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Summary

THE IMMUNE CIS- REGULOME: WHOLE BLOOD CELL CULTURE - DERIVED RESPONSE EQTLS REVEALS POTENTIAL CAUSATIVE GENES IN RISK LOCI ASSOCIATED WITH INFLAMMATORY BOWEL DISEASES

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UEG Week Vienna 2024 | Presentation

IBD | Immunology

October 13, 2024

Immune stimulation-responsive eQTLs in whole blood from 406 healthy individuals enabled colocalization with 11 additional IBD risk loci that would have been missed without stimulation, expanding candidate gene identification.

KEY POINTS

- TLR and TCR stimulation of whole blood from 406 healthy individuals (248 females, 159 males) identified 13,679 cis-eQTLs corresponding to 6,496 eGenes, with 4,695 cis-regulatory modules requiring stimulation to be detected.
- Colocalization analysis yielded 187 correlations spanning 39 IBD risk loci and 68 genes, with 19.2% of correlations associated with response-QTLs that facilitated identification of 11 risk loci overlooked without stimulation.
- The long non-coding gene AJ009632.2 showed opposite correlation patterns in resting versus stimulated conditions, demonstrating that regulatory variants may appear positively or negatively associated with disease depending on immune context.
- CTSS, encoding cathepsin S involved in antigen presentation and extracellular matrix degradation, was identified through reQTL matching to rs4845604 with correlations of -0.72 (p=0.004) under TLR4 stimulation.
- Additional candidate genes identified through response-QTLs include HORMAD1, ZBTB38, and TPPP, with stimulation-dependent correlations ranging from 0.73 to 0.91 across different IBD phenotypes.

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