Tay-Sachs disease not just a Cajun genetic disorder

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Recent publicity regarding a "Cajun" genetic disposition towards a fatal disease called Tay-Sachs, an inherited disorder which affects the neuromuscular systems of humans during infancy, may cause many area residents unnecessary concern.

Nancy Pousson is a resident of the nearby Acadia Parish community of Iota, where the numbers of those afflicted with the disease have increased at an alarming rate. She is trying to educate area residents about Tay-Sachs and trying to lessen fears among natives of the region who may be concerned over recent publicity regarding a "Cajun connection" to the inherited disease.

Those individuals who develop symptoms of the disease usually die by the age of six.

"Cajun heritage alone does not mean that native residents of Acadiana are necessarily at a higher risk of inheriting Tay-Sachs," Pousson said.

Dr. Emmanuel Shapira, a professor of pediatrics at Tulane University's medical school, used genetic markers in his research as a way to determine racial or ethnic related diseases due to intercultural marriages.

Pousson explained that continued research by Shapira seemed to indicate that Cajuns were at a no higher risk for Tay-Sachs or related neuromuscular disorders than any other culture which predominates a certain area.

Pousson referred to an article published by Tulane Medical Center from which Shapira explained, "There's a high frequency of intermarriage in the Cajun population" and Shapira explained that just such an intermarriage would cause a genetic disorder such as Tay-Sachs or its related disorders to appear, with what could seem like alarming frequency.

Information reported recently in Acadiana was primarily based on the continuing research of two professionals studying the fatal Tay-Sachs disease. Tay-Sachs is caused by the lack of a certain enzyme, that is necessary for the breakdown of a fatty compound in the brain and nervous system in the human body.

Dr. Sudhir Sinha, president of GenTest Laboratories in Metairie, recently discovered a specific genetic marking which identifies individuals with Cajun heritage. But, this genetic marking is also individualized in other ethnic or racial groups such as African-Americans, Caucasians, Asians and Hispanics.

The discovery of this genetic marking difference has been used in research related to criminal investigations by helping to narrow the number of suspects in a criminal case. Genetic markings are identified through examination of blood, semen or other bodily fluids which may be collected at a crime scene.

Tay-Sachs disease is only one of 50 inherited diseases related to the same enzyme deficiency, which can make the diagnosis of Tay-Sachs difficult, Pousson noted.

Frederick's Ataxia is another neuromuscular disease related to the same genetic deficiencies that cause Tay-Sachs.

For many years, this inherited trait has been linked to descendants of immigrants from Eastern European areas. Most were members of the Jewish faith, and by custom, married within their faith, remaining in the same area from birth until their death. Because of this, many descendants of Eastern Europeans, particularly members of the Jewish faith, were eventually found through research to have the genetic deficiency identified as Tay-Sachs.

Because of that, many Jewish couples today across the United States have had prenatal genetic screenings to determine the risks of conceiving a child who may inherit Tay-Sachs disease.

Research of the "Cajun connection" may be showing what could be considered a higher-than-normal occurrence of Tay-Sachs disease in Acadiana because of similar circumstances of generational ties to this region of Louisiana.

Pousson and her husband, who are not carriers of the Tay-Sachs gene, are parents of a daughter who died at the age of eight from a condition, which was never clearly diagnosed, but exhibited symptoms of Tay-Sachs. The Poussons are also parents of a 14-year-old daughter who has exhibited none of her younger sister's symptoms.

Pousson has maintained contact with the families in the area who have or have had children affected by the inherited genetic disorder or one of its numerous genetic mutations.

A common ancestor has been found in a few cases of the Iota residents who have children with Tay-Sachs or a related disorder.

"A German immigrant to the Acadia parish area who settled there in the 1700s, Johann Edelmeier, seems to be the European link to Tay-Sachs, appearing in the Iota area, according to information acquired by researchers at Tulane University Medical Center, especially Dr. Emmanuel Shapira," Pousson commented.

Edelmeier, Pousson explained, was born in a region of Germany where the rate of inheriting a genetic disease such
as Tay-Sachs was about 1 in 3,600.

Edelmeir may be the link to the cause of the appearance of Tay-Sachs disease in the predominantly Roman Catholic, Acadian ancestry area found in South Louisiana. This could have resulted from an interfaith or intercultural marriage at some point, which occurred in a region where Eastern European and Jewish ancestry is not common.

This would lead to the possibility that Tay-Sachs and its related genetic diseases, may have been transmitted to Edelmeir’s descendants, yet kept within the Acadiana area due to intercultural marriages by native residents.

During the early stages of his study on Tay-Sachs in July 1990, Shapira took blood samples from 230 volunteer residents of the area in and around Iota.

Of the 230 participants screened, 17 Tay-Sachs carriers were detected. Family members of known Tay-Sachs carriers were screened, and 12 out of those 93 carriers also were found to carry Tay-Sachs.

Finally, 147 individuals with no known family history of the disease were screened and only five of them were found to be carriers of the Tay-Sachs related trait.

Pousson joined with other parents of children who exhibited symptoms linked to Tay-Sachs, hoping to educate other Acadiana residents on genetically inherited diseases related to Tay-Sachs.

A Louisiana Chapter of the National Tay-Sachs and Allied Diseases Association provides an informational and emotional support resource for residents of Acadiana who want to know more about the disease “or simply have another individual to talk to who know(s) what it is like to deal with a genetic disease which often is not detected until symptoms appear,” Poussoni said.