

Metabolic rare diseases are diseases that result from some defect or abnormality in the metabolic process. These diseases are usually caused by genetic factors and affect the body's metabolic processes, such as protein, carbohydrate, and lipid metabolism. Metabolic rare diseases usually lead to serious health problems, including muscle wasting, liver disease, neurological damage, and more. Due to the rarity and complexity of these diseases, sophisticated research tools and techniques are needed to understand their physiopathological mechanisms.

Animal models provide researchers with valuable tools to study disease mechanisms and test potential therapies in a controlled environment. Research in preclinical animal models of LSDs has enabled the development of several therapeutic strategies, including bone marrow transplantation, recombinant enzyme replacement therapy (ERT) using related defective enzymes, substrate deprivation, and gene therapy.