

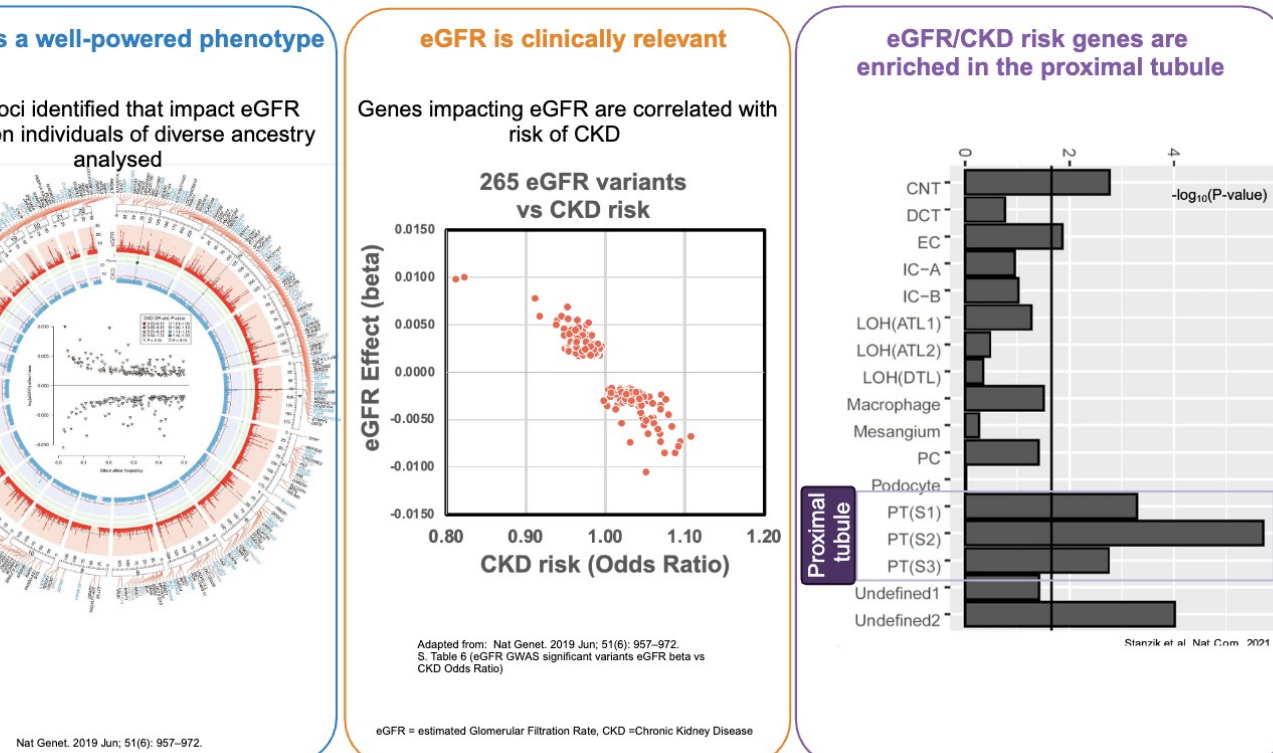
SLC6A19 (B⁰AT1) allelic series: loss of function is associated with improved kidney function

