Do you recognize these scenarios?

CASE 1

A previously healthy 3 month old girl develops a respiratory infection with fever. Her recovery period is prolonged, and she becomes unusually short of breath. Eventually, even feeding leaves her breathless. Investigations show that she has an enlarged heart that is not functioning well; she is in fact in heart failure. She is hospitalized and given a diagnosis of "idiopathic cardiomyopathy" (heart failure without a known cause).

CASE 2

A child with medium chain acyl CoA dehydrogenase (MCAD) deficiency has 1-2 days of persistent vomiting. His parents bring him to the ER. The doctor doesn't want to start intravenous fluids. He tells the parents, "there's no reason; he's not hypoglycemic and his BUN is not that high."

CASE 3

An athletic high school junior has always participated actively in school and community sports programs. He notices over time that he is having more difficulty getting through practices and games because of weakness and some stiffness. He really pushes himself during a semifinal game but after the game, he develops severe pain in his thighs and calves. Later on, his urine turns dark red. He is taken to the local ER and reassured that "these things sometimes happen and shouldn't happen again".

These are some of the real-life situations in the world of patients and families connected with the fatty acid oxidation defects. The essential teaching points that GMCE needs to make are reflected at the end of this article.

In over 30 years as a metabolic clinician, my patients and their families have been my primary teachers. I have learned so much from them about disease, about their symptoms, how treatment works (or doesn't work) for them, and how they cope with their condition. They have generously shared their perspectives with me about their care, the ups and downs of the medical system, and the challenges they face. Having probed and asked more about these challenges, among the top three concerns is almost always the lack of physicians who are knowledgeable about their diagnosis. As

a result, patients and parents find themselves too often needing to educate an unfamiliar doctor in a new encounter...

It is neither unusual nor a bad thing to inform a doctor about your or your child's illness; however, when patients or parents become the primary source of information on disease assessment and management, that can be more than problematic, it's potentially dangerous. Needing to do so places a heavy burden of responsibility upon a patient or family member. In the ER, it's hard enough to just be the patient or the loved one...

While this problem may be widespread in pediatrics, it is even worse in adult medicine.

It may seem at times there is no clear plan for increasing the number of metabolic clinicians nor one for educating medical specialists about inborn errors of metabolism, but there is hope.

The **Genetic Metabolic Center for Education (GMCE)** has recently launched to develop and provide innovative solutions that address the educational needs of all members of the metabolic community with whom we share a common mission: to see that patients are properly diagnosed and treated in a timely manner. GMCE's novel approach is comprehensive, accessible, and technology-based. The outreach is international.

GMCE is developing resources to make information about metabolic disease an integral part of doctors' thinking. Our strategy is to educate caregivers by assisting them in their patient care and providing a variety of symptom-based learning experiences utilizing a symptom-based approach to diagnosing inborn errors of metabolism. Lessons are clear and easy to follow, and interactive learning is maximized.

For clinicians managing metabolic patients but who have never undergone formal training, GMCE offers a Consultative and Clinical Support Service (CCSS) which provides clinical assistance according to the needs and experience of the client, plus access during emergency situations.

GMCE GENETIC METABOLIC CENTER FOR EDUCATION

Promoting metabolic health through technology and education

For specialists who need to consider metabolic disease when evaluating a new undiagnosed patient, GMCE is developing a range of educational resources that include live day-long trainings, online webinars, and specialty-specific training modules. The teaching is specialty-specific so the information is relevant. It is oriented around the symptoms a physician sees in clinic and gives an approach to making a diagnosis. Workshops are accompanied by recorded patient presentations to create a more complete clinical experience. All teaching is practical and case-based, teaching to what the clinician needs to know.

For biochemistry students in medical schools and undergraduate/graduate studies, and for all clinicians and trainees, GMCE will offer online electives moderated by metabolic geneticists using recorded patient presentations, bringing the faces and the stories of metabolic patients to a wide variety of audiences.

For pharmaceutical companies, and other companies producing products for metabolic disease, GMCE will create opportunities for employee training about the diseases they serve, as well as customized brochures for distribution that provide a symptom-based approach for diagnosing diseases that are treatable.

For nations and regions looking to introduce or expand newborn screening, GMCE outlines a team approach that is customized to address the specific needs of the region, including training for caregivers, educational resources, and ongoing clinical support and education.

The GMCE plan is ambitious but an ambitious plan is needed to develop a medical system in which there are more clinicians who are competent to participate actively in the diagnosis and management of metabolic patients. There's much to be done! For example, over the next few years, GMCE is looking to reach all essential subspecialties in pediatrics, and then enter into adult medicine. Along the way, we want to involve patients and families in the teaching process since patient storytelling can be such a powerful teaching tool. There is a need to develop a library of patient presentations representing a wide range of diagnoses. Such testimonials enhance teaching by humanizing these rare medical conditions, and are a vital part of all our educational offerings.

GMCE will make a difference thanks to the support, input, and involvement of those whose lives have been impacted by metabolic disease. The backing of the greater FAOD community continues to strengthen our efforts and help make them sustainable. We would love to hear from you with any input and suggestions on how we can create a positive and lasting impact. As members of the "metabolic community," we must all work together to be successful. Your cause is our cause.

We are grateful to those who have generously offered financial assistance to advance our efforts, including the FOD Family Support Group, as well as the families and individuals from this community who have both given, and pledged their support, financial or otherwise.

An indication of GMCE's response to the case scenarios mentioned at the beginning of this article:

CASE 1

A previously healthy 3 month old girl develops shortness of breath and is found to be in heart failure. She is assigned a diagnosis of "idiopathic cardiomyopathy". **GMCE recommends ruling out metabolic disease (like long chain fatty acid oxidation defects) in patients who present with unexplained cardiomyopathy. The inborn errors represent a treatable cause of heart failure.**

CASE 2

A child with medium chain acyl CoA dehydrogenase (MCAD) deficiency has 1-2 days of persistent vomiting. The doctor doesn't want to start intravenous fluids. **GMCE recommends IV fluids with 10% dextrose (regardless of the glucose level) when vomiting persists. A child with a fatty acid oxidation disorder who cannot take in sufficient calories is at risk for decompensation which can include coma and sudden death.**

CASE 3

An athletic high school junior develops muscle weakness, and after one particularly grueling game, severe muscle pain and rhabdomyolysis (muscle breakdown). **GMCE**

teaches that there are several metabolic causes of exercise-induced muscle damage. Identification and treatment can help minimize the risk associated with such crises and their complications (kidney failure).

* * * * *

-- for more information on getting involved, email volunteer@geneticmetabolic.com
-- for more information on personal giving, contact Kevin at krobinson@geneticmetabolic.com