RARE DISEASE CARETAKER VS. MUNCHAUSEN SYNDROME BY PROXY/MEDICAL CHILD ABUSE PERPETRATOR: A COMPARISON OF PSYCHOLOGICAL PROFILES

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Little Joey, a brown-eyed toddler, is brought into the ER at night. His mother is with him and appears frantic, stating her son had been flailing his arms and legs while his eyes rolled back in his head. Joey is quiet, but responds appropriately when medical staff evaluate him.

Becky, a 13-year-old teenager with long blonde hair, is brought to her third gastroenterologist for consult because she is losing weight. Her mom tells the specialist that her daughter often vomits. Becky has undergone several basic tests, but so far no physician can figure out what is ailing her.

A teacher observes that young Michael often falls asleep at his desk. On occasion, he seems to have difficulty concentrating on activities in the classroom. The issue has been brought up in the past with Michael's parents, the school nurse, and his school counselors. Michael is in the process of being worked up by physicians for a possible physical cause behind his lethargy, but there are also times that Michael, who openly loves sports that involve chasing a ball, has so much energy he is one of the most active children at recess!

The children in these scenarios all need an intervention of some type. But what intervention should be considered, and who should be intervening? Any of these situations might warrant suspicion for a type of child abuse known as Munchausen's Syndrome by Proxy or Medical Child Abuse, but it is also true the children might be suffering from medical conditions that mimic Medical Child Abuse in presentation. There are many situations like these in clinics, schools, day care centers, and other areas where children gather. Some problems present more subtly, while some are more convoluted, but in all cases, working through the clues is a difficult task for anyone not equipped to carefully distinguish between the signs of true abuse versus true illness.

While there are many forms of child abuse, this article focuses on Medical Child Abuse (MCA), formerly known in the United States as Munchausen's Syndrome by Proxy (MSBP), or Caregiver-Fabricated Illness. This article will explain the basic presentations of both caretakers and children involved in MCA cases, and compare and contrast these traits with those seen in caretakers and children affected by mitochondrial diseases ("Mito"), a group of rare, devastating, diseases with potential to cause a variety of serious physical manifestations. There are clear similarities in clinical presentations of patients, as well as perceived caretaker reactions, between both groups, which make it imperative to understand distinctions between a perpetrator of MCA and a caretaker of a child with a rare disease. Although the focus of this article is to show how patients and families affected by mitochondrial diseases in particular can be confused with those affected by medical abuse, it is important to keep in mind that other rare disease communities are similarly affected by false abuse accusations. Lack of understanding, education, and awareness

of what a person or family living with a rare disease experiences in daily life, are issues for all patients and families with complex and difficult medical conditions.

From Munchausen Syndrome by Proxy to Medical Child Abuse: An Evolving Definition

In 1951, a physician named Richard Asher first wrote about patients who engaged in self-harm, or created fictitious medical histories for attention-seeking purposes, naming the condition Munchausen Syndrome, after Baron von Munchausen, who was known to tell outrageously tall tales while he worked within the Russian army in the 1700s (2, 20). In 1977, Sir Roy Meadow, a British pediatrician, coined the term Munchausen Syndrome by Proxy (MSBP) in a paper describing his theory of caretakers who faked illness in a person under their care, usually a child (14). The designation "By Proxy" indicates that the person with the disorder is acting through another person; in this case the child or person under the control of the perpetrator, rather than reporting the symptoms in himself or herself. When a caretaker, usually the mother, purposely exaggerates, falsifies information, or even creates symptoms in a child under her care to make it seem the child is ill, that caretaker is considered to be an MSBP perpetrator. In some cases, it is not understood why the caretaker commits these acts of harm to the child (7), but the belief, according to Dr. Meadow's understanding of the disorder, is that the caretaker may be psychologically ill, with such a strong need for attention that she jeopardizes the health of her own child (6).