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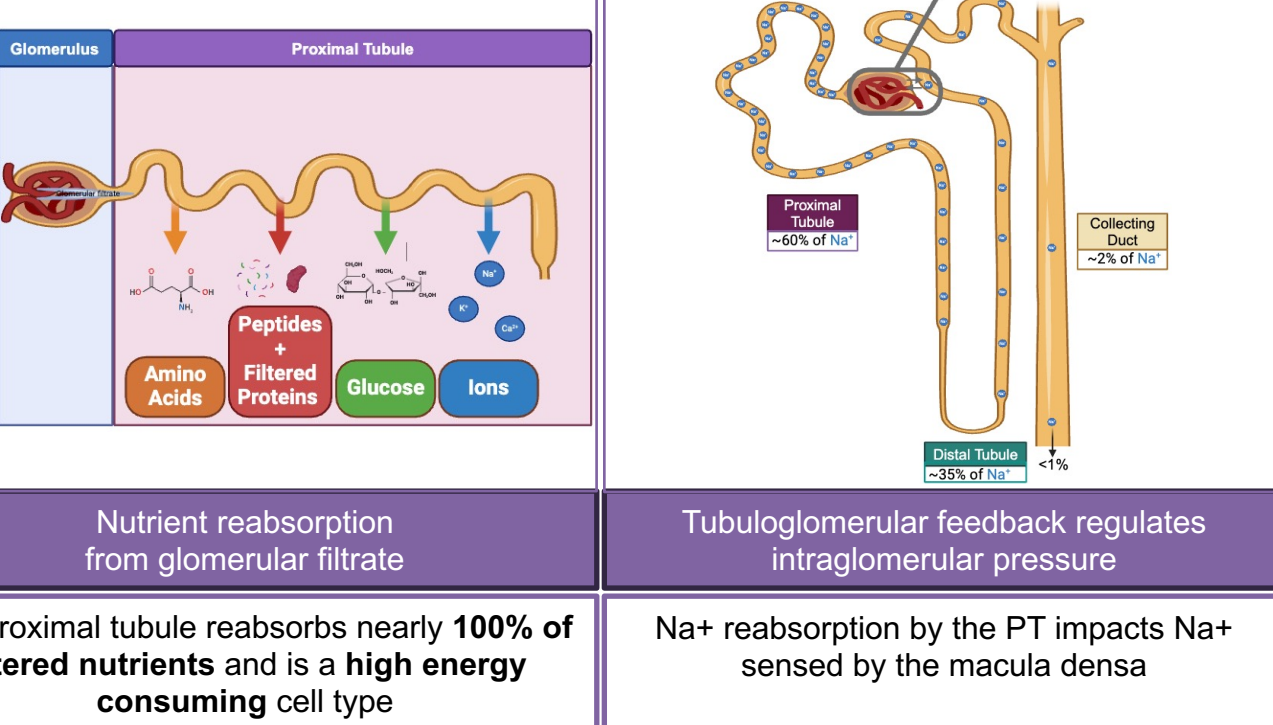
Maze Therapeutics, South San Francisco, CA

Background

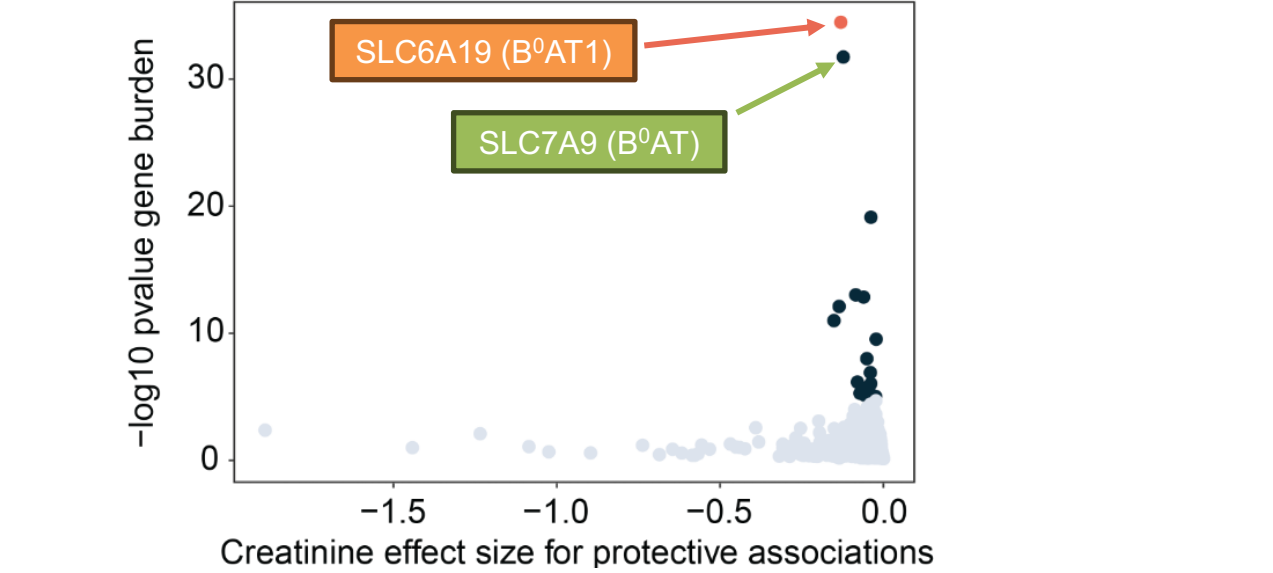
Chronic Kidney Disease (CKD) risk genes are enriched in the kidney proximal tubule



