Summary – Munchausen by Proxy Accusations in Children with Mitochondrial Disease
Dr. Alex Flores

Munchausen's Syndrome is defined in Taber's Medical Dictionary (17th edition) as "a type of malingering or fictitious disorder in which the patient may practice self-mutilation and deception in order to feign illness." Munchausen was a fictional 18th century German baron who entertained others by lying about various illnesses and symptoms. It was around 1951 that Munchausen Syndrome and Munchausen's by proxy were named as diagnoses. Munchausen's by proxy occurs when adults falsify symptoms of illness or even cause them in a child and is considered a form of victimization and abuse.

Because children with Mitochondrial Disease present with different and often changing symptoms, including fluctuating or "impossible" gastrointestinal issues, Munchausen's by proxy is unfortunately the mistaken conclusion drawn by health care professionals who have little or no understanding of Mitochondrial Disease. This discussion is only a beginning step toward addressing this issue, and is intended to educate and open a dialogue that ultimately should improve collaboration and care of the child with mitochondrial disease.

Intestinal Failure
Children with intestinal failure present with many complex symptoms and are among those who are most likely to be involved with false allegations of Munchausen's by proxy. Intestinal failure is the inability to digest or utilize nutrients. Patients with intestinal failure are severely ill and can eventually die from this condition. Patients with other serious GI diseases such as Crohn's disease actually survive longer than those with intestinal failure. Pediatric patients with short bowel syndrome and other congenital GI disorders fall into this category. It is important to note that though you can survive without the large intestine, a person cannot survive without the small intestine.

There are many disorders that can cause gastric dysmotility and there are many anatomical anomalies that can cause some of these severe GI diseases especially among children. However, dysmotility may also present even without an anatomical problem; often this situation occurs due to problems related to the muscles and nerves which control the GI tract (see Dr. Flores presentation about dysmotility in mitochondrial disease).

Efforts to treat gastric dysmotility become paramount because, though a person can live on TPN (Total Parenteral Nutrition) for a long time, numerous and serious problems arise with long-term use. Most frequently, the lines become infected or the patient simply runs out of sites for these parenteral lines. There are not a lot of options for these patients, including children, who cannot eat and therefore cannot survive - and from this perspective, it is little wonder that the parents could become overwhelmed by their care. However, instead of providing support, it is often these very parents who are

most likely to be accused of Munchausen's by proxy, i.e. purposefully harming their child or "creating" the condition.

Characteristics of Munchausen's by proxy
There are certain factors that seem to be the most common when a diagnosis of Munchausen's is made. Numerous unusual and inconsistent symptoms are reported. The reports from parents and health care professionals often are inconsistent. Often diarrhea, constipation, nausea, vomiting and rectal bleeding are all reported. Note that these are often the very same symptoms that children with real disease (i.e., Mitochondrial Disease which affects their GI tract) present with. In 30 years of practice, Dr Flores has only encountered four patients with true Munchausen's Syndrome - all others have had real health issues, which were unrecognized or misdiagnosed.

Other "red flags" related to Munchausen's are symptoms which occur only when parents are present, caregivers who are extremely knowledgeable about the disease and tend to "teach" health professionals, multiple health providers who state different things, absent fathers, multiple hospitalizations, and parents who block the idea of stopping treatment modalities. In order to interpret the symptoms appropriately, the health care providers must be experts in the areas of the patient's primary symptom, must perform motility studies and must be able to accurately interpret these, and must be able to communicate with the patient's team to understand the results as part of the bigger picture.

It is possible that what may be interpreted as "red flags" of Munchausen's may alternatively be attributed to the demands and anxiety related to care of a very sick child. For example, anxious parents may not give a good history, or may "doctor shop" because they are unsatisfied and may be unhappy with the care their child is getting, especially when they feel that no one can actually diagnose, treat or understand the problem. Certain conditions, especially mitochondrial disease, will present with intermittent symptoms, and it will take a skilled and patient clinician to arrive at the right diagnosis - one that is an illness not Munchausen's by proxy. Psychologists have described that the population of patients and parents of children with Mitochondrial Disease are much more vulnerable to a false Munchausen's by proxy accusation simply due to the nature of the disease. In fact, a hallmark characteristic of mitochondrial disease is the presentation of several unrelated symptoms that together, "don't make sense". Clinicians who feel that a parent is intentionally making symptoms appear, is behaving to ensure that the illness continues, and consults multiple physicians may suspect Munchausen's - but should still "trust, then verify." In other words, believe the parents, run appropriate diagnostic tests, seek the input of every part of the child's team, and take very seriously the responsibility to the child to act as an advocate and do no harm.

Management Issues
Because patients with gastric dysmotility present in so many different ways, management can be difficult. Attention must be paid to every minute detail of the

history: diagnostic tests, previous documentation, biopsies, and lab results. It is the responsibility of the individual bringing the diagnosis into question to involve or request a team approach, utilizing nurses, social workers, psychologists, nutritionists, physical therapists and legal advisors. The hospital may not be the best setting for all of this to work. It should only be after careful consideration that the child be separated from the parents, and recognition of the issues surrounding the separation if the child has mitochondrial disease are very important. Symptoms of mitochondrial disease may fluctuate, or be difficult to recognize. Parents caring for a chronically-ill child with a untreatable condition can appropriately cause anxiety, fear, concern and "over-reporting" of symptoms. Caution should also be taken when altering a child's regimen or routine, as the abrupt shift can be devastating for the child suffering from a mitochondrial disorder (a good example is stopping the administration of "unnecessary" fluids, which in a child with mitochondrial disease, may be the buffer that prevents the child from entering a metabolic crisis.)

