Categories

TOC - Accessible
Adult Patients
Education
Genetics
Legal Advocacy
Living with Mito
Medical
- Autism
- Coordinated Care
- Developmental Delay
- Diagnosis
- Dysautonomia
- Mito Cocktail/Supplements
- Mito Minutes
- Pain
- Symptoms
- Treatment

Meeting Announcements
News
Our Stories
POV
Registered-Secure Support

Publications

Symptom Guide for Clinicians
Children's G-tube Book
Patient and Family Handbook
Teleconferences
Media
Podcasts in Itunes
Press Room
Videos

Find a support group

Find A Support Group

Donate

Show your support >

In honor or memory of a loved one >

Mitochondrial Donation (Or 3-Embryo Babies)

By Cristy Balcells on 02/16/2015

Preventing mitochondrial disease in future generations: New techniques may stop the transmission of faulty DNA from mother to child.

- **Cristy Balcells RN MSN**

For patients and parents of children suffering from mitochondrial disorders, there has been great interest in the recent legislation in the UK that would allow women to avoid passing on mitochondrial DNA (mtDNA) mutations to their children and to future generations through an IVF technique called “mitochondrial donation.” Although the concept of mitochondrial donation has been studied in animal models in both the US and Europe for some time, the 2015 vote in Britain’s House of Commons is significant because it implies that, if passed by the House of Lords (debate on Feb. 24), the UK would be the first country in the world that would offer a viable option for women with maternally inherited mitochondrial disease to have biological children without passing on the disease-causing genetic defect.

In every cell, there are a number of mitochondria that make energy for the cell. Mitochondria have their own DNA, called the mtDNA, which resides within the mitochondria themselves. In addition, in every cell there is a nucleus that contains the nuclear DNA (nDNA), which by and large determines who we are and what we look like. Inherited mitochondrial disease can be caused by genetic mutations in either the mtDNA and/or nDNA that result in abnormally functioning mitochondria and, in some cases, devastating symptoms of mitochondrial disease. Importantly, mtDNA is only inherited from the mother while both parents contribute their nDNA to their children. Mutated mtDNA causes about 25-30% of all cases of mitochondrial disease, which includes some of the most common mtDNA disorders (all of which fall under the umbrella of mitochondrial disease), such as LHON, MELAS, MERRF, and Kearns-Sayre Syndrome.

Mitochondrial donation prevents all mtDNA (including the faulty mtDNA mutations) from being passed on while allowing all of the other genetic material from the mother to stay intact during fertilization. On a basic level, mitochondrial donation works by keeping the *mother’s* nuclear DNA and utilizing a healthy*donor’s* egg that has healthy mitochondria and mtDNA. The technique involves implanting the mother’s nucleus into a donor egg by using advanced IVF techniques. In this way, mothers can have children who have all of their parent’s genetic traits while eliminating inherited mitochondrial defects. Further, this process would not only eliminate devastating mitochondrial disorders for these children, but would also have the potential to eliminate maternally inherited mitochondrial diseases from future generations.

One of the scientists behind the technique, Dr. Doug Turnbull, and his colleagues at the Wellcome Trust Centre for Mitochondrial Research at Newcastle University in the UK are well-known and respected around the world for leading advances in diagnosis and treatment of mitochondrial disorders. When developing the technique with colleagues Dr. Mary Herbert and Dr. Alison Murdoch, Dr. Turnbull approached

Latest Updates

- Mitochondrial Donation (Or 3-Embryo Babies)
- MitoAction’s Statement on Nationwide Closures of Mitochondrial Disease Clinics
- Stealth BT mitochondrial myopathy trial
- 2015 Mito Town Meeting
- Complex Medical Needs and Medical Child Abuse Accusations

