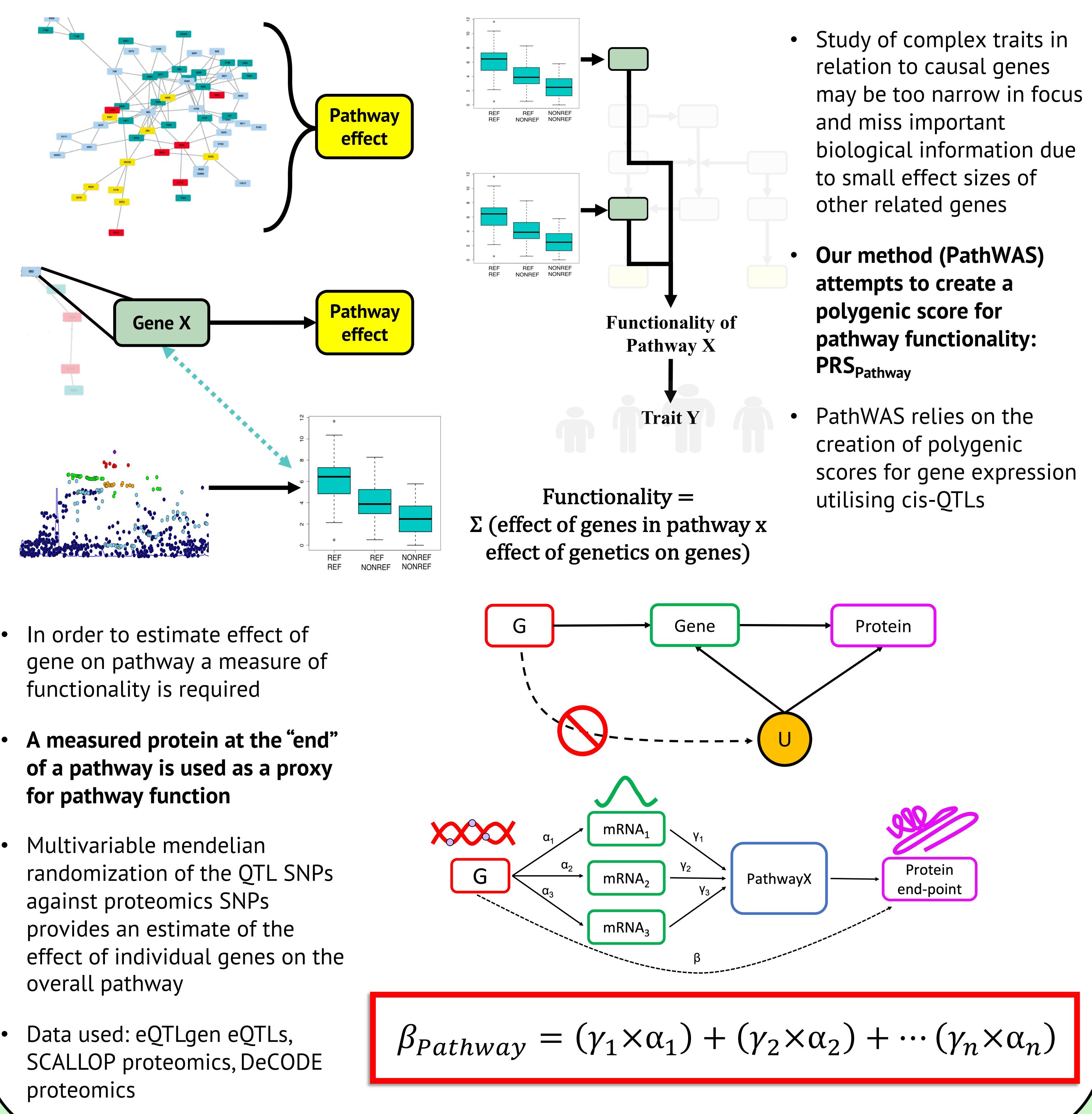
# PathWAS reveals biologically relevant pathways in complex traits



