# Pediatric Neurology Part III: Chapter 172. Lysosomal diseases: biochemical pathways and investigations (Handbook of Clinical Neurology)

*Marie T. Vanier*

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This chapter summarizes our current knowledge of lysosomes and lysosomal proteins referring to recent reviews, general schemes for degradation of substrates, and various causes of lysosomal storage diseases (LSDs). It then discusses the main principles for laboratory diagnosis. Initial screening by study of accumulated substrates in urine is helpful for mucopolysaccharidoses and oligosaccharidoses. A majority of LSDs result from the deficient activity of one acid hydrolase (in some diseases, several). Establishment of the diagnosis in this group of disorders is based on the measurement of the particular enzymic activity. Pseudodeficiencies are a possible source of error. For defects in lysosomal membrane transporters such as cystinosin or sialin, study of substrate accumulation in readily available cells/fluids is still the method of choice. Demonstration of a metabolic block in living cells is rarely used today, except for Niemann–Pick C disease. For primary diagnosis of patients, molecular genetic testing is necessary when no functional tests exist (e.g., many ceroid lipofuscinoses, Danon disease) and it is the preferred strategy when functional tests are too elaborate. Genotyping patients already diagnosed by biochemical methods is, however, essential for genetic counseling in the family; it may also be useful for optimal management.

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