

**Summary -  
Transition for Teens and Young Adults with Mitochondrial Disease  
Presented by Dr. Hope Schreiber**

*Tufts New England Medical Center*

**Introduction:** Dr. Hope Schreiber leads today's discussion, focusing on transitions for teenagers and young adults with mitochondrial disease. Dr. Schreiber is a longtime supporter of MitoAction and strong advocate for the entire Mito community. Schreiber is a clinical neurologist at Tufts Medical Center in Boston as well as an Associate Professor at Tufts Medical School with over 20 years of experience. Schreiber's recent research appears in the December 2012 *Journal of Child Neurology*.

Cristy Balcells, Executive Director of MitoAction, and the mother of a 9-year-old with Leigh's Disease, also joins the discussion. As a nurse with a degree in Public Health, and the author of *Living Well with Mitochondrial Disease*, Cristy brings much expertise to this discussion. Cristy has watched the Mito community grow from one that hardly existed when her daughter was first diagnosed to one that today provides much needed advice, education, and support.

Also participating today are several mothers from around the country:

- Virginia - mother of a recently diagnosed 15-year-old boy (Complex 1,3 & 4).
- San Diego, CA - mother of a young woman beginning college in her mid-20s who carries a diagnosis of "suspected Mito" after searching for a diagnosis for 10 years.
- New York - Mother of a newly diagnosed 12-year-old boy.
- Boston, MA - Mother of a 17-year-old daughter with "suspected Mito" who has been ill for 3 years. This mom has found both the Mito Cocktail as well as the advice in Cristy's book to be very helpful for her child.
- Mother of a 20-year-old who has been diagnosed since she was 1 1/2 years old and has done very well due to early diagnosis.

**Dr. Schreiber's Study:** Dr. Schreiber has been at Tufts for over 20 years, working with many young adults with symptoms and/or issues that no one else could understand. When Dr. Mark Korson joined the Tufts team, working with metabolic disorders, the two doctors worked together, caring for many Mito patients who were often young people struggling to live their lives, go to school, and make the transition from childhood to teenage years to adulthood. Despite Mito presentations being diverse and heterogenous, meaning having a wide range of symptoms and presentations, Dr. Schreiber noted an emerging pattern of struggle and difficulty in patients' lives related to cognitive functions. Schreiber conducted a study to pull together these observations in children aged 13 - 21 years, focusing on executive function and adaptive skill in teens and young adults. Having observed patients over the years, a behavioral rating scale seemed like the most reliable way to study this issue as the actual symptoms are so diverse in the mito population.

As Dr. Schreiber and Dr. Korson's patients reached 17 - 18 years of age, the typical transition issues common to all teenagers became an even bigger problem for the teens

with Mito. Because of the advances in care of Mito patients, these teens were now able to make the transition from high school to college, but facing that transition presented a great challenge. Normal issues of independence and autonomy are heightened for Mito patients. Thoughtful planning is needed by all for this transition to go smoothly, or as smoothly as can be expected with the transition from teen to adult.

### **Terminology**

**Transition:** The move from one place to another, from a more structured environment where one is taken care of to a less structured environment where one must rely more on one's self and one's own resources. Transition is a normal developmental stage. The person becomes his or her own organizer and must own up to the consequences of action or inaction. Teens in general are not proficient at this step, and teens with Mito are not any better. In fact, those with Mito have probably had even less opportunity to plan their own lives and make their own decisions. Teens who do not believe that they need any help tend to have the most difficulty.

**Executive Function (2parts):** **1.Cognitive** - includes organizational skills, self-motivation (i.e., checking a paper for spelling errors), working memory (i.e., ability to perform mental arithmetic, or starting a project taking a break and then returning right where you left off), goal "directedness," and the ability to initiate new tasks. **2.Behavior** - the ability to resist impulses, to recognize the consequences of one's actions and to transition from one task to another easily.

**Adaptive Skills:** The ability to adjust to changes in the environment, to use available help, to anticipate consequences of actions, and to plan accordingly.

Common challenges that teens with Mitochondrial Disease face:

- Acknowledging Vulnerability - Especially difficult for any teen who does not want to "need" other people nor be different than peers. Teens need to acknowledge, "OK, I have some issues," then look at how to face them in light of how to begin a college career, for example. These decisions require maturity and often involve much trial and error. This transition stage is a huge developmental change for teens.
- Understanding Consequences - Challenging for any teenager to plan for and understand the consequences of their actions.
- Generating Realistic Plans - Teens with Mito need to realistically look at what they have been able to do and what they might be able to do moving forward. A teen who has not had enough energy to put in a regular high school day every day likely will not be successful carrying 5 courses in college. Individual adaptations are vital to success.

**The Neuro Psych Evaluation:** Based on a multiple standardized paper and pencil tests that, along with a thorough history, can help map the strengths and weaknesses of a child/teen.

