

Integrated Proteomic and Lipidomic Studies in the Neurodegenerative Disorder, Niemann-pick Type C

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Abstract: Advances in mass spectrometry technology have enabled large-scale differential analysis of proteins and lipids to investigate altered pathways in human disease. Our laboratory studies Niemann-Pick Disease, type C1 (NPC1) a lysosomal storage disorder with visceral involvement and progressive cerebellar neurodegeneration. Using a combination of differential proteomics, lipidomics and mass spectrometry imaging, we have identified new candidate biomarkers in a mouse model of NPC1. These alterations now provide a basis for evaluation in NPC1 patients and can be used for therapeutic development. Several examples will be presented.