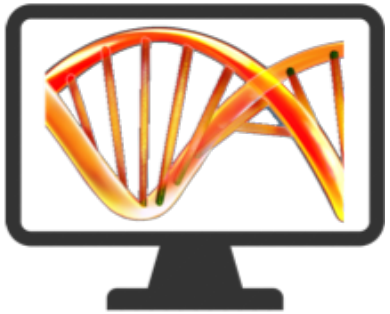
Navigating the ENCODE Encyclopedia: Exploring Candidate Regulatory Elements, Linked Genes, and Genetic Variation with SCREEN



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Part IV – Use Cases

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Use Case #1 - Investigating Gene Expression

RESOURCE

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nature neuroscience

Whole genome sequencing resource identifies 18 new candidate genes for autism spectrum disorder

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MYO5A

- one of three myosin V heavy-chain genes, belonging to the myosin gene superfamily.
- Myosin V is a class of actin-based motor proteins involved in cytoplasmic vesicle transport and anchorage, spindle-pole alignment and mRNA translocation.
- MYO5A is abundant in melanocytes and nerve cells.

Use Case #2 – Annotating GWAS Variants

nature

ARTICLES

Biological, clinical and population relevance of 95 loci for blood lipids

A list of authors and their affiliations appears at the end of the paper.

Plasma concentrations of total cholesterol, low-density lipoprotein cholesterol, high-density lipoprotein cholesterol and triglycerides are among the most important risk factors for coronary artery disease (CAD) and are targets for therapeutic intervention. We screened the genome for common variants associated with plasma lipids in >100,000 individuals of European ancestry. Here we report 95 significantly associated loci ($P < 5 \times 10^{-8}$), with 59 showing genome-wide significant association with lipid traits for the first time. The newly reported associations include single nucleotide polymorphisms (SNPs) near known lipid regulators (for example, *CYP7A1*, *NPC1L1* and *SCARB1*) as well as in scores of loci not previously implicated in lipoprotein metabolism. The 95 loci contribute not only to normal variation in lipid traits but also to extreme lipid phenotypes and have an impact on lipid traits in three non-European populations (East Asians, South Asians and African Americans). Our results identify several novel loci associated with plasma lipids that are also associated with CAD. Finally, we validated three of the novel genes—*GALNT2*, *PPP1R3B* and *TTC39B*—with experiments in mouse models. Taken together, our findings provide the foundation to develop a broader biological understanding of lipoprotein metabolism and to identify new therapeutic opportunities for the prevention of CAD.

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Disclosure for:

ASHG Interactive Workshop: Navigating the ENCODE Encyclopedia: Exploring Candidate Regulatory Elements, Linked Genes, and Genetic Variation with SCREEN

No Relevant Conflicts to Disclose:

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