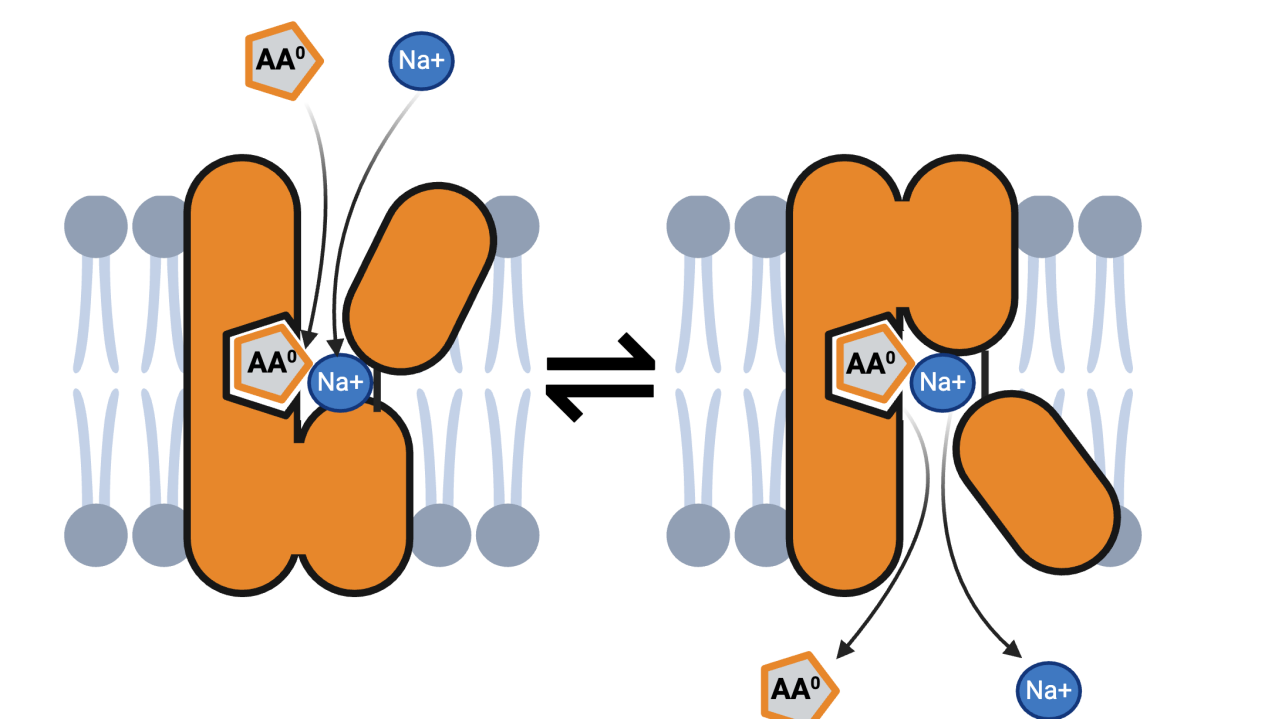
The proximal tubule has a role in nutrient reabsorption and regulation of intraglomerular pressure



