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Integrative genomics finds disease genes

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In the Early Edition of the Proceedings of the National Academy of Sciences Mootha *et al.* describe an integrative genomic approach to discover a gene associated with the human cytochrome c oxidase (COX) deficiency, Leigh syndrome, French Canadian type (LSFC). LSFC is one of five autosomal recessive COX deficiencies; the other four are due to defects in genes encoding COX assembly factors. Mootha *et al.* combined experimental data from genomic, transcriptome and proteomic studies to identify the gene causing LSFC. They systematically analysed potential genes in the LSFC candidate region that had been narrowed down to a 2 megabase genomic region on chromosome 2p16-21. Neighborhood analysis of large-scale microarray data identified genes that are co-regulated with mitochondrial genes; this was combined with organelle-specific mass spectrometry proteomic data. Integrating these DNA, mRNA and protein-based data led to the identification of a promising candidate gene, *LRPPRC* (leucine-rich pentatricopeptide repeat-containing protein). Mootha *et al.* found mutations in *LRPPRC* in patients with LSFC, validating their integrative genomic approach to disease-gene discovery.

References

1. *Proceedings of the National Academy of Sciences*, [http://www.pnas.org]

2. Identification of a gene causing human cytochrome *c* oxidase deficiency by integrative genomics, [http://www.pnas.org/cgi/doi/10.1073/pnas.242716699]

3. Leigh syndrome, French Canadian type, [http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=220111]

4. Cytochrome *c* oxidase deficiency.