This evaluation is often used by schools to make needed academic accommodations. This "mapping" of strengths is very important because it allows planners to use a child's strengths as part of the accommodation. For example, the use of a reading support

system called Kurzweil, “reads” any digital text to the student and offers audio and visual reinforcement of assignments. (<https://www.kurzweilededu.com/default.html>) Reading software can be more important as children move forward in their education, as independent required reading increases. Eyes require a large amount of energy and reducing that energy drain and conserving energy through the use of a computer programs and technology can be very helpful. Dragon Naturally Speaking is another software package that is especially useful, allowing a user to “type” by simply speaking to a computer or other device.

The Neuro Psych evaluation includes a discussion of if and/or where changes might be expected over time. The evaluation provides a baseline for both tracking and planning for the future. Often times, students themselves can tell when there is a change in their status or abilities, but the neurological evaluation provides the map and baseline. The role of a neurological psychologist on the team is to be aware of all these cognitive and behavioral issues, recommending needed accommodations in the academic setting. Education plans for those with Mito can have both a medical component and a neuro psych component. Because fatigue is a huge part of Mito and affects cognition, the neuro psych evaluation is essential. Neuro psych testing is respected by schools, helping establish the need for both accommodations and support. The evaluation can help a teen better understand his or her own needs. Dr. Schreiber states, "Dr. Korson's education plans are a work of art."

**Mito Issues that impact Dr.Schreiber's Study:** Mitochondrial disease is known to commonly present with many and varied symptoms, called heterogeneous symptomology. There is agreement, however, on two basic facts: 1. The impact of fatigue on all Mito patients. Fatigue is a huge part of the disease but can vary in intensity from one day to the next. 2. General executive functioning which consistently affects memory, problem solving, and goal orientation. Symptoms then are dependent on both the individual person and the variability of the disease. Mito is not like PKU, where the cause is a specific enzyme that can be isolated and treated. With Mito, no luxury of a single cause exists, so the treatment, and even tracking of the disease, is much more complex.

**The Study:** The full text of Dr. Schreiber's article may be found at your local university or hospital library.

*[The abstract can be found on pubmed.gov.](#)*

Dr. Schreiber recruited her subjects from the MitoAction website. Twenty (20) people from across the US responded and 70% of these respondents were in the final study. For a rare disease, having 14 participants is considered good. Parents and students were asked to complete two forms: (1) Behavior Rating of Individual Executive Function and (2) Behavioral assessment evaluating anxiety, depression, self-esteem and social skills to assess how parents and students viewed themselves, similarly or dissimilarly. Schreiber's study found that students did indeed view themselves differently than their parents. Parents believed that any variation in child's behavior/performance was due to or related to their health and physical condition. The students, however, felt that how

they felt about school at a particular time, positive or negative, determined how things were going. If things at school were not going well, for example, then they felt anxious or depressed.

**What can we do to help students and parents in this transitional stage?** Planning ahead is vital to success. The social environment, especially of college, is very different from high school. For example, if a student will be living in the dorms, a single room may be needed to ensure that there is enough good sleep time. Socializing will have to be monitored, or the student will get too tired. Finding a balance in new setting is mainly a trial and error procedure, but students need to be able to pace themselves. Parents often struggle with these issues because it can be difficult to figure out what is just "normal" fatigue and what is due to the Mito. Dysautonomia and fluctuations in energy are hard to stay on top of and are very unpredictable.

Parents can stress that their child needs to be an individual and do what they are able to do, not necessarily what his/her peers are doing. Visit various schools to determine which one is the best fit and visit the Office for Disabilities. Try to determine what a typical day would be like and anticipate what life would be like at this school. Sometimes students need to start slowly by living at home first and only taking a few courses, building up to a more regular schedule and student life. Some students like to start full force then pull back when it proves to be too much, but this method may not be best since it may denote failure. Starting small and building successes may be the best way to proceed. Students should feel good about what they are accomplishing. Students should try to find someone in their field of study and shadow that person for a day, providing the student with Mito a realistic view of what it is like to take a particular course or major, and also allowing time to reconsider a course of study or choice of major. Students without Mito often change majors. Remaining flexible and willing to adjust plans is key.

**Depression:** Determining what kind of depression the teens is facing is important. Anyone with a long-term struggle or a chronic disease, looking to what might or might not be in their future, will have depression secondary to these life events. Chronical illness can be demoralizing. Mental health issues, however, can also be neurobiological due to decreased energy and directly related to the Mito diagnosis. New research is looking at this kind of depression and anxiety. The exact connection is not clear, but it appears that this depression and anxiety are more than simply situational. The brain is being affected, and there may be an actual chemical component. This depression should be treated differently, taken seriously, and managed medically.

**Fatigue:** Dr. Schreiber noted that all parents mentioned that fatigue was the most prominent and most unpredictable issue. Unfortunately, living with Mito includes the inability to predict energy levels. Therefore, fatigue should be treated like low blood sugar in a diabetic - a symptom of the disease that needs to be consistently addressed and treated.

