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# Chapter Five - Tandem Mass Spectrometry of Sphingolipids: Applications for Diagnosis of Sphingolipidoses

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### Abstract

In recent years, mass spectrometry (MS) has become the dominant technology in lipidomic analysis. It is widely used in diagnosis and research of lipid metabolism disorders including those characterized by impairment of lysosomal functions and storage of nondegraded“degraded substrates. These rare diseases, which include sphingolipidoses, have severe and often fatal clinical consequences. Modern MS methods have contributed significantly to achieve a definitive diagnosis, which is essential in clinical practice to begin properly targeted patient care.

Here we summarize MS and tandem MS methods used for qualitative and quantitative analysis of sphingolipids (SL) relative to the diagnostic process for sphingolipidoses and studies focusing on alterations in cell functions due to these disorders.

This review covers the following topics:

- “ Overview of the biochemistry of SL under normal and pathological conditions (lysosomal storage disorders, LSD)
- “ Overview of MS and its applications to the analysis of SL: evidence of pathological storage of nondegraded SL in cells and body fluids focused on a laboratory diagnosis of LSD, isoform profiles and deacylated forms of SL as new biomarkers, applications in enzymology and metabolic experiments in living cells using mass-labeled substrates.

Tandem MS is sensitive and robust in determining the composition of sphingolipid classes in various biological materials. Its ability to establish SL metabolomic profiles using MS bench-top analyzers, significantly benefits the first stages of a diagnosis as well as metabolic studies of these disorders. It can thus contribute to a better understanding of the biological significance of SL.



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## Keywords

Sphingolipids; Tandem mass spectrometry; Electrospray ionization; Sphingolipidoses; Lysosomal storage disorders; Deacylated sphingolipids; Enzymology

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