Clinical presentation of a child in the care of an MSBP perpetrator may include symptoms such as seizures, vomiting, bleeding, cardiac arrest, apnea, sepsis, and multiple organ involvement (25). MSBP perpetrators have been known to do such things as smother their children to unresponsiveness, administer poisons, and withhold foods to make their children ill enough to need medical intervention (9). The unifying feature for all MSBP cases however, is that even if the clinician did not witness the event, the child's symptoms are brought on by something the perpetrator did to the child. Cases have been proven by surveillance videos in hospital rooms, in which hospital personnel observed a caretaker engaged in actions such as withholding a needed medication, smothering, administering poisons or foreign substances to the child, and other disturbing behaviors (6, 8). Results of MSBP perpetrators' actions can have mild to life-threatening effects on the child. MSBP is a very serious disorder that endangers the health, well-being, and possibly the life of a child under a perpetrator's care.

Frequency of occurrence of MSBP is not fully known, but is thought to be exceptionally rare (5). Some studies have shown a prevalence of 2-2.8 out of every 100,000 children under age one are victims of MSBP (13), but it is difficult to determine the true prevalence rate of this syndrome. Some cases of MSBP will be missed due to the subtlety of the signs, while other cases will receive mistaken diagnoses of MSBP simply because the symptoms of the child, and the behaviors of the family in dealing with a true medical condition, will be confused with signs and symptoms of abuse. In many states, child protective services will only agree to return children to the home if parents agree to "admit" they committed MSBP abuse (15). This presents strong incentives for innocent parents to agree they are guilty, if only to have their children returned. MSBP must be acknowledged to be very real, inasmuch as some cases have been proven by surveillance, but it is equally important to understand that there are limitations and uncertainties

to a positive diagnosis if the alleged perpetrator was not physically observed inducing or fabricating medical symptoms.

Medical Child Abuse (MCA) is a more recent abuse label used for situations where children have multiple consults with physicians, or undergo many medical tests, procedures, or surgeries, in an attempt to diagnose and treat symptoms the caregiver describes to the doctors (1). If the child does not truly have medical problems as the caregiver has described them, and the medical care given is actually unnecessary, the child is considered a victim of "overmedicalization." Overmedicalization means the child has had unnecessary and harmful, or potentially harmful, medical procedures and/or interventions, for no true medical reason. As MCA is currently defined, accusations may be made even if the child in question actually has a true illness, but doctors believe the child has too many interventions for the level of symptoms the illness creates. Unfortunately, with the current vague definition, accusations can be made even when the parent has no intent to derive personal attention or gain from the illness of the child, and no intention of hurting the child at all, but simply asks for medical interventions that one or more doctors think are unnecessary. Such a nebulous definition of "abuse" creates broadening scrutiny of parents dealing with children who have poorly understood or controversial medical conditions. It puts rare disease families at especially great risk of being falsely accused of abusing or overmedicalizing their sick children in situations where physicians don't understand the child's medical problems.

What is a Mandated Reporter?

A mandated reporter is a professional who, by virtue of his or her duties and training, is legally required to report to child welfare authorities any suspicion of harm to a child, the elderly, the disabled, or other vulnerable populations (11,18). In some states, any citizen is expected to report an act of suspected abuse, and in all states, there are certain professionals who are mandated to report what they perceive to be a danger to a protected person (11).

It is the professional responsibility of all mandated reporters to educate themselves regarding what constitutes child abuse, what to consider as signs of abuse in a child, and when to be concerned about a possible adult perpetrator who may be inflicting harm. Some states require training by a mandated reporter on what is considered child abuse or neglect and how to begin the reporting process, as well as what to expect thereafter (20). All states have information available on which professions are mandated reporters, and the importance of taking this role seriously. Many states offer online programs that grant completion certificates. At least one state offers educational credits to social workers upon completion of their online program (3), and at least one state devotes material to recognizing MSBP perpetrators (3). Those mandated to report suspected abuse include, but are not limited to: physicians, nurses, law enforcement officials, social workers, physical therapists, CNAs and home health aides, coroners, teachers, coaches, counselors, child care providers, pharmacists, and firefighters (4).

