In this issue:
Read about what exemplary AEPhi women are doing
Alissa Hess Marino (Phi Phi) met her husband, Chris, when the two were undergrads at SUNY New Paltz. They married in June 2001 and moved to Long Island, NY. In August 2003 their daughter Danielle was born – in the middle of the East Coast blackout – Alissa remembers. The two sensed something was wrong with their baby but didn't receive an official diagnosis until she was 18 months old. Danielle had Mucolipidosis Type IV (ML4). “Symptoms are similar to cerebral palsy. We went to a geneticist and they didn't even know about ML4. An ophthalmologist ultimately gave us the diagnosis,” Alissa says.

First identified as a genetic disease in 1974, ML4 causes mental retardation, retinal degeneration and results in a limited lifespan. There is no treatment, no cure. The Marinos learned that ML4 is an inherited autosomal recessive disease. If both parents are carriers of the gene, their child has a 25 per cent chance of inheriting the disease. It is so rare that many doctors don’t even know about it. “Children with ML4 typically begin to exhibit symptoms during their first year of life. The disease causes clouding of the corneas and developmental delays,” explains Alissa. The carrier rate in Ashkenazi Jews is one in 100.

One in four Ashkenazi Jews is a carrier for at least one of 19 different genetic diseases which can strike in childhood, have no cure, require significant medical intervention, and, for some, lead to an early death. Sephardic Jews are at risk for carrying over 20 disease genes common in that population. “I had no idea there were Jewish genetic diseases other than Tay Sachs for which I was tested back in 2003,” Alissa says. Ironically while Alissa is Jewish, her husband Chris is only a quarter Jewish.

Like all parents faced with the devastating diagnosis that their child has a rare disease, the Marinos sought support. They are part of a very small, private Facebook group – with other families whose children also have ML4. “We chat about different things, share pictures and see videos,” says Alissa. Danielle is now 11, uses a wheelchair and can't walk without help. “She can't talk and ML4 also causes neurological, gross and fine motor issues,” she adds.

“Kids with ML4 are very social and not really interested in toys. If my daughter had someone to hang out with her all day she would be happy. Thankfully she doesn't seem to be in pain.” Danielle attends a private school for special needs children. “She gets physical therapy –everything is done at school,” Alissa explains. “The goal is to try and help her to advance as much as possible to do whatever she can do.”

Another family’s experience with ML4 may help to change the outcome for families like the Marinos. When Randy and Carolyn Gold’s daughter, Eden, was diagnosed with ML4 in August 2009 – also at 18 months old – they decided to help change the fate of other families. The Atlanta couple was instrumental in the creation of JScreen – a simple genetic screening program. A pilot program was launched in 2010. Initially it was called Atlanta Jewish Genes because of its Georgia location. With funding from the Marcus Foundation, JScreen was launched nationally in September 2013.
in partnership with Emory University’s Medical School. “JScreen provides an at home saliva testing kit that screens for a panel of about 80 genetic diseases including 40 that are more common in the Ashkenazi and Sephardic Jewish populations,” explains Karen Arnovitz Grinzaid (Omicron), senior director of Outreach Initiatives at JScreen and co-director of Emory’s Gene Screen Program.

Hillary Kener, Outreach Coordinator for the JScreen Program, explains how the program works. “It’s very easy – log on to our website and request a kit. You simply watch an informative three minute educational video to know what to expect. Fill out a form with your background, family history, how many grandparents of Ashkenazic and Sephardic descent and enter your physician’s information. Upon completion of the form – we send out a six-inch by six-inch saliva kit. Follow the directions, zip up the envelope with prepaid postage and drop it in the mail. The lab receives it and results are available in about a month and we schedule a genetic counseling session over the phone through HIPAA-compliant video-conferencing. JScreen also sends information and results directly to your physician.”

