THE WALL STREET JOURNAL.

http://www.wsj.com/articles/SB10001424052702304458604577488842861558400

HEALTH & WELLNESS

Study Looks at Irish Risk for a Rare Fatal Disease

By AMY DOCKSER MARCUS

June 25, 2012 6:37 p.m. ET

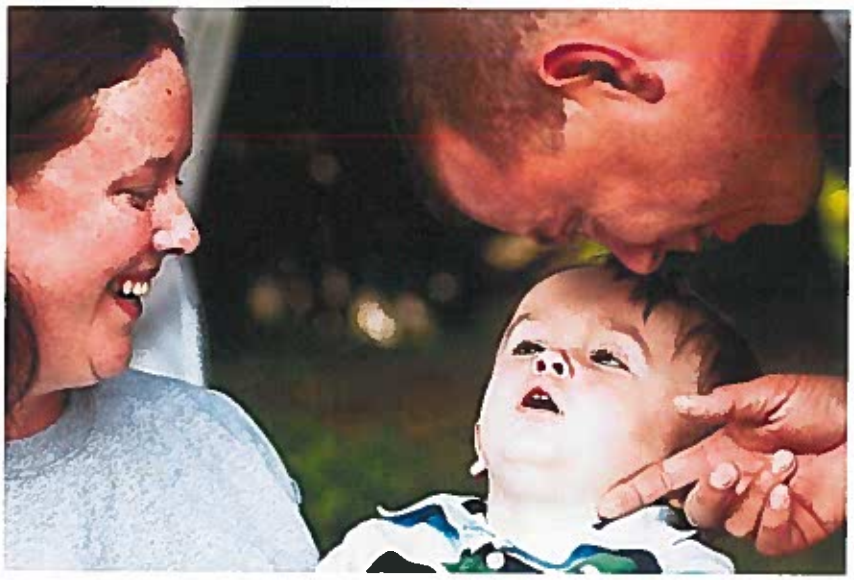
Tay-Sachs disease is generally known as a Jewish genetic disorder. Now, researchers are beginning to focus on another community at risk: people of Irish descent.

Tay-Sachs is a rare neurodegenerative disease that hits fewer than 30 children in North America a year. It is always fatal. Widespread testing to check if prospective parents are carriers of the Tay-Sachs gene has sharply diminished the disease's occurrence among Jews of Central and East European ancestry, the ethnic group long associated with Tay-Sachs. But not everyone knows that other groups, including Irish, French Canadians and Cajuns, also are vulnerable.

"It is not an exclusively Jewish genetic disease," says Miriam Blitzer, professor and geneticist at the University of Maryland School of Medicine, who says most cases today involve non-Jews. "We have been trying to teach that for years."

Some parents of children with Tay-Sachs are helping to spread the word. Kathryn and Aaron Harney of Downingtown, Pa., handed out literature at an Irish cultural festival this month aiming to enroll participants in a new study to identify carriers of the disease. The Harneys' 2-year-old son, Nathan, who has Tay-Sachs, can no longer sit up, walk or crawl. The couple never heard of the disease before his diagnosis. "No one seemed to know it was Irish," says Ms. Harney. "Our doctor thought it was only a Jewish disease."

To pass the disease to a newborn, both parents must be genetic carriers. About 1 in 300 people in the general population are estimated to have a faulty gene that can lead to Tay-Sachs. That rate is much higher, about 1 in 30, for Jews of Central and Eastern



Kathryn and Aaron Harney, of Downington, Pa., play with 2-year-old Nathan, who has Tay-Sachs, a rare neurodegenerative disease. *SCOTT LEWIS FOR THE WALL STREET JOURNAL*

European descent and for some French Canadians and Cajuns. About 1 in 50 people of Irish descent are believed to be carriers, although the rate hasn't been scientifically proved.

If both partners are carriers, there is a 1 in 4 chance that a newborn will have Tay-Sachs. Couples may choose instead to adopt a child or try in vitro fertilization using preimplantation genetic diagnosis. They may also use a donated egg or sperm. People with Tay-Sachs lack a crucial enzyme, hexosaminidase A, needed to break down fatty substances. The fatty material accumulates, eventually destroying nerve cells in the brain and spinal cord.

MORE

Making Gene Mapping Part of Everyday Care

May 14, 2012

Families of Ill Children Try Drug Development

Dec. 27, 2011

A pilot study to identify carriers of the disease among people of Irish ancestry has been launched in Philadelphia by the Einstein Medical Center Philadelphia and the nonprofit National Tay-Sachs & Allied Diseases Association of Delaware Valley. The program aims to enroll 1,000 people with at least three of four grandparents who can trace Irish ancestry.

Researchers plan to measure levels of the enzyme in participants' blood to find carriers. These people will then have DNA testing. If already-known genetic mutations aren't found, then researchers will look for other

possible mutations that can lead to Tay-Sachs. Genetic mutations related to Tay-Sachs in the Irish community can be different than those found in Jews and other ethnic groups.

Nathan can no longer sit up, walk or crawl. *SCOTT LEWIS FOR THE WALL STREET JOURNAL*

At the Irish-American Festival at Penn's Landing earlier this month, vendors sold T-shirts and bands played Irish music. The Harneys, both of whom have grandparents of Irish ancestry, also were there, handing out pamphlets about the study and introducing people to their son, Nathan. "It was putting a face on Tay-Sachs," says Ms. Harney, who works at an auto-insurance company.

Another couple, Cathy and Jeff Mitchell, of Langhorne, Pa., sold shamrocks and souvenirs and talked about the Tay-Sachs study at Pennsylvania's Bucks County St. Patrick's Day parade in March. The Mitchell's son, Harrison, died from Tay-Sachs in 2010, shortly before his sixth birthday.

"I don't think many people of Irish descent realize the threat of this disease," says George Galloway, president of the parade committee.

Adele Schneider, director of clinical genetics at Einstein Medical Center Philadelphia and principal investigator of the current research, says the study came together after she saw three families of Irish descent who all had children with Tay-Sachs in the past few years. Organizers of the current study plan to expand their efforts to Irish communities in Boston and New York.

Dr. Schneider says that in Ireland people with a family history of Tay-Sachs may request testing, but routine screening for the disease isn't done. That is partly because the carrier rate for Tay-Sachs in the Irish population hasn't been scientifically proved, she says, adding that the current U.S. study is expected to provide data on the risk faced by

people of Irish ancestry.

Michael Kaback, emeritus professor of pediatrics and reproductive medicine at the University of California, San Diego School of Medicine, says trying to determine the frequency of carriers for Tay-Sachs among the Irish is likely to be complex. Sometimes people who volunteer for screening know of carriers in their family, which makes it harder to get accurate statistics, says Dr. Kaback, who helped drive screening in the Jewish community.

Another problem could be defining who is Irish. "What do you do with someone with one English, one Scottish, one Irish, and one American grandparent?" says Dr. Kaback. "Are they Irish or not Irish?"

Write to Amy Dockser Marcus at amy.marcus@wsj.com