

Lysosomal storage diseases are inherited metabolic diseases that are characterized by an abnormal build-up of various toxic materials in the body's cells as a result of enzyme deficiencies. There are nearly 50 of these disorders altogether, and they may affect different parts of the body, including the skeleton, brain, skin, heart, and central nervous system. New lysosomal storage disorders continue to be identified. While clinical trials are in progress on possible treatments for some of these diseases, there is currently no approved treatment for many lysosomal storage diseases. As a group, lysosomal storage diseases are believed to have an estimated frequency of about one in every 5,000 live births. Although the individual diseases are rare, the group together affects many people around the world. Some of the diseases have a higher incidence in certain populations. For instance, Gaucher and Tay-Sachs diseases are more prevalent among the Ashkenazi Jewish population. A mutation associated with Hurler syndrome is known to occur more frequently among Scandinavian and Russian peoples. Prenatal diagnosis is possible for all lysosomal storage disorders. Early detection of lysosomal storage diseases, whether before birth or as soon as possible afterward, is important because when therapies are available, either for the disease itself or for associated symptoms, they may significantly limit the long-term course and impact of the disease.