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Case report

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Angiokeratoma – When is a few too many?

Colm O'Mahony[1], Andrea Franks[2] and Rhiannon Llewellyn[2]

Abstract
Many patients have few scattered angiokeratoma and we reassure them that this it is normal; however, if they are numerous, Fabry disease should be considered and check family history.

Keywords
Fabry, angiokeratoma, alpha galactosidase

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Introduction

Fabry disease is an X-linked lysosomal storage disorder due to alpha galacotosidase 'A' deficiency. This results in the accumulation of globotriaosylceramide (Gb3) within lysosomes in cells throughout the body, but especially in vascular endothelium. The more deficient the alpha galactosidase activity, the more Gb3 accumulates. It is an uncommon condition and diagnosis is often delayed because of the multiple systems involved and no standard pattern of presentation. However, one of the features is multiple angiokeratoma. In sexual health clinics, we frequently see patients who have a few scattered angiokeratoma in the genital area and we reassure them that this is normal. However, when they are multiple, Fabry disease should be considered.

Case report

A 33-year-old man from Bulgaria attended the GUM clinic in March 2012 because he had noted 'spots on his body'. He was fit and well, and there was no past history of any significance and, on retrospective questioning, no family history which might have alerted suspicion. His elder brother was well; however, his father died of a myocardial infarction at the age of 50 years. On examination, he had multiple angiokeratoma in the genital area and multiple, tiny angiokeratoma were noted on both flanks (Figures 1 and 2). C O'M was concerned with this clinical pattern and took photographs and emailed them, along with a referral letter, to AF in Dermatology. Ten days later, AF biopsied the angiokeratoma and considered as Fabry disease, confirmed by histology. The alpha-gal blood test was also sent along with routine investigations, and this test result was positive. The patient was recalled and the diagnosis was discussed with him. He was referred to the Department of Adult Inherited Metabolic Disorders at Salford Royal Hospital. After further investigation, the diagnosis was established as Fabry Disease gene mutation p.D92Y. The patient commenced Replagal which is agalsidase alpha. It is an intravenous infusion, given every two weeks. The cost is about £8400 per month.

Discussion

Because of the multiple possible manifestations with Fabry disease, it is usually diagnosed late. Neuropathic pain is common, as is abdominal pain and there can also be cardiac, renal and cerebral damage in early adulthood. Studies[1] indicate the correct diagnosis was usually obtained after a mean of almost 20 years in one survey and 27 years in a French study.[2] Neuropathic pain especially in the extremities is often a presentation. Check for family history of early death in males, especially cardiac or renal

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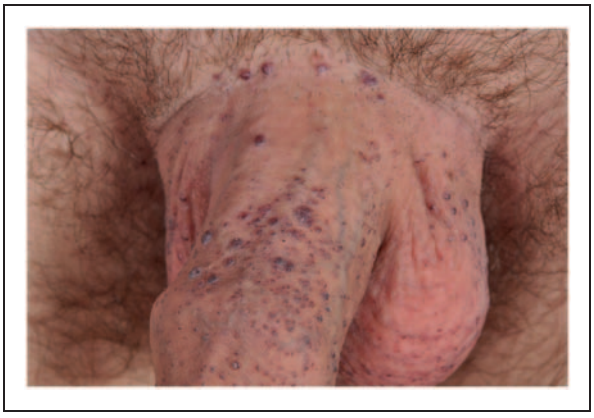

Figure 1. Multiple genital angiokeratoma.

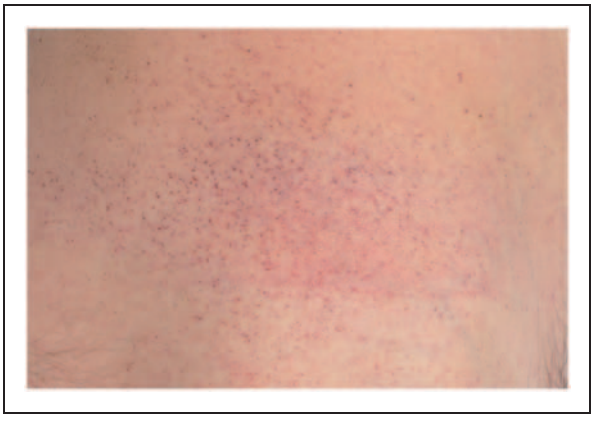

Figure 2. Image of right flank; numerous small angiokeratoma were present on both flanks.

failure. Hypohydrosis and unexplained recurrent abdominal pain can begin in childhood. Most have multiple angiokeratoma. Worryingly, however, one case report shows how a family had two further affected males and four carrier females detected simply because a 39-year-old man had a single angiokeratoma and family history of early onset cardiac disease.[3]

As many cases are diagnosed late, it is hard to establish how useful treatment is, but it has been shown that the treatment can alter the course of the disease if instituted early.[2]

So, consider the alpha-gal blood test (cost is about £104) in someone who has more than a few of 'normal' genital angiokeratoma and a suspicious family history. Early diagnosis and treatment can prevent progression and avoid future costly management of renal failure, i.e. dialysis and transplant.

Conflict of interest

The authors declare no conflict of interest.

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