Metabolomics: Towards a Better Understanding of Desmoid Tumors

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Metabolomics involves the study of the low molecular weight complement of cells, tissues, and biological fluids, and is now being broadly used in medical research to reveal biomarkers to detect or stage disease or monitor intervention. By mapping metabolites to biochemical pathways, insights into mechanisms can be gained which can lead to novel target identification for drug discovery. This presentation will be an introduction to metabolomics, and the proposed study design will be discussed for a cell-based metabolomics project in collaboration with the Desmoid Collaboration for a Cure. We have used similar research methods a recent project in Barth Syndrome. NMR and LC-MS were employed to study differences in metabolite profiles in plasma from a cohort of BTHS affected individuals compared to age-matched controls. Overall, a clear distinction between metabolite profiles of individuals with BTHS and controls were determined. The discriminating metabolites were found to be involved along mitochondrial and extra-mitochondrial biochemical pathways including: lipid metabolism, biogenic amine metabolism, amino acid metabolism, tRNA biosynthesis, insulin signaling and cholinergic signaling. Taken together, this data indicates broad cellular lipid dysregulation in Barth Syndrome with wide cellular effects.
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