Mitochondrial Diseases: A Representative Group of Rare Diseases

Mitochondrial diseases (Mito) are a group of disorders caused by defects in the mitochondria, which are the energy-producing powerhouses of the cells. Disease-causing genetic mutations, and sometimes environmentally induced damage to the mitochondria, decrease energy supplies to organ body systems, which in turn causes a wide variety of different medical problems. As a result, mitochondrial diseases typically present as multisystem disorders. Any body system, or even several body systems, may be affected, including the ears, eyes, brain, and cardiovascular, endocrine, renal, gastrointestinal, and respiratory systems (12).

Current research shows that mitochondrial diseases likely affect as many as 1 in 2,500-3,000 people (16). Scientists are discovering that mitochondria play a huge role in diseases such as Alzheimer's, diabetes, cancer, and many other well-known disorders (15). As such research advances, it is suspected researchers may discover mitochondrial diseases are even more common than currently thought. Since it has been a slow process in many ways toward awareness, education, and research into mitochondrial diseases, it is essential to strive toward better understanding, more efficient treatments, and cures for those affected by this devastating group of illnesses.

Mito occurs in children and adults alike, and multiple people in a family may be affected if they share a disease-causing genetic link (16). The disease may present as a spectrum of illness from mild to severe, and symptoms may wax and wane over time within an affected person. A person with dysfunctional mitochondria is not always able to keep up with the energy demands made by the organs and systems within his or her body. Symptoms in one body system may improve as he/she "recharges" with rest, only to have another body system show problems as energy is again depleted by activities of daily living. At times of significant stress on the body, such as illness, heavy physical activity, or anything that increases the body's need for energy, the energy depletion from dysfunctional mitochondria can be severe enough that the cells in a body system die. There are no cures for mitochondrial diseases (22). People die -- even children die-- and families diagnosed with Mito are acutely aware of this fact. Mitochondrial diseases are complex, overwhelming, unpredictable, and at times terrifying.

Unfortunately, mitochondrial diseases are not well known by the general public, or even in the medical world. Symptoms of mitochondrial diseases are not always understood by the different medical specialities involved with a person suspected of, or diagnosed as having, Mito. Health care workers, including physicians and nurses, are not taught much, if anything, about mitochondrial diseases during their training. Partly as a result of this, there are few specialists dedicated to mitochondrial medicine in the United States; so few, in fact, there are not even enough to represent every state (10). Physicians who do specialize in mitochondrial medicine are often overwhelmed with the needs of their many complex patients. Due to the lack of awareness and education about Mito in the medical community, mitochondrial specialists must carefully maneuver through an often ignorant, and sometimes hostile, medical system to make sure their patients have access to proper medical care.

What Connects Medical Child Abuse and Rare Diseases Such as Mitochondrial Diseases?

Since Mito is a complex and unpredictable group of diseases in which even most medical personnel lack education, there is significant opportunity for misunderstandings in the presentation of physical symptoms. Most medical practitioners lack time to research or fully analyze one particular patient's case. For some professionals there is an unwillingness to learn about unknown conditions, and some feel threatened if a patient knows more than the doctor about a disease. Combined, these problems can create an unfavorable environment for patients and families seeking medical care for mitochondrial diseases. Doctors and nurses pressed for time, and lacking education or a desire to learn about mitochondrial problems, sometimes perceive Mito patients as describing strange symptoms that don't make sense, and as asking for medical interventions they don't think the patients really need. This can lead to suspicion that patients or family members are exaggerating or fabricating medical problems. Busy, distracted, and sometimes even arrogant professionals may feel it is safer and easier to simply report all potential issues as suspicion of abuse, and let someone else handle the situation, rather than get involved in a long process of testing and treating unusual and difficult medical problems. In this manner, false accusations of Medical Child Abuse can be made against families struggling with legitimate illnesses like mitochondrial diseases. A diagnosis of a rare disease such as Mito does not rule out the possibility of abuse, and it is possible to have both at the same time within the same family. However, mitochondrial diseases are thought to occur in the population of the United States at a rate of about 1 in 2,500-3,000 people (16), and the few studies done on abuse in a medical setting have been done on MSBP, with an estimated prevalence at about 0.5 out of every 100,000 children under the age of sixteen (13). This means the average medical professional is roughly 16 to 20 times more likely to encounter a child suffering from a mitochondrial disease than Medical Child Abuse such as MSBP!

