The Mucopolysaccharidosis (MPS) and Mucolipidosis (ML) disorders are inherited diseases with pediatric onset, which are often both severe and fatal in the childhood period. These multi-systemic disease syndromes cause severe developmental and progressive damage to the bones, heart, eyes, respiratory system, and brain.

The Committee continues to urge NIH to put a high priority on better understanding and treating MPS and ML diseases. The Committee commends the NIH for allocating funds to discover, develop, define, and make available for research animal models of human genetic disease. The Committee encourages expanded research of treatments for neurological, chronic inflammation, cardiovascular, and skeletal manifestations of MPS and ML, with an emphasis on gene therapy.

The Committee thanks the NINDS, NIDDK, and NCATS/ORDR for past funding that supported the Lysosomal Disease Network through the Rare Disease Clinical Research Network and for funding lysosomal research meetings. The Committee encourages the NIH, NCATS, NIDDK, NICHD, and NINDS to increase funding to 17 million dollars to grantees specifically addressing mucopolysaccharidosis and mucolipidosis type II and III disorders to incentivize MPS and ML research, particularly given the aging and small population of current researchers. Understanding the manifestations and treatments of both the skeletal and neurological disease continues to be the greatest areas of unmet need.

The social cost of rare diseases, a legislatively defined group of conditions which include the MPS and ML disorders, is considerable, and could be substantially lessened by policy incentives and investments in both drug approval policies and public health measures that target early diagnosis of rare disease of childhood. The burden on families, care givers, and the healthcare delivery infrastructure is estimated at nearly one trillion dollars per year. Of the 12 MPS and ML disorders, six have FDA therapies available, and there is active progress developing therapies for all. Early initiation of therapy fundamentally changes the lives and health trajectories of MPS and ML individuals. Awaiting a clinical diagnosis invariably limits optimal treatment. Improvements in the federal mechanism for evaluating and recommending diseases for newborn screening approval at the federal level, and incentivizing states to fully implement federal level recommendations, will save lives, substantially improve health outcomes, reduce total economic costs, reduce federal health expenditures, preserve family structure, and improve family well being and prosperity.