

GUNTHER'S DISEASE AND ANGIOFIBROMAS OF THE FACE

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INTRODUCTION

Angiofibromas of the face occur in association with Tuberous Sclerosis.¹

We report a case of Gunther's disease which developed Angiofibromas on the face during the course of the disease.

To the best of our knowledge this is the first case report with such an association in the world literature.

CASE REPORT

A thirty years old man complained of recurrent blisters on the exposed parts of the body, photosensitivity and reddish discoloration of the urine since early childhood. For the last four years he developed multiple nodular lesions on the face. His parents were first cousins.

Clinical examination showed mutilation and scarring of both hands and face along with Hepatosplenomegaly. There were multiple skin coloured papules and nodules on both cheeks (Fig. 1 & 2), and beads of papules along the free margins of both eye lids (Fig. 4). There was also brown discoloration of the teeth and hypertrichosis.

Investigations revealed a positive test for urinary porphyrins, Hepatosplenomegaly and Haemolytic Anemia.

Skin biopsy was reported as Angiofibroma (Fig. 3). There was also PAS positive material in the dermis.

On the basis of clinical history, laboratory investigations and biopsy report, a diagnosis of Gunther's disease with Angiofibromas was made.

DISCUSSION

Congenital erythropoietic porphyria (Gunther's disease) is a rare, autosomal recessive disease which presents with severe photo-sensitivity, mutilation and haemolytic anemia with splenomegaly.^{2,3} Not more than 200 cases are reported in the World's literature upto 1987.⁴

Some of the additional clinical features include hypertrichosis brown coloured teeth, fluorescence of teeth and urine reddish pink under Wood's lamp.² Interestingly both these features were seen in our case. One case previously reported by us⁵ also had these features. Beaded papules along the margin of the Eyelids which are classically present in Lipoid proteinosis⁶ were also seen in our patient. Further studies are needed to find out whether this was a chance occurrence or an association with this disease.

Angiofibromas are only seen in association with Tuberous Sclerosis but we have observed this association in Gunther's

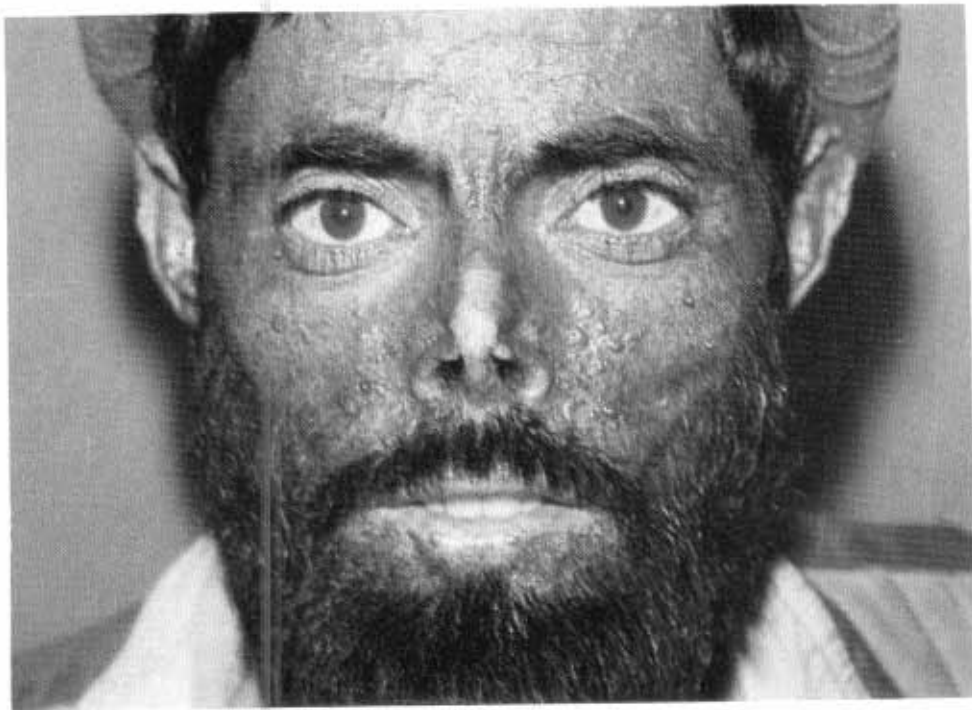


Fig. 1. Multiple papules and nodules on the face with scarring of the nose.



Fig. 2. Mutilation of the exposed parts of the hands.

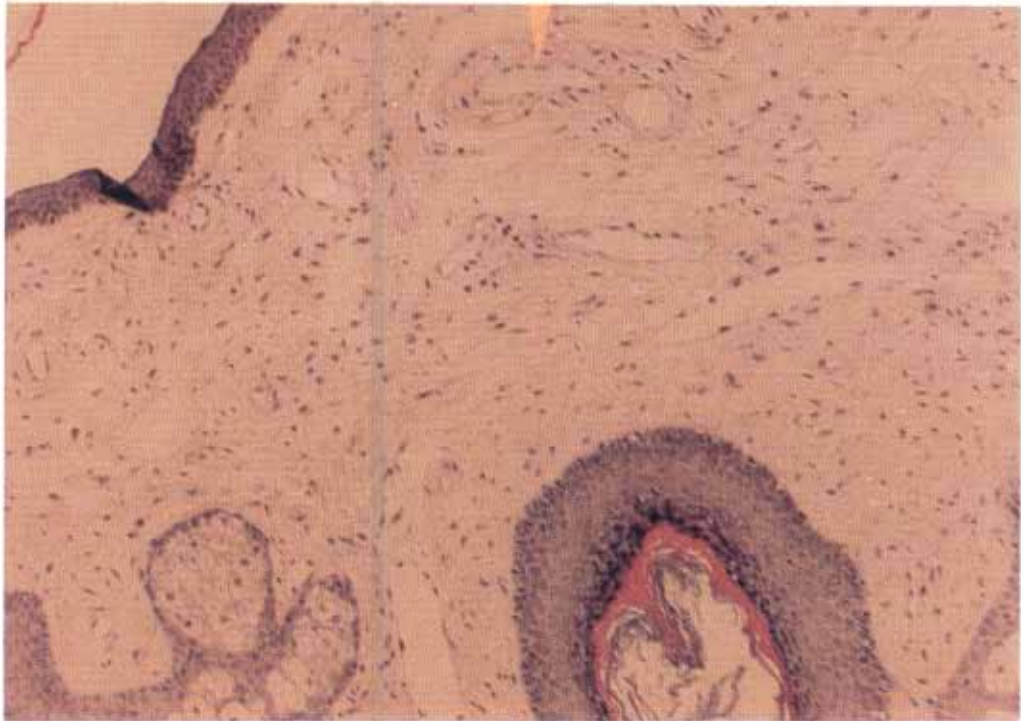


Fig. 3. Angiofibroma: proliferation of blood vessels in the dermis with fibroblastic stroma.



Fig. 4. Beaded papules along the free margin of the upper eye lids.

disease. It may be a chance occurrence or an association with this disease. Similar case has been diagnosed by Major General Ashfaq Ahmed Khan (personal communication). This makes the later probability more likely.

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