Parents Accused of MSBP/MCA

For the reasons listed above, parents and caretakers of patients with rare diseases like Mito are more vulnerable to accusations of Medical Child Abuse than other parents of children affected by disease. Adding to this vulnerability is the unfortunate fact that the behavioral traits supposedly displayed by medically abusive parents are very similar to normal behaviors seen in parents of legitimately ill children. This only increases the confusion medical professionals might have regarding whether a child has an abusive parent, or simply a parent stressed with the many burdens of caring for someone with a difficult illness.

Listed below are behavioral traits supposedly indicative of Medical Child Abuse and/or Munchausen's Syndrome by Proxy, with explanations of how perfectly normal and attentive parents might display the same behaviors when caring for a child with a disease like Mito.

Caretaker Profile for Medical Child Abuse And Munchausen Syndrome By Proxy (REFERENCE)	Caretaker Profile of child with serious chronic illness (such as a mitochondrial disease).
Primary Caretaker is usually the mother. (1)	It is common for the mom to be primary caregiver for a child, especially when that child is ill and cannot function in a standard daycare or school setting.

The caretaker is extremely knowledgeable about the child's supposed illness. (9)	The caretaker of a child with serious illness is usually well-educated on that illness. She has listened to many physicians explain different issues in the child's illness. The caretaker has spoken with others affected by the same illness in support groups, and on advocacy hotlines. Out of concern for her child, the caretaker has often done her own research as well.
Caretaker may have medical background or work in the healthcare field. (1)	People with medical backgrounds have children. Some have children with severe illness or disease.
Caretaker stays close by the child during hospitalization. (15)	A caretaker's goal is to care for and protect, her child. When a child is hospitalized, there is legitimate concern for the health of that child. The child usually feels ill and is outwardly showing signs he/she wants the caretaker around. The child is stressed and needs familiarity. Why would a parent disregard the child's need for his/her mother or other caretaker? Also, caretakers naturally want to stay close to see that the care given is what the child needs.
Caretaker becomes friendly with the medical staff. (1)	A chronically ill child has probably been in hospital clinics, or even hospitalized inpatient, frequently. Some ill children spend weeks, even months, in a hospital setting. The parent has had ample conversations with the child's medical providers. Friendship and/or pleasant relationships are a natural result of some of these conversations.
The child is taken to multiple physicians and may be seen in multiple facilities. (9)	A child with a suspected or confirmed diagnosis of a rare disease, such as mitochondrial disease, may often be referred to different physicians, and even other care facilities for specialized care. A team of knowledgeable physicians may not be available in just one facility. Also, the majority of physicians are not familiar with rare diseases, so parents may need to consult several physicians before finding one who is willing or able to help.
The caretaker does not seem relieved by normal test results and may demand further testing or procedures, and even a second opinion. (15)	A caretaker often has an intuitive sense that something is wrong with her child. If a child is exhibiting troublesome symptoms, she will want to know why. A physician who says everything looks normal on paper, "so be happy," only makes a parent feel fearful and helpless when he/she has witnessed the child's symptoms, and knows the child is ill. A second opinion is not only a natural thought, but an intelligent one, for parents in such circumstances.