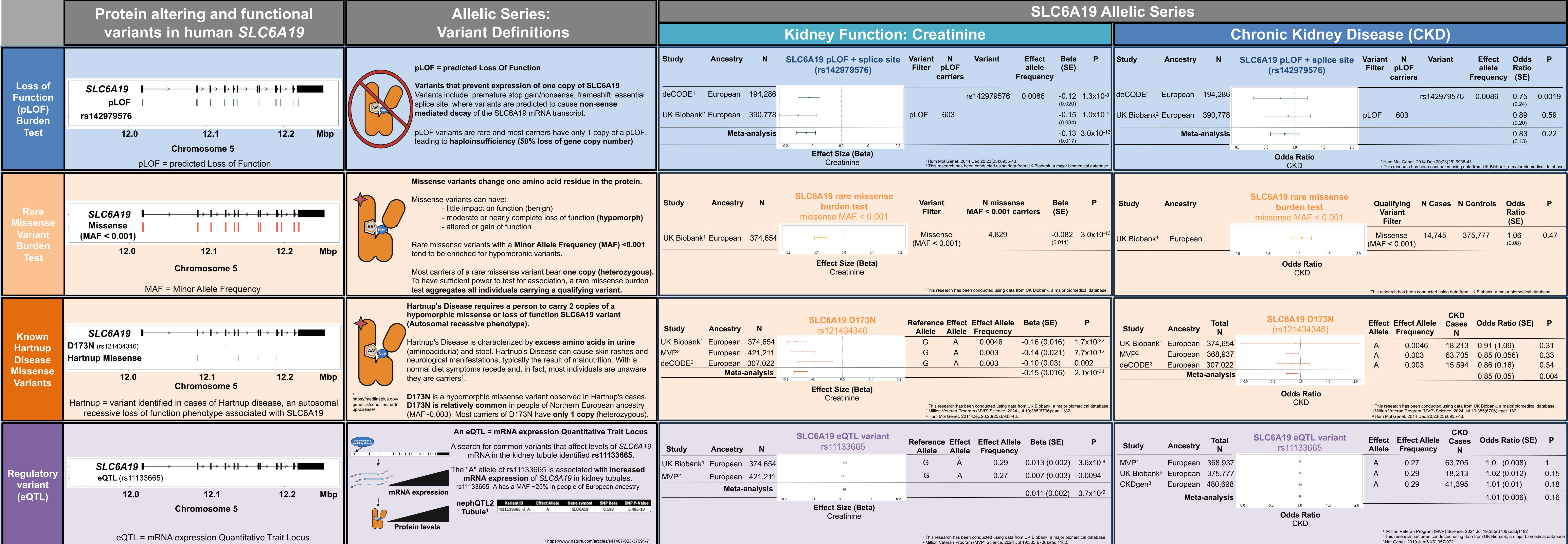
Loss of apical proximal tubule amino acid transporters associates with improved kidney function



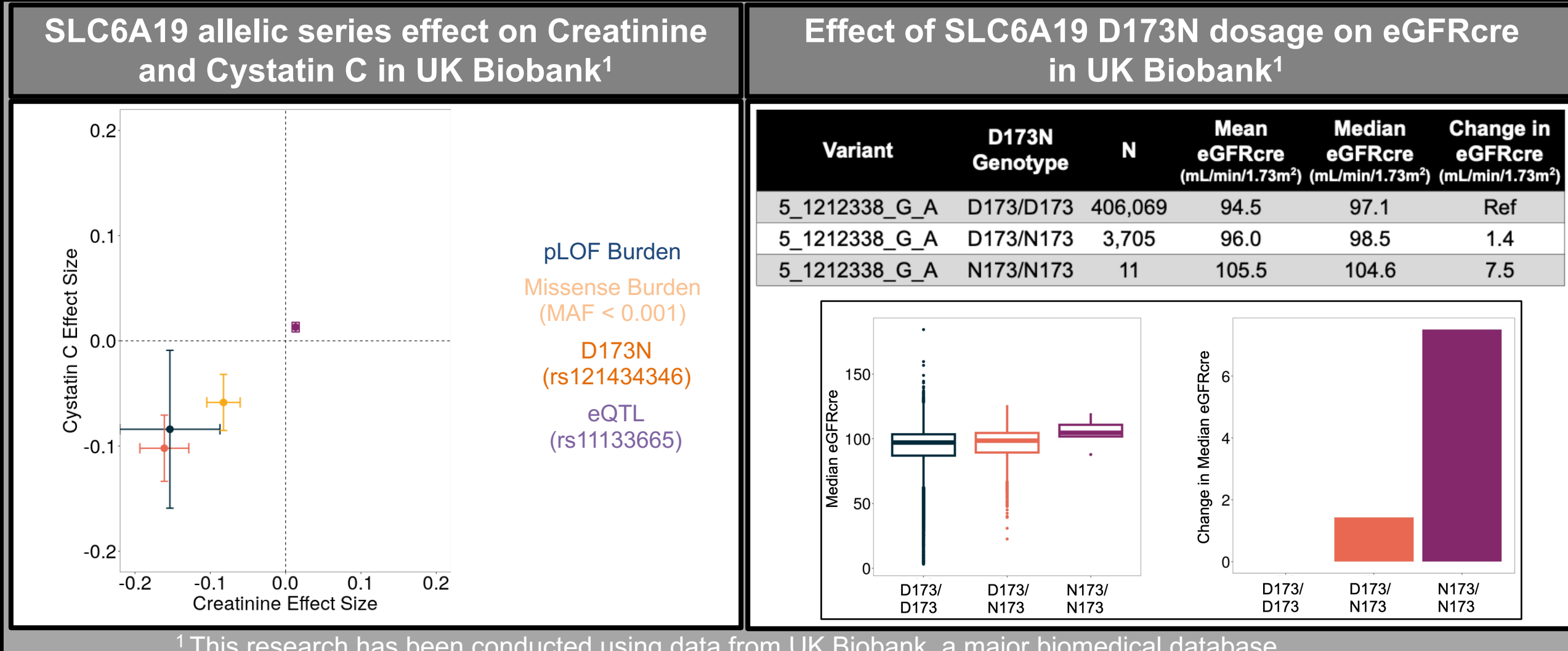
SLC6A19 is a sodium dependent amino acid transporter that reabsorbs nutrients from the diet (intestine) and filtered blood (kidney)



