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Genetic miracle work

Art and Wendy Kessler with their son, Benjamin.



CAPTION: Given his past, Art Kessler is overjoyed about the miracle of his present, and elated at the promise of his future all thanks to scientific breakthroughs in Jewish genetic disorders.

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Special to JUF News

Kessler, diagnosed at age 12 with early-onset dystonia a genetic brain disease that causes involuntary muscle movements and forces the body into twisted, painful postures refused to father a child at risk of having the disease and the condition he described as a nightmare.

Now, because of the discovery of the DYT1 gene, genetic and prenatal testing, and a groundbreaking procedure called preimplantation genetic diagnosis (PGD), Kessler and his wife, Wendy, are the parents of dystonia-free Benjamin, born Sept. 9 weighing 7 pounds, 2 ounces. Benjamin is the first child ever to be born using PGD to prevent another life from being burdened with dystonia.

Kessler hopes that Benjamin will be an encouragement to other couples facing a similar family decision, and Karen Litwack, director of the Chicago Center for Jewish Genetic Disorders, hopes Benjamin will be a reminder for couples planning to marry to be screened for Jewish genetic disorders and take advantage of resources available locally.

When scientists, after a 20-year search, identified the gene for early-onset dystonia and named it the DYT1 gene in 1997, there was no change in Kessler's life. Married in 2001, Art and Wendy planned to remain childless and talked about adoption until they learned of PGD, a form of in vitro fertilization.

Although PGD does not guarantee pregnancy, when a successful pregnancy does occur, it allows most couples to be almost 100 percent sure that their child will be born without the abnormal gene for early-onset dystonia. Currently, early-onset dystonia, which affects about one-third of the 350,000 North Americans who suffer with dystonia, is the only form of dystonia to which PGD is applicable.

Through the Reproductive Genetics Institute in Chicago, Wendy's eggs were fertilized outside her body and the conceptions were screened for the DYT1 mutation before they became viable embryos. Genetic screening is possible when the conceptions grow to the size of eight cells. Conceptions free of the mutation were selected and returned to Wendy's womb. Wendy didn't become pregnant until the second attempt.

Kessler, a board member of the Dystonia Medical Research Foundation, is quick to point out that PGD is not for everyone. "It's expensive. It's still hard to get covered by insurance," he explains. The two procedures the first unsuccessful, the second resulting in Benjamin cost the Kesslers \$20,000 and were not covered by insurance, but Benjamin's father, 36, isn't putting a price tag on what the end of dystonia in his family means.

Wendy and I are elated. For my immediate family, Benjamin means this is the end of dystonia in our family. It's great! This is a huge relief for us, says Kessler. He describes Benjamin as a typical baby he smiles, he blows bubbles, he laughs. He's a happy kid except when he's hungry.

Kessler, whose gait and posture are affected by the disease but otherwise considers himself lucky to have lived a pretty normal life, cautions that the scientific breakthroughs that have given Benjamin a dystonia-free life are not the ultimate achievement when it comes to dystonia. This is not a treatment or a cure. There is still a huge need for scientific research. People who want to give money need to give to the Center for Jewish Genetic Disorders or the Dystonia Foundation.

Dystonia is one of several Jewish genetic disorders including Bloom syndrome, Canavan disease, Familial Dysautonomia, Fanconi anemia (Type C), Gaucher disease (Type 1), Mucopolysaccharidosis IV, Niemann-Pick disease (Type A), Tay-Sachs disease, cystic fibrosis (CF), and Beta-thalassemia.

Because everyone carries six to eight disease-producing genes, which would be harmful if passed on to children by both the mother and father, the Chicago Board of Rabbis and the Chicago Center for Jewish Genetic Disorders remind couples planning to marry that knowledge is the key to prevention. The two organizations urge couples to talk with their physicians and be screened before starting a family.

The Chicago Center for Jewish Genetic Disorders is a collaborative effort of the Jewish Federation of Metropolitan Chicago, Children's Memorial Hospital, and the Illinois Jewish Genetic Disorders Committee. For additional information, call (312) 357-4718, e-mail jewishgeneticsctr@juf.org, or visit www.jewishgenetics.org.