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Expression Quantitative Trait Loci for *ALMS1* and Their Influence on the Symptoms of Alström Syndrome

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ABSTRACT Genome-wide expression quantitative trait loci (eQTL) were identified for *ALMS1*, which is known as a causal gene for Alström syndrome. Genetic associations of nucleotide variants were examined with expression of the gene encoding *ALMS1* in lymphoblastoid cell lines derived from 373 Europeans. The analyses revealed six cis-regulatory eQTLs for *ALMS1* ($P < 5 \times 10^{-8}$), showing a strong signal ($P < 10^{-20}$) upstream of the gene. Transcriptome-wide association analysis with these eQTLs revealed that 16 genes shared eQTLs with *ALMS1* ($P < 1.02 \times 10^{-4}$). These genes included MOCS3, CDH3, C1orf100, CXCL14, CDK4, and PCBD1, which are involved in the symptoms of Alström syndrome. This study suggested six novel eQTLs for *ALMS1* and their associations with 16 additional genes. The candidate genes whose expressions were associated with eQTL for *ALMS1* might directly or indirectly cause symptoms of Alström syndrome. Further studies are required to understand their underlying regulatory mechanisms of the eQTLs to improve our understanding of the function of *ALMS1* and other causal genes for Alström syndrome.