More

Monthly Archive

- February 2015
- January 2015
- December 2014
- November 2014
- October 2014
- September 2014
- August 2014
- July 2014
- April 2014
- February 2014

more

Recent Comments

- Is there a reason that you neglected to mention the hugely deleterious effects of fluoroquinolone antibiotics (cipro/ciprofloxacin,... *Read More*
- Those with mitochondrial damage that is not one of the identified hereditary diseases should look through their medical records to see if... *Read More*
- Re: Lactate/Pyruvate ratio -- Each lab has its own reference range. I usually only consider the ratio if the lactate is abnormal.Re: Fatty... *Read More*
- The Seahorse is not ready for prime time yet but see papers by Victor M. Darley-Usmar, PhD at University of Alabama. At this point I... *Read More*
- We think that the levels needed in the blood just cannot be absorbed through the gut. I recommend giving the injections after the child... *Read More*

more

the problem while thinking of the many children, adults, and families who suffer from the devastating impact of mitochondrial disease. Says Dr. Turnbull, “We decided to try and develop this IVF technique to prevent the transmission of mitochondrial DNA disease after one of our patients lost 7 children. These IVF techniques will give new hope to families and more reproductive choice to mothers who carry these mtDNA mutations.”

Members from MitoAction’s Medical Advisory Board (www.MitoAction.org) are by and large supportive of the technology. Dr. Katherine Sims, Director of the Neurogenetics Program at Boston’s Massachusetts General Hospital, believes this technique holds great promise for women of childbearing age with mtDNA mutations, but cautions that “careful study and observation/assessment for potential complications of this sophisticated protocol for separation of nuclear DNA and mitochondrial DNA will remain critical as use of these techniques goes forward in women.” Criticism of mitochondrial donation generally has not stemmed from concern about the mechanics of the technique, but more from an ethical fear of manipulating our DNA in general and “designing babies.” Most of the negative responses to mitochondrial donation, both in the public point of view and in focus groups held by Britain’s Human Fertilisation and Embryology Authority (www.hfea.gov.uk), involve spiritual or ethical protests about manipulating inheritance traits in children in general. It is important to recognize that in the UK all fertility treatment is carefully regulated and these new IVF techniques would be limited to prevent transmission of mtDNA disease.

However, for the worldwide community of affected families and the physicians who care for them, mitochondrial donation holds great promise and hope to prevent transmission of mtDNA diseases. “For families who have long dealt with the devastating recurrence risks and burden of multiple affected children and family members associated with mtDNA mutations causing a host of incurable diseases, this breakthrough offers them the hope of healthy children and one way to eradicate a number of these non-treatable, progressive, fatal diseases,” says Fran Kendall MD, a geneticist and mitochondrial medicine expert affiliated with the University of Georgia and owner of VMP Genetics in Atlanta, GA. “While these scientific advances are not immune to greater discussions involving the ethics and morals of ‘genetic engineering,’ the positive aspects provided to this long-suffering patient population outweighs those issues, in my opinion.”

Mitochondrial disorders are thought to be more common than cystic fibrosis and more deadly than pediatric cancer in children, with an estimated incidence of about 1 in 2,000 people. Symptoms of mitochondrial disease are progressive and, at this time, science offers little in terms of treatment or a cure. Dr. Amel Karaa from Massachusetts General Hospital provides care for many adults with mtDNA diseases. “While many efforts are underway to find a treatment and a cure for patients with mitochondrial disease, progress has been slow. Affected mothers would have the choice of conceiving a healthy child without worrying about the devastating effects an mtDNA mutation could have on their future. It is already hard enough for the families to have an affected parent; it is unbearable to watch a child grow knowing that he/she carries a fatal genetic defect that can reveal itself and progress unexpectedly. Women all over the world have the opportunity to carry a pregnancy free of chromosomal abnormalities and gene defects by using prenatal testing and preimplantation diagnostics. It is time for the women with mitochondrial disease to have that same opportunity.”

For more information:

Newcastle Wellcome Trust http://www.newcastle-mitochondria.com/

NAMDC (North American Mitochondrial Disease Consortium) online survey http://www.rarediseasesnetwork.org/namdc/

Mitochondrial Donation on HEFA.gov.uk http://www.hfea.gov.uk/6896.html

MitoAction: Genetics http://www.mitoaction.org/blog/genetics-mitochondrial-disease

NEJM Editorial http://www.nejm.org/doi/full/10.1056/NEJMc1500960

BBC News http://www.bbc.com/news/health-31069173

The Lily Foundation http://www.thelilyfoundation.org.uk/

United Mitochondrial Disease Foundation
http://www.umdf.org/site/c.8qKOJ0MvF7LUG/b.9166823/k.2E25/Mitochondrial_Replacement_Therapy.htm#Position

Genetics
News
Treatment
POV