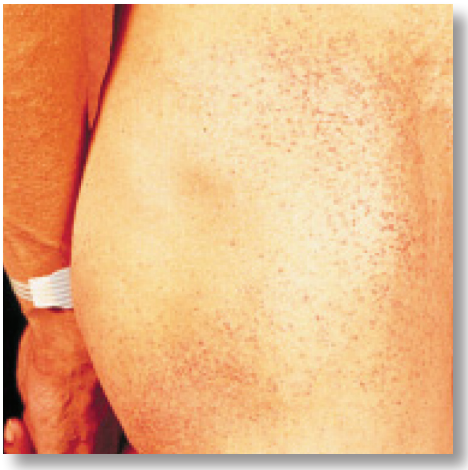
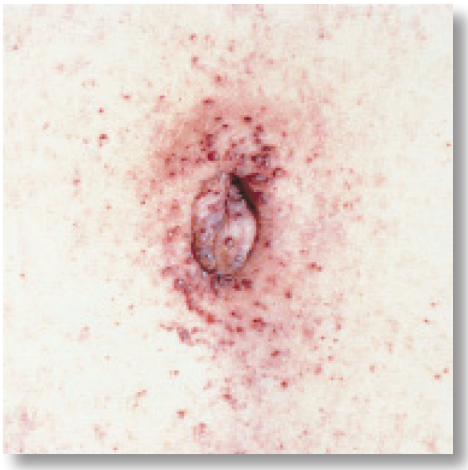
If you see a patient with angiokeratomas

It could be **FABRY DISEASE**

Information for **Dermatologists**





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Clusters of these characteristic angiectases are typically found between the umbilicus and thigh in patients with Fabry disease. (Left buttock shown left; umbilicus shown right.) Angiokeratomas may appear in childhood or adolescence, are most dense between the umbilicus and knees, and do not blanch with pressure.[1]

Dermatologists have the opportunity to identify patients with this progressive, often life-threatening genetic disease.

While Fabry disease is rare, it may be more common within a Fabry family.

In addition to angiokeratomas, patients with Fabry disease may present with[1]:

- Hypohidrosis/anhidrosis
- Lymphedema of the legs
- "Burning" pain in the hands and feet
- Heat/cold and exercise intolerance
- Gastrointestinal problems

Other manifestations include[1]:

- Progressive and/or unexplained chronic kidney disease
- Premature cardiac disease
- Corneal and lenticular abnormalities (seen through slit lamp—generally do not affect vision)
- Premature stroke

1. Desnick RJ, Ioannou YA, Eng CM. α-Galactosidase A Deficiency: Fabry Disease. In: Valle D, Beaudet AL, Vogelstein B, Kinzler KW, Antonarakis SE, Ballabio A, Gibson K, Mitchell G. eds. New York, NY: McGraw-Hill; 2014. http://ommbid.mhmedical.com/content.aspx?bookid=971&Sectionid=62644837

FABRY DISEASE

Silently Progressive. Increasingly Debilitating. Often Life-Threatening.

FABRY DISEASE PROFILE

Fabry disease is an inherited disorder that affects men, women, and children of all ethnicities. It is a multisystemic disorder that can result in irreversible, potentially life-threatening disease of the kidney, heart, and brain. The disease is characterized by the progressive cellular accumulation of a lipid substrate called globotriaosylceramide (or GL-3). Ongoing build-up of this substance is caused by deficiency of the lysosomal enzyme alpha galactosidase A (or α-GAL), which usually metabolizes GL-3 and keeps it from accumulating. Without enough of this essential enzyme, GL-3 accumulates in the lysosomes of most cell types over the course of a lifetime, often causing debilitating symptoms in childhood and adolescence and potentially irreversible tissue damage by adulthood.

DISEASE RISK IN FAMILIES

- Unlike many other X-linked disorders, females with the mutated gene are affected to varying degrees due to random X inactivation.
- Males with the disease pass the mutated gene on to all of their daughters and none of their sons.
- Females have a 50% chance with each pregnancy of passing the mutated gene to both their sons and daughters.
- If you identify a patient with Fabry disease, family testing should be considered.

LEARN MORE

Visit **www.fabrycommunity.com** for more information on Fabry disease or call Sanofi Genzyme Medical Information at 800-745-4447, option 2.

DIAGNOSIS

- Although Fabry disease usually presents in childhood, the disease often goes unrecognized by physicians until adulthood, when the underlying pathology is advanced.
- Delayed diagnosis may be the result of disease under-recognition and/or symptoms being mistaken for those of other disorders, such as rheumatoid or juvenile arthritis, rheumatic fever, erythromelalgia, multiple sclerosis, or lupus.
- Diagnosis is confirmed in males by enzyme assay (blood test) detecting low or absent levels of alpha-galactosidase A (α-GAL), or in females through genetic testing to detect a mutation.

Dermatologists are in a unique position to identify patients and families at risk for Fabry disease.

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