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Deposited research article

Heart Specific Genes Revealed by EST Sampling

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deposited research

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Heart Specific Genes Revealed by EST Sampling

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Running title: Human Heart Specific Genes

Abstract

Background: Cardio-vascular diseases are the first cause of death worldwide, particularly in the developed countries; the identification of genes specifically expressed in the cardiac muscle is thus of major biomedical interest. In this study, we performed a comprehensive analysis of the expression profiles to identify genes over-expressed in the human adult heart using the public Expressed Sequence Tags (ESTs) database. The initial set of genes expressed in the heart was constructed by clustering and assembling ESTs from human adult heart cDNA libraries. Expression profiles were then generated for each of these genes by counting their cognate ESTs in all libraries. Differential expression was assessed by applying to these profiles a previously published statistical procedure.

Results: We identified 35 "cardiac specific" genes significantly over-expressed in the heart, some of them exhibiting significant co-expressions. Some genes had clear functional association with the heart, and others had no previously recognized cardiac function. Of the 35 genes, 32 were mapped back onto the human genome sequence. According to OMIM, 5 genes were previously known as heart disease genes and one gene was located in the locus of a bleeding disorder. The analysis of the core promoter regions of our collection of "cardiac specific" genes provides the first list of putative regulatory elements associated with differential gene expression in the heart.

Conclusion: This study shows that ESTs are still a powerful tool to identify differentially expressed genes: we presented a list of genes specifically expressed in the human heart, one of them being a candidate for a bleeding disorder. In addition, we provided the first set of putative regulatory elements, the combination of which appears correlated with heart-specific gene expression.