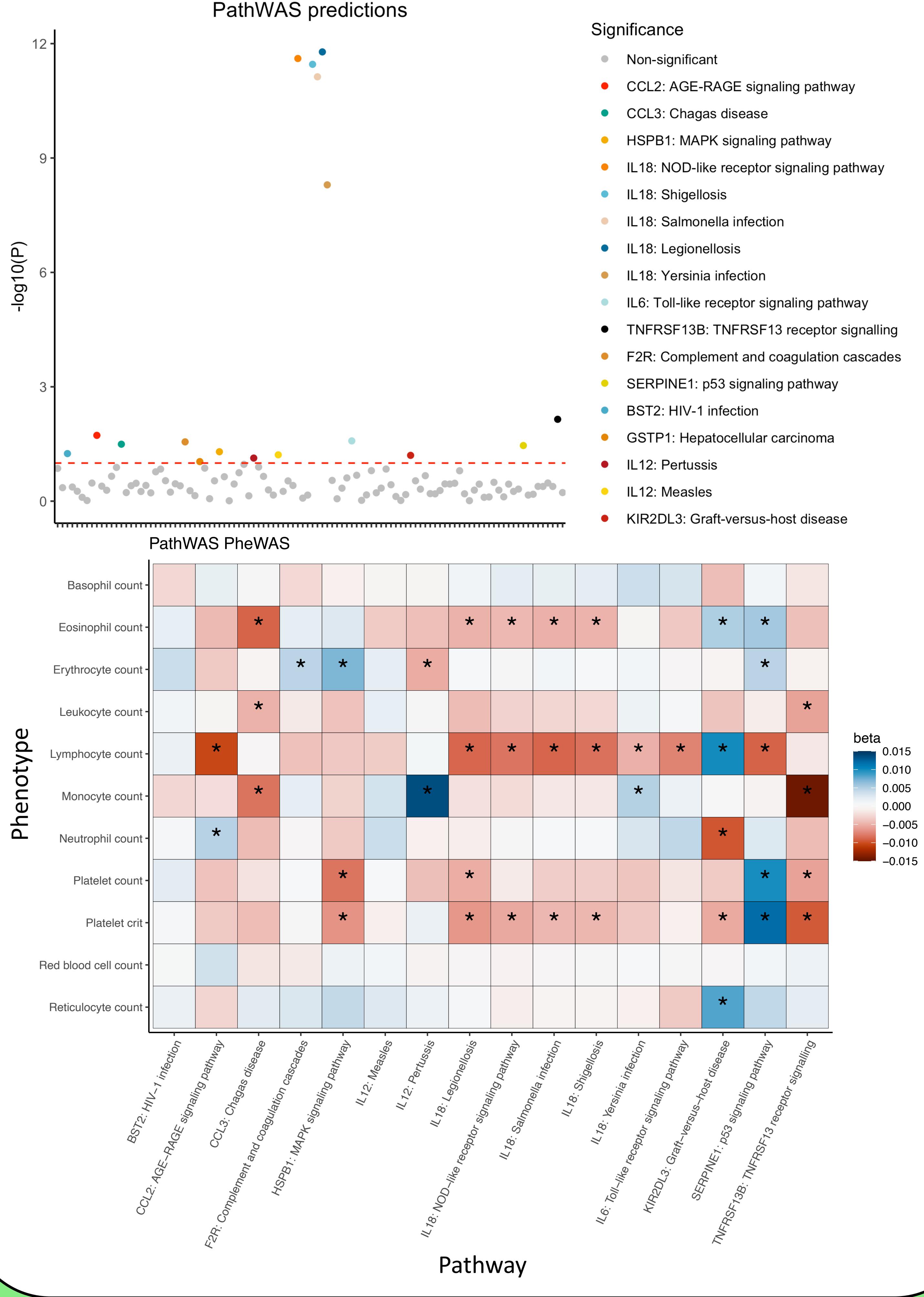
THE UNIVERSITY  
of EDINBURGH

MRC Medical Research Council  
Usher Institute

## 1. PathWAS rationale and methodology



## 2. SCALLOP PathWAS and proof of concept



## 3. DeCODE PathWAS finds hundreds of associations

- 4907 proteins in DeCODE subjected to PathWAS analysis

- 666 pathways tested against 60 phenotypes in UK Biobank

- 2515 significant results acquired and subjected to sensitivity analysis

- 393 significant pathway-trait associations discovered displayed here:

- Sensitivity analysis (Leave-1-Out) and Z-scoring used to exclude results which were driven by individual genes or end-point protein

