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Lysosomal disorders are a family of rare metabolic disorders that include Pompe disease and mucopolysaccharidosis type 1 (MPS1). Pompe disease is caused by deficiencies in the lysosomal enzyme alpha-glucosidase (GAA). This diagnosis is generally divided into infantile and late onset, but can exist along a spectrum. The main feature of Pompe disease is progressive muscle weakness, and in the infantile onset form, cardiomyopathy is present. MPS1 results from a deficiency in the lysosomal enzyme alpha-L-iduronidase (IDUA). Features include progressive neurological disease, skeletal abnormalities, cardiac disease, corneal clouding, hearing loss and hepatomegaly. The diagnosis has been stratified into MPS1 or attenuated MPS1, and severity or development of symptoms is dependent on the type. Both Pompe disease and MPS1 were recently added to the newborn screening panel. With their addition, a number of challenges have occurred. The purpose of this study was to examine the experience and perceptions of parents of an infant who had a positive newborn screening result for Pompe disease or MPS1. A survey was distributed via email and mailer, to parents followed by the UPMC Children’s Hospital of Pittsburgh and the Children’s Hospital of Philadelphia. The survey consisted of questions specific to newborn screening of lysosomal diseases. Parent responses, were consistent with the literature, indicating that this period was categorized by stress and uncertainty. Of the five respondents, 100% described initial diagnostic of the newborn screening results as difficult. Responses suggest that negative emotions were fueled by lack of information provided about the results, and provider’s lack of knowledge. Similar to published literature, this study indicated that the majority of infants are not diagnosed with the most severe forms of the disorders. Results of this study may have implications for how genetic counselors care for these families, how they communicate with other providers, and standard of care.

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134 Impact of SARS-CoV-2 on patients with lysosomal diseases in a major NYC hospital system

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During the height of the pandemic in NYC (March–June 2020), the NYU Langone Health Lysosomal Storage Disorders (LSD) Program reached out to 183 patients to provide information on how to mitigate exposure to COVID19 and to ascertain who had been exposed and/or infected. 139 patients were successfully contacted. Recommendations on how to safely continue enzyme replacement therapy (ERT) or substrate reduction therapy (SRT) were provided. 135 of the 139 respondents during March 2020–June 2020 had Gaucher disease (GD). Twenty-six patients with GD endorsed 2 or more symptoms consistent with COVID19 infection and/or were confirmed to have COVID19 either through RT-PCR test for SARS-CoV-2 RNA or through antibodies to the virus. The remaining 4 who had suspected or confirmed COVID19 infection were patients with Fabry (2), Pompe (1), and Mucopolysaccharidosis Type IIIA (1). This case series describes the impact of COVID19 on 30 patients with LSDs with details of symptomatology, duration of illness, and treatment. Baseline demographics were collected including age, sex, genotype, current disease burden, LSD treatment history, biomarkers and co-morbidities. At time of infection, 21 patients were on ERT (20 GD, 1 PD), 3 on SRT for GD, and 6 were naive to therapy. There was only 1 hospitalization of a 55 year old woman with GD on ERT that resulted in ARDS who subsequently died due to SARS-CoV-2. Her co-morbidities included morbid obesity, COPD, hypertension and diabetes. Her GD burden was minimal. The rest of the affected patients had a mild to moderate COVID19 course. In conclusion, patients with LSDs experienced varied symptomatology and severity from COVID19 infection, ranging from asymptomatic to critically ill. Risk factors included baseline health status regardless of specific LSD, age, and associated co-morbidities. The sample is too small to make conclusions on specific impact of treatment status on COVID19 severity.

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