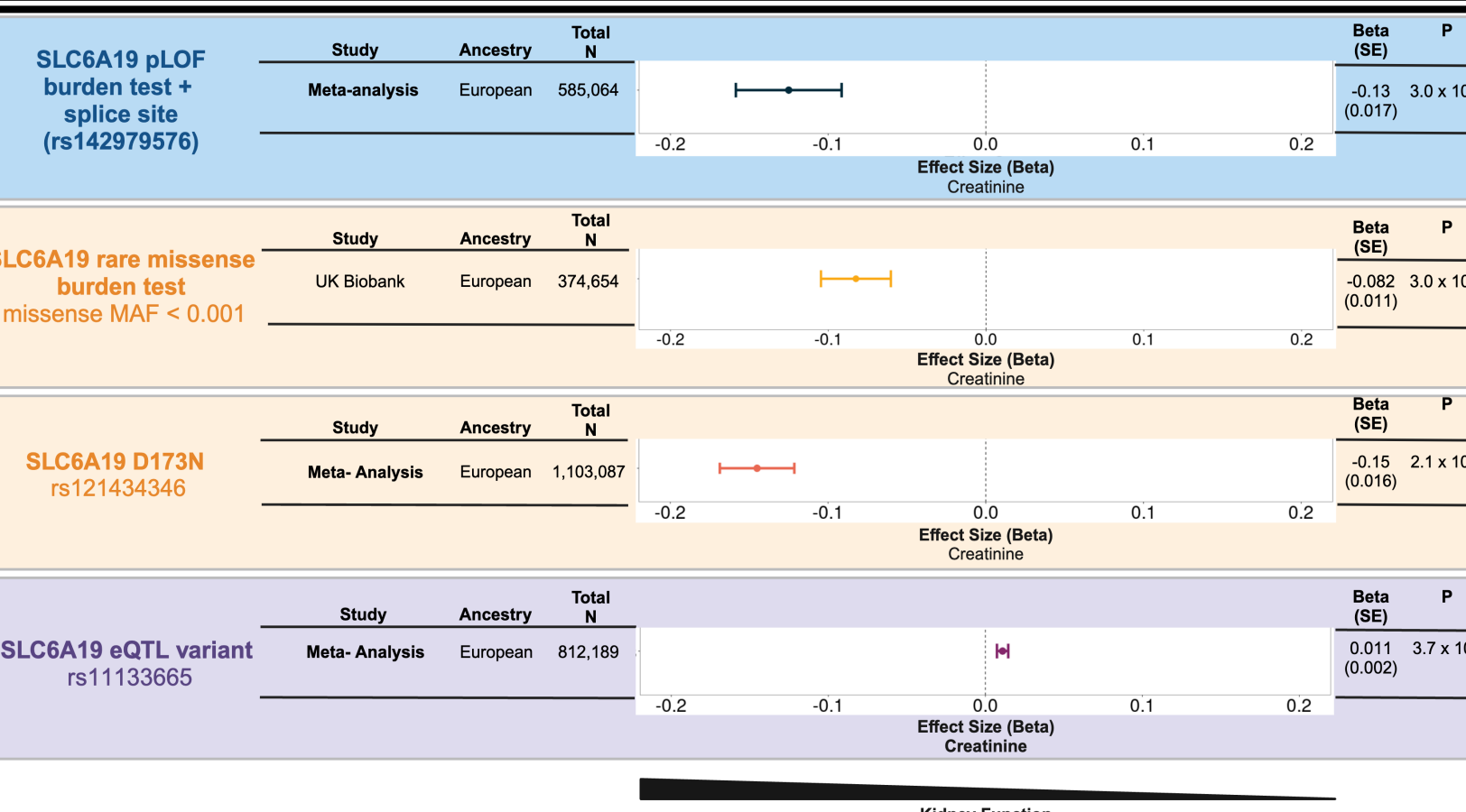
Results: An allelic series of SLC6A19 functional variants is associated with kidney function



