



DEPARTMENT OF HEALTH AND HUMAN SERVICES

Public Health Service



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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 organic acidemias (OAs). As many of you are aware, infections can play a significant role in triggering life-threatening acute metabolic crises in children and adults with IEM, especially in those with OAs. While the standard of care for IEM patients is routine vaccination for childhood and seasonal illnesses in most US centers, there have been no studies to investigate whether the response to vaccination is normal in OA patients. We know that the enzymes deficient in IEM may also be deficient in immune cells, and we wonder whether the enzyme deficiency 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 new 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 OAs. Participants will be invited to the NIH Clinical Center in Bethesda, Maryland for an evaluation. Travel costs will be provided for patients and their families. Additional visits may be suggested dependent upon the level of subject participation. The visits will typically be 2-3 days long. At the first visit, we will perform a physical exam and do a detailed nutritional and immunologic assessment for all study participants. We will offer Hepatitis A vaccination, which is part of the current Pediatric Vaccination Schedule

(<http://www2.aap.org/immunization/IZSchedule.html>).

These recommendations are relatively recent, coming in 2005, and many children over the age of 8 years may have missed getting vaccinated against Hepatitis A. Therefore, you or your child would potentially benefit from receiving this vaccine. During “flu” season, the influenza vaccine is recommended for all children with chronic illness, including those with OAs, and will also be offered. As part of the assessment, we will measure whether or not you or your child’s immune system was able to respond appropriately to vaccine(s).

Additional tests **may** include:

- body composition testing, including a DEXA scan
- energy expenditure testing
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To be eligible, participants must:

- Be at least 2 years of age
- Have a definitive diagnosis of an IEM
- 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.

Important: You are still eligible to enroll in the protocol and be evaluated at NIH even if you or your child have already had HepA or “flu” vaccines or do not want to receive a vaccine. For those patients not undergoing vaccination in the study, we will be able to determine whether you or your child have made an immune response to other vaccines such as measles, mumps, and rubella (the “MMR” vaccine).

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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