FABRY DISEASE…

Floyd Stockinger called me and stated that Chuck's wife has this disease and gets $2500 treatments for it to "extend her life" to 75 years of age. (She works at Anderson Trucking Company on 94)

Fab're

Told him to ask him what the treatments are doing for her as it is an X-linked DNA problem and thus the treatments must be for a specific problem.

E.g. Fatique- PROTEIN

Tinnitus = B-Complex and Vita-E (this would also be great for the heart)

Neuropathy – Vivix and/ or Alfalfa

Skin issues – Protein & Omegaguard

Just the general program would help her live at a more optimal health level!

http://www.medicalnewstoday.com/articles/263333.php

Fabry disease is a rare, inherited disease caused by the deficiency of an enzyme called alpha galactosidase A (a-GAL A), which is encoded by the GLA gene. In patients with Fabry disease, a-GAL A is either lacking or deficient. The disease is also known as Anderson-Fabry disease, alpha-galactosidase A deficiency, or angiokeratoma corporis diffusum.

A genetic fault (mutation) in the gene that controls a-GAL A causes a poor breakdown of lipids, which leads to a harmful accumulation of lipids - specifically globotriaosylceramide (GL-3) - in the cardiovascular system, autonomic nervous system, kidneys and eyes.

There are several lipid storage disorders; Fabry disease is the only X-linked (inherited) one.

Fabry disease is estimated to affect 1 in every 55,000 males. Many more males have the defective gene, but with no symptoms.

The mutated gene is carried on the mother's X chromosome, meaning there is a 50% chance she will pass it on to her sons, and a 50% risk her daughters will be carriers.

If females do have symptoms, they tend to be much milder than those seen in males. However, some females with Fabry disease may be affected severely. Young boys may have serious eye manifestations, particularly cloudiness of the cornea.

The accumulation of lipids, specifically GL-3, can lead to problems with arterial circulation and a higher risk of stroke or heart attack.

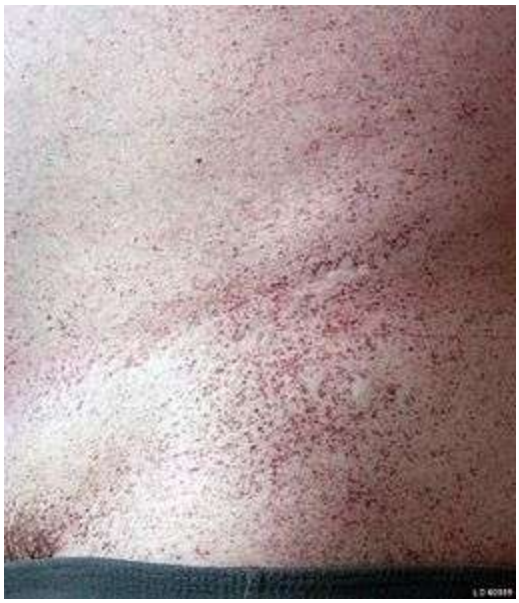
A study published in the *International Journal of Clinical Practice* found that Spanish patients with Fabry disease react differently to those in the rest of Europe.

What are the signs and symptoms of Fabry disease?

Fabry disease may be hard to detect because several of its signs and symptoms overlap those present in other diseases and conditions. Patients do not typically have all of the symptoms associated with Fabry, while others may develop them at different times throughout their lives.

It is important that the doctor finds out whether there is a family history of Fabry disease whenever it is suspected.

Fabry disease in males



Angiokeratoma

Signs and symptoms usually start during childhood.

- **Pain** - the child experiences episodes of discomfort and pain in the hands and feet (acroparesthesias). These can be triggered by stress, fatigue, fever, physical activity, and even changes in the weather.

- **Skin rash** - known as angiokeratoma, A dark, red, spotted skin rash appears. The rash is seen most densely between the navel (belly button) and the knees.

- **Hypohidrosis** - reduced ability to sweat.

- **Keratopathy** - clouding of the corneas. This does not affect vision

Fabry disease usually develops slowly over many years as GL-3 gradually accumulates. After the age of 30 years, patients may experience:

- **Gastrointestinal problems** - patients with Fabry disease commonly experience stomach problems, which may include nausea, vomiting, and diarrhea.

- **Cardiovascular problems** - years of GL-3 accumulation eventually result in damage to blood vessels that supply the heart, as well as damage to the heart itself. Problems include:

  - heart failure
  - arrhythmia
  - heart attack
  - faulty heart valves
  - enlarged heart

- **Renal (kidney) problems** - decades of GL-3 accumulation can lead to renal failure or renal insufficiency. Often, Fabry disease is first detected when the patient comes to the doctor with kidney problems.

- **CNS (central nervous system) disorders** - small blood vessels in the brain can become affected after years of GL-3 buildup. Patients are at a higher risk of having:

  - stroke
  - dizziness
  - numbness and tingling (pins and needles)
  - headaches
  - weakness
- **Hearing problems** - for the same reasons, GL-3 accumulation, the patient may gradually lose his hearing, or experience ringing in the ears (tinnitus).
- **Emotional and psychological problems** - living with the symptoms associated with Fabry disease, many of which get worse with time, causes anxiety, fear, and also depression. There may be a feeling of guilt if the disease is inherited by an offspring.

