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Metabolomics: A Pipeline for Biomarker Discovery in Genetic Diseases

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Abstract: Genetic disorders (GD) affect hundreds of millions of lives worldwide, leading to specific molecular perturbations in the metabolic profile within a certain biological matrix. Metabolomic studies use advanced technologies (nuclear magnetic resonance (NMR) and mass spectrometry (MS)) with bioinformatics to identify and quantify a set of small molecules (such as carbohydrates, nucleic acids, amino acids, and lipids) present in a biological system. As metabolites represent the downstream product of gene and protein activity, they most likely reflect the phenotype of an organism at a specific time. Metabolomic studies provide novel insights into the underlying disease pathophysiological mechanisms, evaluate the progress of the disease, and identify unique biomarkers for the prediction of disease and therapeutic outcomes. This chapter highlights the applications of metabolomic techniques for biomarker discovery in GD. It discusses the workflow followed, the methods used for sample analysis and data interpretation, and the major challenges and limitations in applying the metabolomic approach for biomarker discovery in GD. Moreover, the chapter provides an overview of the biomarkers identified in four GD: cystic fibrosis, Down syndrome, sickle cell anemia, and glycogen storage disorders, highlighting the promising role of metabolomics in clinical applications.