Angiokeratoma Corporis Diffusum

A 13-year-old boy presented with history of fever and burning pain of the hands and feet for 10 days. He had four similar febrile episodes with burning pain of the hands and feet over the last one year. He also had hypohidrosis and heat intolerance. On examination, the child had multiple discrete reddish-brown papular non-blanchable lesions with keratotic surface extending from umbilicus to the knee in the 'bathing trunk distribution' (***Fig.* 1**). Based on the clinical findings a diagnosis of Fabry's disease (FD) was considered. Biopsy of one of the papular lesions showed multiple dilated capillaries with plenty of red blood cells within the lumen and the overlying epidermis showed mild hyperkeratosis suggestive of angiokeratoma corporis diffusum (ACD) (***Fig.* 2**). The enzyme α-galactosidase (α-GAL) in the leukocytes revealed very low levels of the enzyme (0.38 nmol/hr/mL; fluorometry method), thus confirming the diagnosis of Fabry's disease.

For many years, ACD was considered synonymous to Fabry disease, a systemic X-linked disease caused by deficiency of the lysosomal enzyme 'α-GAL' resulting in the accumulation of 'globotriaosylceramide'. ACD can also occur in association with other diseases related to deficiencies of enzymes that are involved in the metabolism of glycoproteins which include fucosidosis, sialidosis, α-mannosidosis, GM1 gangliosidosis, and Kanzaki disease . The disorders other than FD is typified by characteristic facies, mental retardation and organomegaly which were absent in the present case and the occurrence of acral paraesthesia and hypohidrosis were further suggestive of FD. Differential diagnosis of the cutaneous lesions must exclude the angiokeratoma of Fordyce spots, angiokeratoma of Mibelli, and angiokeratoma circumscriptum, none of which has the typical histologic or ultrastructural lysosomal storage pathology of the Fabry lesion.

Treatment of FD may include enzyme replacement therapy (ERT) using recombinant human α-GAL, analgesics (carbamazepine, or gabapentin), ACE inhibitors and dialysis or renal transplantation for end stage renal disease.

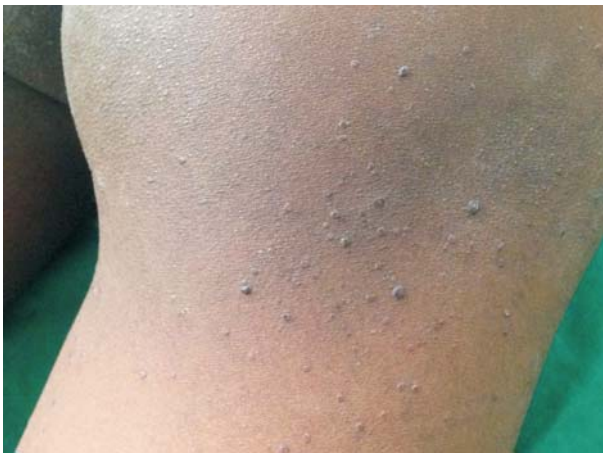

Fig. 1 *Multiple discrete reddish brown papular lesions of angiokeratoma.*

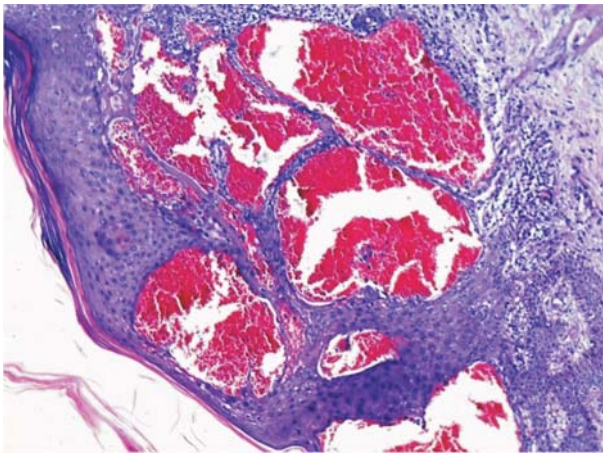

Fig. 2 *Biopsy of the papular lesion showing multiple dilated capillaries and hyperkeratosis of overlying epidermis.*

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