Omics approaches reveal clues for mitochondrial disease diagnosis and pathogenesis

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Mitochondrial disorders involving the respiratory chain are a large group of different disorders, with onset at any age, symptoms originating from any organ system, with any inheritance pattern, and with a large variety of different molecular backgrounds. During the last two decades, knowledge on the genetic backgrounds has increased, but still many patients remain without a specific diagnosis. Furthermore, little is known of the physiological consequences of the various mutations, to explain tissue specificity and the spectrum of clinical outcomes. Recently developed "omics" approaches offer a powerful tool for both understanding molecular background and developing diagnosis. We have utilised a versatile molecular tool set, including transcriptomics, metabolomics and exome analysis, to study samples of mitochondrial disease patients and of our mouse models to reveal molecular pathways involved in mitochondrial disease. These approaches have enabled us to recognise new mitochondrial disorders as well as identify wide-spread physiological consequences of respiratory chain deficiency to the whole-body metabolism in mice and in human patients. The identified molecular signatures offer practical tools for first-line diagnosis of mitochondrial disorders.