Fabry's Disease

Fabry disease is a fat storage disorder. It causes an abnormal deposit of a particular fatty substance in blood vessels walls. It is caused by a lack of an enzyme. The mother needs to be a carrier to produce an affected child. Her sons have a 50 percent chance of having the condition, and her daughters have a 50 percent chance of being a carrier. Some of the female carriers exhibit signs of the condition, especially cloudiness of the cornea.

Symptoms males have include:

- Eye problems.
- Burning sensations (feeling) in their hands and feet that is worse with exercise and hot weather.
- Small, raised, reddish-purple blemishes on their skin.

As they grow older, they may have impaired arterial circulation leading to early heart attacks and strokes. The kidneys become progressively involved. Many patients require kidney transplantation or dialysis. A number of patients have trouble with their digestive track. Symptoms of this include frequent bowel movements shortly after eating.

TREATMENT

The pain in the hands and feet usually responds to medicine such as Tegretol (carbamazepine) and dilantin. Gastrointestinal hyperactivity may be treated with metoclopramide or Lipisorb® (a nutritional supplement). Recent experiments indicate that enzyme replacement is works for patients with this disorder.

WHAT WILL HAPPEN

Patients with Fabry disease usually survive into adulthood. They are at risk for:

- Strokes.
- Heart attacks.
- Kidney damage.

The hope is that enzyme replacement and gene therapy will eliminate these difficulties.

RESEARCH BEING DONE

NINDS supports research to find ways to treat and prevent lipid storage disorders such as Fabry disease.

Research is continually being done to learn more about this problem.

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