The child has one or more medical issues that do not present or respond to treatment as expected. (15)	Mitochondrial diseases are unpredictable and involve many body systems; symptoms may not be consistent. One of the hallmarks of these diseases is uncertainty in presentation and treatment response.
The caretaker seems abnormally calm when the child is hospitalized. (15)	People handle crisis differently as a result of learned responses, cultural differences, or coping mechanisms.
The child's symptoms are not witnessed by medical staff. (1)	There are multiple reasons symptoms may not be witnessed by people who are not close to a sick child, and around him/her on a continual basis. There may be few family members or friends available for daily care of the child, meaning the same person does the majority of caretaking and sees the most symptoms. Some symptoms are brief and leave no after-effects; symptoms may have resolved by the time the child reaches an ER, or the caretaker may have successfully given treatment before the child is seen by the medical team. For example, witnessing a focal seizure in a child can cause a caretaker to seek help, but such seizures can present briefly with no after-effects, making it very easy for doctors or others to miss all signs of the problem.
There may be a history of similar illness or death in a sibling. (15)	Mitochondrial diseases, and many other rare disorders, present mainly as genetic diseases. Any, or even all, siblings may be afflicted with the disease. Some may die.
There may be discrepancies in clinical findings and histories. (15)	A patient with Mito is complicated. The patient may have many clinical notes in various facilities. Specialists often take clinical histories rapidly while talking to parents at appointments; it is common for mistakes to occur. It is also common for communication between doctors and hospitals to be less than perfect or accurate. Many complicated patients have varying accounts of their medical problems in their records.
Caregiver is demanding, or seems to require constant attention. (15)	The caregiver of a child with a rare, complex, and unpredictable disease such as Mito is emotionally drained. The caregiver knows well that this disease may take his/her child's life. The caregiver watches his/her child struggle every day with symptoms. A caregiver knows not all medical staff are trained to deal with unusual diseases, and may fear an issue will be missed causing harm to the child. It is natural for parents in such circumstances to experience stress and appear emotionally needy at times, particularly if the child is struggling more than usual.

What Harm Can Be Done by Mistaken Accusations? A Great Deal!

Mandated reporters are often encouraged to believe it is better to mistakenly report an innocent parent for child abuse than overlook reporting a parent who is guilty (3). This "better safe than sorry" frame of mind can easily discourage careful thinking about the legitimacy of the concerns before reporting. Mandated reporters must report suspected abuse, but need to consider whether what they observe reasonably amounts to a true suspicion of abuse, and not simply confusing of potentially concerning behaviors that have other clear explanations. To report without thoughtful consideration of other explanations assumes that little or no harm comes to children and parents involved in false accusations of Medical Child Abuse. In reality, a false accusation of Medical Child Abuse against a caretaker can have truly devastating effects on the child, family, and entire community, such as:

- The child being removed or separated from the family. Children removed from their families on suspicion of Medical Child Abuse have spent many months, and even years, separated from a loving family (23). Separating a parent from a child at a moment of great emotional, mental, and physical need can negatively affect a child psychologically for years, or even a lifetime.
- Generating the breakdown of the accused family's relationships.
- Interfering with medical care the child truly requires. Medical care for the child is often removed or stopped as part of a Medical Child Abuse investigation. A sick child can be medically harmed as a result. Some children involved in Medical Child Abuse investigations have even died as a result of insufficient medical care during the investigation (24).
- Creating mistrust of the patient and family for medical staff and social workers that will continue for a lifetime, as well as mistrust of the clinic, facility, or school from which the accusation originated. In the long term, this can lead to Post-Traumatic Stress Disorder (PTSD) in the patient and family, and even jeopardize the patient's future health, as the family will fear seeking help from doctors or the ER.
- Creating mistrust against the medical community in the accused family's friends, and the rare disease community. The stigma of handing out false accusations will hold tightly against the accuser and/or facility involved.
- Severe financial burden on the family who must hire attorneys to defend against the false accusation.

False reports of Medical Child Abuse harm children and families. A professional wondering whether or not potential concerns about a family reasonably amount to "suspected Medical Child Abuse" should consider these questions:

- Has this child been strongly suspected of, or diagnosed with, a complicated disease?
- If yes, have I researched this child's reported or suspected diagnosis? Have I spoken with someone well informed about this disease, such as a specialist or a patient advocate for the disease?
- Can my potential concerns be explained as symptoms of this disease?

