29 September 2014

Dear Patients and Families,

We are writing to announce a new National Institutes of Health sponsored study to learn about the function of the immune system in patients with inborn errors of metabolism (IEM), especially Mitochondrial Disorders. As many of you are aware, infections can exacerbate underlying mitochondrial dysfunction. We know that the enzymes deficient in Mitochondrial Disorders may also be deficient in immune organs or cells, and we wonder whether this can affect immune system function.

The NIH MINI Study: Metabolism Infection and Immunity in Inborn Errors of Metabolism (www.genome.gov/mini) is an exciting study at the NIH Clinical Center (clinicalcenter.nih.gov). The main goal of our study is to learn about the function of the immune system in metabolic disorders, especially Mitochondrial Disorders. Participants will be invited to the NIH Clinical Center in Bethesda, Maryland for an evaluation. Travel and lodging will be provided for patients and their families. Additional visits may be suggested dependent upon study findings and the level of subject participation. The visits will typically be 1-2 days long. At the first visit, we will perform a physical exam and do a detailed immunologic assessment for all study participants. As part of the assessment, we will measure whether or not you or your child’s immune system responded appropriately to childhood vaccinations and naturally acquired infections.

Additional tests may include:
- body composition testing
- energy expenditure testing
- nutritional assessment
To be eligible, participants must:

- Be at least 2 years of age
- Have a diagnosis of a Mitochondrial Disorder
- Be able to travel to the NIH Clinical Center in Bethesda, Maryland

Results of all clinical testing will be provided to the participant and if desired, their home medical teams. Research findings, when available, will also be communicated to participants.

The NIH MINI team is available to discuss eligibility for this protocol with anyone that may be interested in participating and welcomes all inquiries.

In order to participate in the study, please contact the either myself or the MINI study coordinator by phone or by email. See our contact information below.

Thank you for reading our letter and we hope you will be encouraged to investigate study participation further. Please do not hesitate to contact me with any questions or concerns.

Sincerely,

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