

Lysosomal storage diseases (LSDs) are rare inborn errors of metabolism, occurring in 1 in 7,000-8,000 live births. Approximately 50 different LSDs manifest in the neonatal period and are seldom considered when evaluating a sick newborn. A greater awareness of these disorders may help to reduce misdiagnosis and promote the early detection of these disorders.

This presentation will focus on recognizing specific symptoms and clinical findings that present in the perinatal period, including neurologic, respiratory, endocrine, and cardiovascular manifestations.