**Kirste Casel**, Educational Consultant to MitoAction, is an educational expert and has two adult children with mitochondrial disease. Kirsten confirms many of the insights gained from Dr. Schreiber's study in real life, noting that executive function does change when there is an illness. Setbacks can be due to a virus, some other illness, or just severe fatigue. Any highly emotional state, which uses more energy, can also set up fatigue. For example, a student who found that she was admitted to college was so overjoyed and emotional, that her energy level was totally depleted right after this highly emotional experience.

College is a big change from the routine of high school, and taking a reduced course load and asking for more time when needed can help. A special parking space may also be needed to conserve energy getting to and from classes. All of the energy conserving that was implemented as part of high school needs to be looked at again in terms of the college plan.

Kirsten highly recommends the neurological psych testing because it helps point out the specific areas where help and adjustments may be needed. Several callers agreed, adding that not just having extra time for a test, but taking tests in certain areas of the school made a difference. Dragon Naturally Speaking use to limit typing also made a world of difference for a student. Another caller noted how the neuro psych evaluation helped validate which executive function skills were affected, and adaptations based on the that evaluation, along with the adoption of the Mito cocktail, has helped with education.

Insurance payment for neuro psych evaluations can be an issue. Kirsten suggested that parents should request the neuro psych evaluation as part of the IEP while the student is in high school (or earlier). The primary care doctor may request this testing, but the school will give the eval. If not happy with the school evaluation, or disagree with the results, the eval can be re-administered by a third party, again with a letter from your physician. Health insurance should cover the expense. For more information about neurological psych evaluations, go to the MitoAction site, and under the School and Legal Advocate tab, select a sample IEP or a .pdf file for neurological psych testing. Insurance is more likely to cover accommodations when physiological changes affect executive function (i.e., ability to retain information). The fatigue of mitochondrial disease is a physiological reason to cover testing. Insurance will often deny coverage if the accommodation is for educational reasons only.

Justification for a neuro psych evaluation is at times difficult, but becomes easier with a physician's support. The physician can be your pediatrician, neurologist or PCP and does not have to be a Mito doctor. Asking the doctor to be a partner also means that he or she won't have to actually develop the plan, so this step can save the doctor time. A caller noted her own daughter's struggles with this transition from high school to college. Her daughter was not able to make that transition right out of high school, feeling that she "could not do it." The young adult has built self esteem since then, has received more treatment for physiological symptoms (i.e., Mito cocktail), and now at age 20, has applied to college. Issues faced now are social: "How will I fit in? I'm older

than other students." These real issues make this transition from teen years into adulthood so difficult, not just for those with Mito, but also for all.

A typical college plan for someone with Mito is not 4 years, but realistically is 5 or 6 years. The required course load can be spread over summers and stretched out. Set realistic expectations from the beginning. Expect ongoing trial and error when devising a plan. For example, an 8 am class for a freshmen student with Mito probably won't work, so work with the college early on to plan for these things.

Another caller asked how to determine whether academic progress deteriorations is caused by mitochondrial disease or by something else. There is no easy answer to this question. The first step is to get others involved to assess the student medically and psychologically. Isolation is not good for parent or child and can lead to a downward spiral. Keep trying and use different resources as needed. Remember how difficult and overwhelming this transition stage can be.

Next Step is a Mito group for older teens who are experiencing this transition phase of their life, offering activities, retreats, and a connection with others with whom they can relate. Online chat groups and MitoAction's Facebook page where teens can find support can be very therapeutic. For teens 15 years and younger, Camp Korey in Seattle, Washington offers a way to find those therapeutic connections and support. MitoAction helps fund children so they can go for one week in the summer to meet other children with Mito.

### **How do you get teens/young adults to accomplish their goals themselves?**

Consider that it will take longer than 4 years to complete a Bachelor's Degree.

- Encourage flexibility in taking courses in college.
- Begin study at local college or community college and move on to a bigger institution when successful and ready for a bigger challenge.
- Don't just survive Mito, live your life!
- When attending a university event, plan ahead to reduce energy expenditure before hand.
- Do not be afraid to speak with an accommodation specialist on campus. Be creative and look for alternative solutions. Asking for help cannot hurt as the worst thing that can happens is someone will say, "no."
- Adult children should make decisions and set their own goals (it is hard for parents to let go).

**Conclusion:** Dr. Schreiber notes how impressed she has been with both the courage of Mito families, as well as their willingness to deal with so much uncertainty in their lives. The need to continue to define the issues that teens and young adults face in life remains. Creating strong, yet somewhat flexible, supports for teens as they transition to adulthood is vital. Mito patients of any age should not feel alone.

Submitted by Joanne M. Turco, RN, MS Edited 5/18/15 MBH, RN, MSN