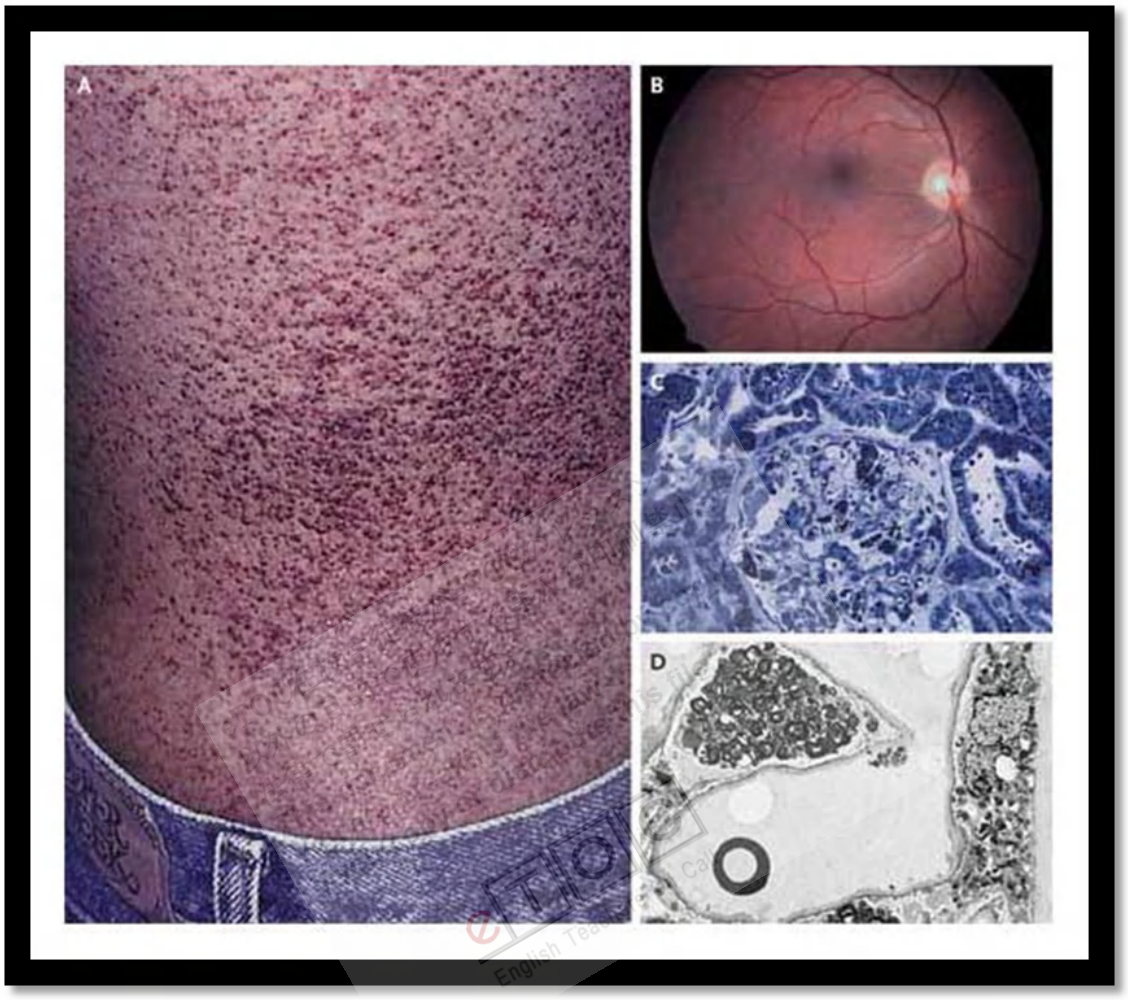
Fabry Disease



http://www.nejm.org/doi/full/10.1056/ENEJMicm000011

What is Fabry disease?

Fabry disease is caused by the lack of or faulty enzyme needed to metabolize lipids, fat-like substances that include oils, waxes, and fatty acids. The enzyme is known as ceramide trihexosidase, also called alpha-galactosidase-A. A mutation in the gene that controls this enzyme causes insufficient breakdown of lipids, which build up to harmful levels in the eyes, kidneys, autonomic nervous system, and cardiovascular system. Since the gene that is altered is carried on a mother's X chromosome, her sons have a 50 percent chance of inheriting the disorder and her daughters have a 50 percent chance of being a carrier.

What are the symptoms of Fabry disease?

Some women who carry the geneticmutation may have symptoms of the disease. Symptoms usually begin during childhood or adolescence and include:

- burning sensations in the hands that gets worse with exercise and hot weather, and
- small, raised reddish-purple blemishes on the skin.

Some boys will also have eye manifestations, especially cloudiness of the cornea. Lipid storage may lead to impaired arterial circulation and increased risk of heart attack or stroke. The heart may also become enlarged and the kidneys may become progressively involved.

Other symptoms include:

- decreased sweating,
- fever, and
- gastrointestinal difficulties, particularly after eating.

Fabry disease is one of several lipid storage disorders.

Is there any treatment for Fabry disease?

Enzyme replacement may be effective in slowing the progression of the disease. The pain in the hands and feet usually responds to anticonvulsants such as phenytoin (Dilantin) and carbamazepine (Tegretol, Tegretol XR , Equetro, Carbatrol). Gastrointestinal hyperactivity may be treated with metoclopramide (Reglan, Octamide, Maxolon). Some individuals may requiredialysis or kidney transplantation.

Reference:
http://www.medicinenet.com