## 5. Refined DeCODE results

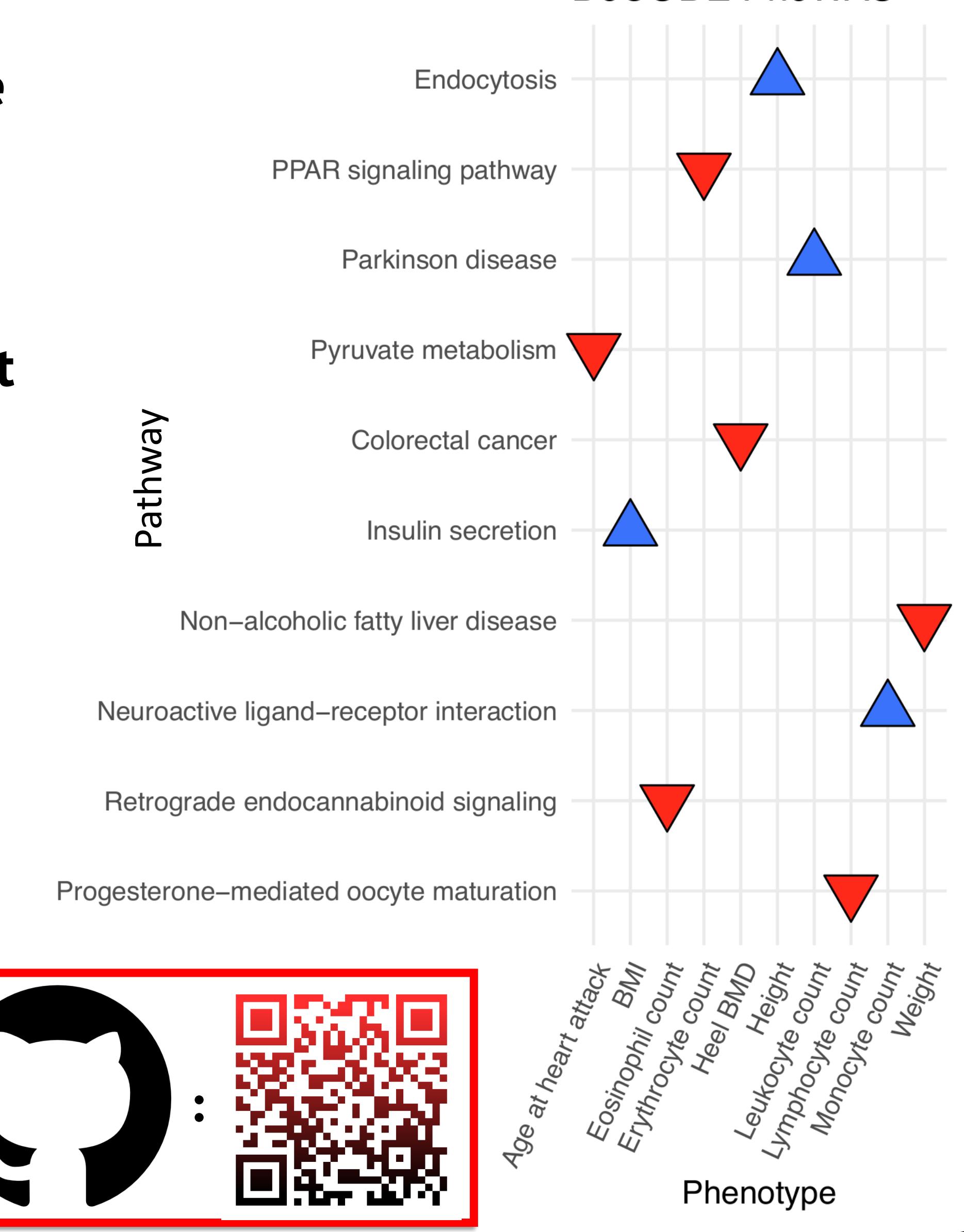
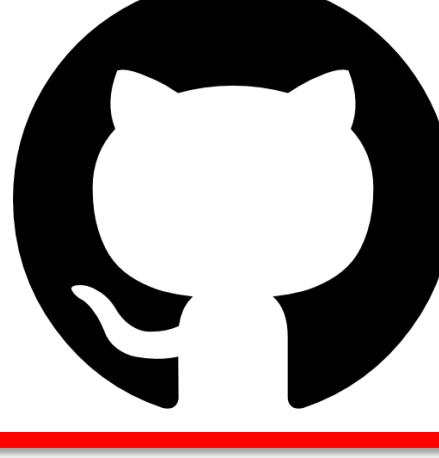
- Many results validated in literature

- Pyruvate degradation to lactate leads to increased risk of heart attack at earlier age

- Parkinson's disease pathway increases leukocyte count

- Insulin secretion increases BMI

PathWAS Package



Sebastian May-Wilson<sup>1</sup>, Erin Macdonald-Dunlop<sup>1</sup>, Linda Repetto<sup>1</sup>, SCALLOP Consortium, James F Wilson<sup>1,2</sup>, Nicola Pirastu<sup>1</sup>



@SebastianMayWi1

@ErinMcD46

@lindarepetto

@Npirastu

s.may-wilson@ed.ac.uk