Fabry disease in females

Females, who are also carriers of the genetic mutation, generally either have no symptoms or much milder ones than males. However, a small percentage may suffer as much as their male counterparts.

In milder cases, symptoms usually appear during the patient's childhood/teen years, and may include skin rashes, corneal cloudiness, pain in the extremities, and hypohidrosis. There is also the possibility of chronic diarrhea and abdominal pain. As they get older the left side of the heart may become enlarged (left ventricular hypertrophy); there is also a possibility of heart-valve problems.

In more serious cases, the girl/woman has the same signs and symptoms as males with severe symptoms.

How is Fabry disease diagnosed?

The doctor may suspect Fabry disease if the patient has the associated signs and symptoms. If one of the patient's relatives is found to have the disease, the physician will order a blood test to measure a-GAL A activity.

Screening females for Fabry disease is not so simple, because the blood test can be misleading, due to the random nature of X-inactivation, i.e. the faulty gene may be switched off, so the enzyme a-GAL A won't be affected. A chromosomal analysis of the GLA gene is much more accurate than a blood test.

If excessive GL-3 accumulation is detected, a kidney biopsy may help.

Fabry disease is commonly misdiagnosed by pediatricians and internists.

What are the treatment options for Fabry disease?

Doctors say that prevention is the first level of treatment for patients with Fabry disease.

- **Pain and discomfort** - episodes of pain are nearly always linked to certain triggers, such as exposure to heat, temperature changes, sun exposure, exercise, and fever. The patient must learn to avoid these pain triggers.

  For patients with severe and frequent episodes of pain, the doctor may prescribe an anticonvulsant, such as carbamazepine (Tegretol, Tegretol XR , Equetro, Carbatrol) or diphenylhydantoin (Dilantin). They should be taken daily.
- **Enzyme replacement therapy (ERT)** - ERT is a medical treatment that replaces an enzyme which is either absent or deficient in patients. In Fabry disease patients' cases, the missing enzyme is alpha galactosidase A (a-GAL A).

  There are two recombinant GLA preparations for ERT on the market today: 1. agalsidase alfa (Replagal, Shire Human Genetic Therapies, Cambridge, MA, 0.2 mg/kg per infusion). 2. agalsidase beta (Fabrazyme, Genzyme Corporation, Cambridge, MA, 1 mg/kg per infusion).

  In the USA, only Fabrazyme is FDA approved. Genzyme Corp. writes on its website "The lowering of GL-3 suggests that Fabrazyme may improve how Fabry disease affects your body; however a relationship of lower GL-3 to specific signs and symptoms of Fabry disease has not been proven."

  ERT has one serious drawback - it is extremely expensive.

The other complications related to Fabry disease, such as skin, heart, kidney and psychological problems are treated separately as they occur, by specialist doctors, i.e. if it is a heart problem, the patient will be treated by a cardiologist.

What is the life expectancy for somebody with Fabry disease?

Patients who are diagnosed early and can receive treatments promptly have longer lifespans. According to the Genzyme Corporation, new treatments have lengthened life expectancy from 41 to 50 years.

Researchers published an article in *Genetics in Medicine* saying that "The life expectancy of males with Fabry disease was 58.2 years, compared with 74.7 years in the general population of the United States. The life expectancy of females with Fabry disease was 75.4 years, compared with 80.0 years in the United States."

http://www.ncbi.nlm.nih.gov/pubmed/19745746

Genet Med. 2009 Nov;11(11):790-6. doi: 10.1097/GIM.0b013e3181bb05bb.

Life expectancy and cause of death in males and females with Fabry disease: findings from the Fabry Registry.

Waldek S, Patel MR, Banikazemi M, Lemay R, Lee P.

Author information

Abstract

PURPOSE:

To evaluate life expectancy and cause of death among patients with Fabry disease, an X-linked lysosomal storage disorder.

METHODS:

Data from 2848 patients in the Fabry Registry were summarized using descriptive statistics. Life expectancy at birth was compared with that of the United States general population.

RESULTS:

As of August 2008, 75 of 1422 males and 12 of 1426 females in the Fabry Registry were reported to have died. The 87 deceased patients were diagnosed at a much older age than other patients in the Fabry Registry: median age at diagnosis was 40 vs. 24 years in males and 55 vs. 33 years in females. The life expectancy of males with Fabry disease was 58.2 years, compared with 74.7 years in the general population of the United States. The life expectancy of females with Fabry disease was 75.4 years, compared with 80.0 years in the United States general population. The most common cause of death among both genders was cardiovascular disease. Most (57%) patients who died of cardiovascular disease had previously received renal replacement therapy.

CONCLUSIONS:

Most deceased Fabry Registry patients exhibited serious cardiac and renal dysfunction. Late diagnosis may have contributed to the early deaths of these patients.

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[PubMed - indexed for MEDLINE]

Publication Types, MeSH Terms

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