“Regardless where you live, this test is easy, convenient and affordable. It’s no fuss, no muss, it brings screening to your doorstep. The out of pocket cost is $99 with private health insurance. The actual cost is a lot higher but JScreen subsidizes the testing to make it affordable for anyone and everyone,” adds Karen.

Both Karen and Hillary emphasize the importance of people getting screened before they have their families. “Whether you are single, married, engaged, just adding to your families, these tests are crucial. We recommend that whether you are an Ashkenazi or a Sephardic Jew, even if one parent is not Jewish, that you take the test,” Karen says. “The beauty of JScreen is that we are accessible 24/7 whether you live in North Dakota or Florida you have the same access as everyone,” Hillary adds. JScreen has screened over 2000 people from 41 states since its launch. Most surprising? One in four individuals screened are identified as carriers for at least one disease. Hillary encourages future grandparents to order a JScreen gift certificate to help loved ones plan for a healthy future.

JScreen could have changed our story, Alissa contends. But she cautions that even with testing, a child can still be born with autism or other developmental delays. “But if you do what you can to prepare yourself to be knowledgeable, it can make a huge difference. We left the genetic testing up to our doctor. Today it’s different – there’s so much more understanding and options,” she says.

“With JScreen you can do a saliva test – so why wouldn’t you? It’s a no brainer if you are thinking about having kids someday. Even if you are in college and not planning a family right now – it’s good to be aware of what could be a possibility for your future, for your future children’s future. There’s no reason not to get tested. Don’t think it couldn’t happen to you,” she cautions. Alissa hopes that by sharing her story, it will help others. “I hope my story brings awareness to people before it’s too late. Learn what’s in your family’s background. Everyone should – even with no Jewish history.”

**Are you Ashkenazic or Sephardic?**

**Ashkenazic** Jews are the Jews of France, Germany and Eastern Europe and their descendants. Most American Jews today are Ashkenazim, descended from Jews who emigrated from Germany and Eastern Europe from the mid 1800s to the early 1900s.

**Sephardic** Jews are the Jews of Spain, Portugal, North Africa and the Middle East and their descendants.
What is JScreen?

Based out of Emory University's Department of Human Genetics, JScreen is a nonprofit, community-based health initiative dedicated to providing easy, at-home genetic screening for over 80 different genetic diseases, including those that are more common in Ashkenazi Jews.

JScreen's process is easy, from start to finish:

▲ Order your JScreen at-home saliva-based genetic testing kit from www.jscreen.org. Cost of the test is $99 with private health insurance.

▲ Receive your test kit in the mail. Unbox and, following the directions, fill the tube with your saliva sample.

▲ Send your sample to the laboratory in the included pre-paid mailer.

▲ Receive your results in approximately 4 weeks. If your results indicate that you are a carrier for any of the diseases, a JScreen genetic counselor will contact you to discuss your results.

Why it matters:

• 1 in every 4 people of Ashkenazi Jewish descent is a carrier for at least one of the diseases that is more common in that population. Carriers are healthy and do not have symptoms.

• When both parents are carriers for the same condition, each of their children faces a 25% risk for actually having the disease.

Some of the diseases JScreen can test for are:

• Tay-Sachs Disease
• Cystic Fibrosis
• Usher Syndrome Types I & III
• Canavan Disease
• Gaucher Disease
• Spinal Muscular Atrophy
• Familial Dysautonomia
• Nemaline Myopathy
• Lipoamide Dehydrogenase Deficiency
• Bloom Syndrome
• Walker Warburg Syndrome
• Fanconi Anemia
• Mucolipidosis IV
• Niemann Pick Disease
• Joubert Syndrome
• ABCC8 Hyperinsulinism
• Maple Syrup Urine Disease
• Glycogen Storage Disease
• Type 1A

A new Second Avenue program focusing on JScreen will be made available to chapters and colonies early next year. The program will include a video about the Marino family, discuss genetics and why people are not getting screened, and the solution that JScreen offers.