Procedures: ER

The most important thing to do if faced with a patient with an FOD in the emergency room is to get them to a room and follow their protocol letter quickly. Even if the protocol letter doesn't make sense to you, it was written for a reason by a doctor who fully understands the rare condition. Also look for medical alert bracelets. Thirdly, with a patient already diagnosed, the family members often know much of what the patient might need. Listen to them.

Symptoms

When left untreated, FOD patients have multiple symptoms. Here are a few:

- Lethargy
- Confusion
- Severe muscle cramps
- Difficulty breathing
- Seizures
- 'Reye' like syndrome

The most important thing is to provide calories. If in the hospital, an IV with 10% Dextrose is usually the most effective accomplish this.

FODs can only be diagnosed through specialized blood, urine, or muscle/skin tests, so if the patient shows these symptoms, it is best to order those tests immediately.

References & sources

Much of this information is from https://fodsupport.org/index.htm or personal experiences. This brochure is only a summary of the most important information. For more information, please check out the website and the additional sources and links on it.

Permission given by

Deb Lee Gould, MEd, FOD Group Director
to use their material in my brochure

Fatty Oxidation Disorders

What are they and why do they matter?

A media guide to FODs;

why FODs matter,

symptoms and procedures,

and personal experiences

Why do FODs matter?

FODs are metabolic disorders. Common sicknesses, such as the flu and other simple illnesses, can be lethal. People affected with FODs have a high risk of death unless diagnosed and treated early, because they cannot process fat for energy. Instead, they process glucose. Patients may present with high glucose as the body "dumps" trying to stay alive. While glucose level is a way to put a number to a possible problem, it doesn't accurately show metabolic distress. Most FODs are included in newborn screening now, but there are still children and adults who are undiagnosed or misdiagnosed.

All FODs are autosomal recessive, which means both parents must be at least a carrier, if not affected. However, there are cases of affected carriers, although they are unusual. There are many types of FODs, such as:

Primary Carnitine Deficiency, CACT, CPT I and II, VLCAD, LCHAD, TFP, MCAD, SCAD, SCHAD/HADH, HMG, GAII, and Unclassified FODs.

For a more complete list, refer to https://fodsupport.org/list.htm

Jennifer's Story

Jennifer had barely turned one before she got terribly sick with croup. She was refusing to eat and had a temp of 102. Jennifer had been restless all night, and by 7:30 in the morning could not even crawl anymore. Her mom took her to the doctor, but the nurse took one look at her and sent them off to the ER. When they got there, Jennifer was in a coma with a blood sugar of 20. If she did not get treatment soon she most likely would have died. Even still, Jennifer was not responding to anything until she got a bolus of sugar. She was then sent by ambulance to the Cardinal Glennon Children's Hospital, with her IV bag decreased from D10 to D5 (it needs to be D10). When they arrived, Jennifer was lethargic again, and again had a blood glucose of 20. She was in ICU for one and a half days, then she was moved out because the doctors had decided she was stable as long as they kept her on the dextrose. By day 4, she was diagnosed with MCAD. Most children do not survive their first illness if they were not diagnosed at birth. Jennifer was one of the lucky ones who survived.

Michelle's Story

Michelle was born as a healthy child, with a normal life up until her second birthday. Then she became ill. Michelle had severely low blood sugar, and was in the hospital for five days in a coma. All of the seventy tests the doctors ordered came back negative. Michelle eventually got well, and went back home. Her life was fine until she was twenty and she went camping with some friends. The day after they climbed a mountain, Michelle was sick at 5am. At 8pm, she went to a hospital, but they felt they would be unable to treat her and sent her back to her family and the hospital there. She was lucid, and her blood pressure, temperature, and heart rate were fine. Blood and urine tests came back with negatives for blood alcohol levels, illegal drug use, with glucose and electrolytes "well within range" according to her doctors. She also had a CT scan which also revealed nothing. Michelle was moved to Observation, and three hours later, she died. Michelle's autopsy produce no hints to her cause of death, but her mother keep looking for an answer. Fourteen months after Michelle's death, the physician who took care of her in Observation contacted Michelle's mother with the answer – Michelle had MCAD! Needlessly, it was fatal for Michelle because it was not diagnosed early and treated on a daily basis.