Since January 2020 Elsevier has created a COVID-19 resource centre with free information in English and Mandarin on the novel coronavirus COVID-19. The COVID-19 resource centre is hosted on Elsevier Connect, the company's public news and information website.

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In summary, NBS for MPS1 is based on reduced IDUA activity in dried blood spots. In our experience, the majority of samples submitted for NRE analysis as follow-up of an abnormal NBS, had results that excluded a diagnosis of MPS1. We identified a group of infants with mild elevation of urine NREs, who cannot be classified at present; this pattern may represent a very attenuated phenotype. This group would benefit from continued clinical monitoring and repeated determination of these biomarkers. About 20% of cases submitted had a clear pattern of NREs and HS consistent with MPS1. The concentration of HS and its NREs in these cases may correlate with the severity of the disease. The integration of clinical assessment with specific and sensitive biochemical methods, like the NRE method, and with molecular and enzymatic data can help in defining the appropriate management of infants identified by NBS.

**Poster # 52**

**RISK FACTORS FOR EXPOSURE AND MORBIDITY ASSOCIATED WITH COVID19 IN THE MITOCHONDRIAL DISEASE POPULATION**

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**Background:** Individuals with mitochondrial disease (MD) are vulnerable to decline with infection. We aimed to understand the impact of the recent COVID19 pandemic on the MD community by identifying risk factors for COVID19 exposure and severe COVID19 disease in this population.

**Methods:** An online questionnaire was distributed through MD advocacy groups for 8 weeks in April and June 2020. Patients with MD or their caregivers completed the survey.

**Results:** The survey received 688 respondents, with 29% representing pediatric MD patients. The most common MD type were, MD NOS, Mitochondrial Myopathy and Leigh Syndrome. 62% of patients reported MD associated with a known pathogenic variant. 5 positive COVID19 cases were reported. 68 requested SARS-COV-2 test and 14/68 (21%) were unable to receive testing. Symptoms that overlap with COVID19 occurred frequently, including fever (22%) and new or worsening cough (16%). The most common risk factors for exposure to SARS-COV2 were frequent visits to healthcare settings (63%) having a member of the household that is an essential worker (37%). 73% reported at least one condition recognized by CDC as risk factors for severe COVID19. Overall 35% reported respiratory muscle weakness, 31% reported immunodeficiency, and 25% reported asthma.

**Conclusion:** Few people with MD reported COVID19 despite common risk factors for exposure. Many patients had symptoms that overlap with COVID19 and it is unclear whether these are part of their MD or COVID19, given insufficient testing was available at the time of the survey. Most people with MD have at least one risk factor for severe COVID19. Many families affected by MD are at increased risk of exposure due to essential workers in the household and frequent visits to healthcare settings.

**Poster # 53**

**A Milder Phenotype of Asparagine Synthetase Deficiency and Initial Response to Asparagine and Glutamine Supplementation**

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**Background:** Asparagine synthetase deficiency is an autosomal recessive condition caused by mutations in the ASNS gene. It is typically characterized by microcephaly, refractory seizures, spastic quadriplegia, and severe developmental delay. Biochemical testing is not a consistent marker of disease and diagnosis has largely been molecular, with many affected individuals born to consanguineous parents. Asparagine synthetase synthesizes asparagine and glutamate from aspartate and glutamine. Dietary supplementation of asparagine is a logical metabolic approach for these patients. To date there have been three reported cases of asparagine supplementation in patients with asparagine synthetase deficiency. One initially had mental status improvement but developed worsening seizure frequency and supplementation was discontinued after 27 days. The next two patients were siblings, of which the first experienced increased interaction with the environment, while the other remained stable without clear improvements. Here we present a 14-year-old female who is compound heterozygous for previously unreported variants in ASNS. Clinically, she has microcephaly, intractable seizures, and developmental delay. However, her delays are less severe than the majority of previously reported patients. She was started on asparagine and glutamine supplementation after diagnosis and we present her case and initial outcomes after supplementation.

**Case presentation:** The patient was adopted at 5 years old. She started having seizures in infancy, typically absence seizures, that became medically refractory by early adolescence, typically generalized tonic-clonic seizures. Developmentally, she was nonverbal, but able to use a simple augmentative communication device, ambulate for short distances, and ate and drank by mouth. On exam she had microcephaly, spasticity of her extremities, and 3+ reflexes. Brain MRI showed prominent lateral ventricles, thinning of corpus callosum, and progressive...