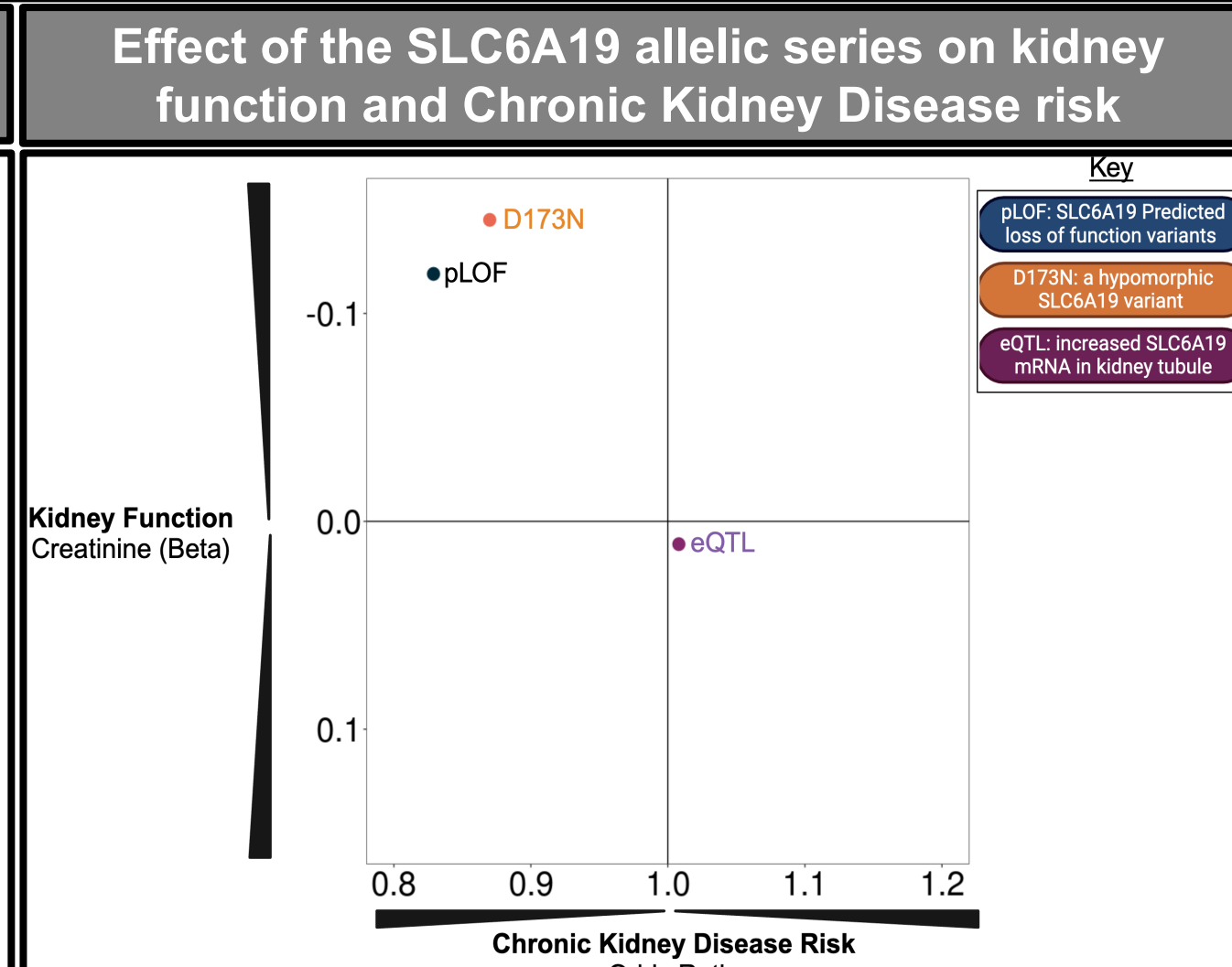
Results: Creatinine/Cystatin C and D173N dosage



A bi-directional allelic series of SLC6A19 loss of function, protein altering and regulatory variants impacts kidney function



Summary



SLC6A19 loss of function: Potential Mechanisms for Renoprotection