- How common is the suspected or diagnosed disease? Is is significantly more prevalent than Medical Child Abuse? Statistically, which am I more likely to be dealing with?
- Can my potential concerns about the caregiver's behavior or the child's care be explained as a natural product of caring for a child with a complicated and difficult illness?
- Given the answers to these questions, is it *reasonable* or *necessary* to suspect child abuse is the best explanation for the behaviors or situations I find potentially concerning? Do I actually suspect abuse is the cause of my potential concerns?

The lives of patients and families living with a rare diseases can be challenging. In addition to medical challenges, families struggle to deal with fears, anger, and uncertainty over what tomorrow might bring. The burden of worrying about false accusations of Medical Child Abuse should not have to be added to the many struggles families already face. Professionals who truly desire to help patients with complicated illness will be thoughtful and careful in their reporting of suspected Medical Child Abuse. False reports harm children and families. Patients and families trust medical professionals to help them, not harm them. Everyone on the team participates in that trust, and therefore everyone on the team has equal responsibility to carefully consider the risks and benefits of making suspected abuse reports.

Special Considerations for Adults Strongly Suspected or Diagnosed with Mitochondrial Disease

Children and parents are not the only ones to suffer from false accusations and misunderstandings about their disease. Adults with Mito are often suspected of exaggerating or fabricating their symptoms, or even speculated as having a psychological source to their ailments, rather than a legitimate medical condition. Many adults spend years going to clinics with maladies that physicians find perplexing. They are diagnosed with depression, anxiety, or even more extreme psychological conditions such as conversion disorder, malingering, somatoform disorder, or factitious disorders. Some of these adults spend years in therapy trying to treat what really are true physical symptoms caused by Mito.

Feelings of depression and anxiety are natural emotions someone with a complex, rare disease might experience at one time or another. Furthermore, an adult with Mito may have a true psychological component to his or her disease process in addition to other symptoms. This possibility should be evaluated carefully. It is detrimental, however, to mistake emotional and psychological reactions to a chronic disease as the actual source of physical disease symptoms. Some adults report accusations of recreational drug use because their symptoms are considered disproportionate to what doctors understand about their medical condition. Some very medically ill patients have ended up admitted to hospital psychiatric floors because ER staff did not understand the physical effects of their disease. Many are characterized as chronic complainers, and exaggerators. It can be emotionally damaging to not be taken seriously while suffering troublesome symptoms. It can be frightening to feel so ill and have no one understand the symptoms to have a real physical basis. For these adults it is important to validate the physical symptoms they experience as real.

Conclusion

In summary, Medical Child Abuse is a very rare form of child abuse in which a caretaker causes unnecessary medical care to be given to a child. This includes purposely harming a child through poisons, smothering, or fabricating symptoms for the caretaker's emotional benefit, as seen in conditions such as Munchausen's Syndrome By Proxy. It can also involve exaggeration or fabrication of the child's medical symptoms for other reasons. The result is that the child is harmed, or potentially harmed, through medical care that is unnecessary.

Mitochondrial diseases are difficult to diagnose neurometabolic disorders that are complex, produce unpredictable symptoms, and may have devastating effects, including body system shutdown and death. These diseases are not well known in the general community, or even in the medical community. A patient with a mitochondrial disease may present clinically in a way not understood by his or her medical team, leading to confusion over the cause of medical symptoms.

Physicians, nurses, social workers, teachers, and others who may not have regular contact with people touched by rare diseases such as Mito, may be unaware of the similar presentations between a family committing Medical Child Abuse and a family dealing with a rare disease. Such confusion can, and has, led to mistaken accusations of Medical Child Abuse against people caring for children with Mito and other rare and difficult diseases. The consequences of these false accusations are always serious, and sometimes devastating, to the child and family. With increased awareness and education, the goal is to reduce false accusations of abuse, and increase support for struggling families caring for children and adults with rare diseases.

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