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Case Report

Extensive Angiokeratomas of the Trunk-An Unsual Presentation of Fabry Disease

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Abstract

A 23 year old man presented with erythematous non-pruritic rash over his trunk since childhood. Family history was non-contributory, and he did not have any major or chronic medical illness in the past. There was no history suggestive of paresthesias, exertional dyspnea or focal neurological deficit.

Keywords: angiokeratomas; fabry disease; skin redness

Case Report:

A 23 year old man presented with erythematous non-pruritic rash over his trunk since childhood. Family history was non-contributory, and he did not have any major or chronic medical illness in the past. There was no history suggestive of paresthesias, exertional dyspnea or focal neurological deficit.

On examination, he had a diffuse macular rash over his trunk, with multiple scattered cherry red painless papules. Systemic examination was unremarkable.

Based on the typical angiokeratomas on trunk, a possibility of Fabry disease was considered. Confirmation was by enzyme analysis which showed markedly reduced α-galactosidase activity (3.8 nmol/hr/mg [8.1-28.5]).

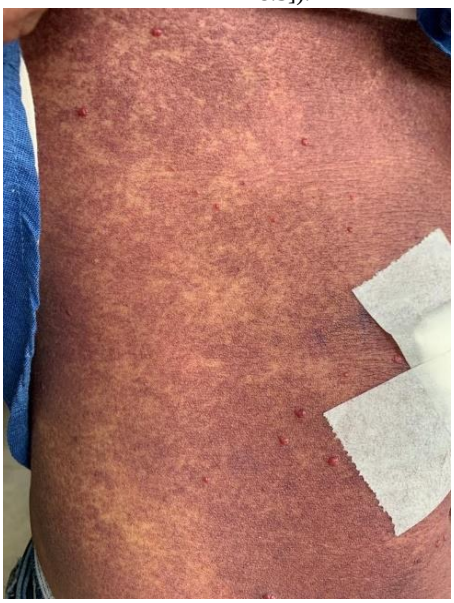

Figure 1: *Colour photograph showing reddish brown papular angiokeratomas*

Discussion:

Fabry disease was first described by Johannes Fabry and William Anderson in 1898, and results from mutation of GLA gene, leading to decreased levels of α-galactosidase enzyme [1]. Mode of inheritance in X-linked recessive, and females are occasionally affected due to inactivation of one X chromosome.

Neuropathic pain is the commonest manifestation (81.4%) caused by deposition of globotriaosylceramide in the dorsal root ganglion, followed by corneal opacities (76.9%), angiokeratoma (36%), hyperhydrosis, cardiac and renal involvement [2].

Angiokeratomas are the commonest dermatological manifestation, with age of onset at 5-10 years, seen mainly in the "bathing area". It is typically described as non-blanching, red to blue-black lesions ranging from 1 to 5 mm in diameter. Microscopically, it is composed of a vascular proliferation in the papillary dermis, with overlying acanthosis and keratosis [3].

Treatment of choice is recombinant enzyme infusion given at 2 weekly intervals, which results in resolution of dermatological lesions in more than 95% of cases [4].

Fabry disease, though being one of the common lysosomal storage disorders, is generally not diagnosed in childhood. A holistic approach is crucial for early and correct diagnosis, so that early enzyme replacement therapy can be initiated to ensure better outcome and improved quality of life.

Take Home Messages:

- Angiokeratomas in the bathing area is virtually pathognomic of the disease
- A holistic approach is crucial for early and correct diagnosis.
- Early enzyme replacement therapy should be initiated to ensure better outcome and improved quality of life.

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