Case Studies

Dr. Flores presented two case studies as examples. The first is a patient who currently is a 25-year-old female with complex GI disease due to a mitochondrial disorder. Her history includes use of long-term TPN and a gastrectomy.

When this patient was 20 months old she presented with many GI symptoms and several relapses. At 2 years of age, because of the reoccurring symptoms, her mother was investigated by a very reputable hospital for Munchausen's by proxy. At age 10 she had additional treatment, developed infections, and demonstrated abnormal GI motility. Finally, at age 16 she received a muscle biopsy and was diagnosed with Mitochondrial Disease and is currently being treated and managed as well as possible. It took many years and many diagnoses to get to where she is today.

In contrast, Dr. Flores remembers a case of a 19 year old female who presented with bowel related symptoms, had multiple surgeries and even a bowel transplant and a splenectomy. Eventually it was determined that this was a real case of Munchausen's by proxy. The mother refused psychiatric care. Clinicians should feel obligated to finding a correct diagnosis before judging, and must be willing to recognize that the medical establishment does not always have the answers.

Questions from the MitoAction Listeners

Some of the "red flags" of Munchausen's's by proxy - use of medical terminology, multiple specialists,reporting of symptoms, etc. - may be exactly the pattern that parents of a child with mitochondrial disease demonstrate by necessity....how do you distinguish?

This is true. The lack of continuity of clinicians often occurs with Mito patients because parents feel that they have to be the coordinators of the child's care because no one knows very much about Mito. Here in New England we are lucky because we have medical centers where Mito is known and managed, but in other parts of the country this is not the case. Whenever possible, parents with children with Mito or with inconsistent symptoms, should seek out a major medical center as near home as possible. The key

is to find a physician who is interested in your child and is willing to spend time with you and to get to know you - old fashioned "good doctoring."
In areas of the country where there is not a great deal of information or knowledge of Mito, in order for parents to avoid being considered "over labeling" which can be a red flag, they should refer clinicians to the Mito web site and to the organization Mito Action. Doctors can also be given the names of experts in other parts of the country who they can contact about Mito - to convince them that the symptoms the child is suffering are real!

The Clinicians Guide which is online at the MitoAction site provides an excellent resource for those in parts of the country where knowledge about Mito might be minimal. In this way, the parent is not "labeling" the child with a specific disease. The clinician can find out himself or herself just what the symptoms are and how they might present in children and come to the diagnosis him/herself rather than through the parent.

MitoAction can also act as an advocate and help to explain the complexity of Mitochondrial Disease and the variability potential of symptoms. Mitochondrial Disease is still a disease in its infancy as far as what we know about it and how to treat/manage it, as was gastric dysmotility when the disease was in its infancy 20 years ago. Another argument against false accusations of Munchausen's by proxy when the patient has Mitochondrial Disease is that it is very possible that the child may not "look sick". In many ways, Mito is truly an invisible disorder and is confusing due to variable presentation even within a same family with the same phenotype or genetic inheritance of the disease.

How do you deal with a child where Munchausen's was first diagnosed, then real disease was discovered later?

This may be devastating and should be considered before intervention (separation) is forced upon the family. Children do remain frightened and fear that separation from parents may be long term. The child should be reminded, however, that the parent did win out in the end and was persistent enough to finally get a real diagnosis for the child. Some parents who have experienced this trauma report that their children are still anxious, fearful, and unable to recover from the psychological damage years after the wrongful accusation and separation. For children with mitochondrial disease, they are often unable to control or monitor their symptoms and rely on parents to act as their voice and their advocate.

Is there any other way to distinguish Munchausen's from real disease other than separating the child and parent when the symptoms only occur in the parent's presence?

In Dr. Flores experience, his opinion is that separation is sometimes necessary, but unusual if necessary for more than three to five days. However, even a short separation can be very traumatic for the child and the family, and should be handled

with great sensitivity and respect by all involved. The family is already stressed because they have a very sick child and no definitive diagnosis. They are afraid that their child may die. Clinicians hold the important responsibility to advocate for the child and respect the family.

Another factor here to remember is that when a child is at home, parents and children understand symptoms and describe them differently. Fatigue in a child with Mitochondrial Disease does not always mean that the child looks sleepy; instead, a child with mitochondrial disease related fatigue may become "unorganized", irritable, unfocused, less verbal, avoidant, complain of pain, as he or she becomes fatigued. In many cases, parents are able to recognize and identify the nuances of the child's symptoms better than others who are less involved with the child, including teachers, nurses, etc. The manner in which symptoms are manifested becomes very important and should be described in detail when sharing and taking the child's history. It cannot be emphasized more clearly that, with mitochondrial disease, it is not unusual for the entire body to be potentially affected - the battery is literally running on low. Interpreting this and working through the complexity of the symptoms is essential to get to an accurate diagnosis.

One listener shared the story of her daughter who had multiple episodes of regurgitation at home and none recognized at school. Further evaluation demonstrated that the child had over 100 episodes in one school day, but the school personnel only recognized very obvious vomiting rather than the smaller but real episodes which this young patient was having. This highlights the need to describe and understand the presentation of the child's symptoms in detail.

A listener who acts as a patient advocate shares that it is also possible that a child who is separated from home/parents will indeed improve while hospitalized because there is air conditioning (home has none), he is getting lots of rest (no school), lots of fluids (IV's), and reduced stress (no expectations, school, work etc). Again, perhaps this appears to be a case in which the parents are causing harm, but could be a legitimate result of misinterpretation of the effects of the environment on mitochondrial disease. Mitochondrial disease is a diganosis that places significant stress on a family and caregiver burden on the parents. Awareness of the disease, support of the patient and family, and recognition amongst residents, medical fellows and physicians about the potentially devastating impact of a false Munchausen's accusation are critical in order to offer hope to the family caring for a child with mitochondrial disease.