

Health facts Q&A on Fabry disease

Q: What is Fabry disease?

A: Fabry disease is an inherited, progressive disease that occurs worldwide among all ethnic groups. The disease may lead to a shortened life span due to decreased kidney and heart functions. It occurs in about 1 in 40,000 males.

Fabry disease is an inherited lipid storage disease. It is triggered by an inherited defective gene located on the X chromosome, thus hampering the production of an important enzyme. Without the enzyme, Fabry disease interferes with the body's ability to break down lipids, causes damage to cells and organs, and results severe pain, decreased ability to sweat, hearing loss, red-purple skin lesions, stroke, as well as bowel, kidney, heart problems and shortened life span.

Q: What are the signs and symptoms?

A: Fabry disease symptoms range from sudden, severe pain attacks to numbness or burning in hands and feet, impaired ability to sweat or adjust to temperature changes, hearing loss or ringing in the ears and heart and kidney damage. The disease gets progressively worse with age.

Fabry Disease is easy misdiagnosed as conditions such as rheumatoid arthritis, irritable bowel syndrome, inflammatory bowel disease, growing pains in children, for example, and other types of nerve damage.

Q: What is the treatment?

A: Treatment of Fabry disease had traditionally involved symptom and pain management, cardiac intervention and dialysis. In the last eight years, specific enzyme therapies have been

developed, allowing for the replacement of the missing enzyme.

In fact, the Canadian Fabry Disease Initiative has recently been established to examine the progress of all Fabry Disease patients in Canada on enzyme replacement therapy. All patients in Canada with Fabry disease are eligible for enrolment in this study, which consists of the collection of clinical findings to learn more about the nature of this highly variable disease. Fabry patients who meet the criteria for treatment will be provided enzyme replacement therapy.

Q: Why are there so many cases of Fabry disease found in Nova Scotia?

A: Nova Scotia has a great number of patients with Fabry Disease because of what's called a founder effect. There was an individual who moved here 16 generations ago, and it's her descendants who have stayed in Nova Scotia that have this condition. The funding of enzyme replacement therapy in Nova Scotia means that patients are now seeing their symptoms, including bouts of fever and sensitivity to heat and cold, arrested or receding.

Fabry Disease can be diagnosed by a simple blood test to identify the defective gene. Nova Scotians who suspect they, or a family member may suffer Fabry disease should consult their family physician or call 877-998-9797 toll-free. For information on the Canadian Fabry Disease Initiative, see www.fabrycanada.com.

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Nova Scotians urged to participate in study of Fabry Disease Treatment with missing enzyme contributes to pain and symptom relief

(NC)—As a girl, Debbie Deveau listened as her high-school-aged brother moaned with pain. Being farm folk on Nova Scotia's south shore, her brother's symptoms weren't talked about; the suspicion being that he was 'malingering' to avoid chores. Occasionally, though, there was mention of Fabry Disease, a disorder no one knew much about. By her mid-twenties and married with children, Debbie discovered from an eye and blood examination that she and her sons Jason and Sean, aged one and two at the time, were afflicted with Fabry, the same disease her brother had been diagnosed with.

X-Linked Inheritance

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Signs and Symptoms

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Fabry Disease is easy misdiagnosed as conditions such as rheumatoid arthritis, irritable bowel syndrome, inflammatory bowel disease, growing pains in children, for example, and other types of nerve damage.

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enzyme therapies have been developed, allowing for the replacement of the missing enzyme.

Dr. Michael West, Chief of Nephrology, Dalhousie University, is Lead Investigator of the Canadian Fabry Disease Initiative which will examine the progress of all Fabry Disease patients in Canada on therapy.

According to Dr. West, "Nova Scotia has a great number of patients with Fabry Disease because of what's called a founder effect. There was an individual who moved here 16 generations ago, and it's her descendants who have stayed in Nova Scotia that have this condition."

Funding of enzyme replacement therapy in Nova Scotia means that Debbie, her sons and people like them are seeing their symptoms, including bouts of fever and sensitivity to heat and cold, arrested or receding.

Last summer was the first time Debbie can remember that her sons reported enjoying summer and were not stuck indoors avoiding the heat. Told she could expect general health benefits within five years, after two year's on ERT, Debbie already reports improved heart and kidney functioning.

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Ryan and Debbie Deveau