A clinical quiz that turns heads

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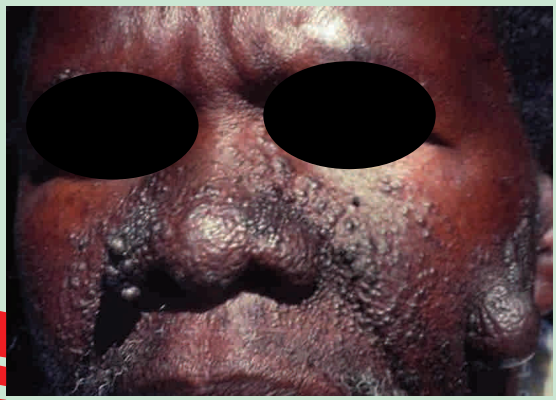
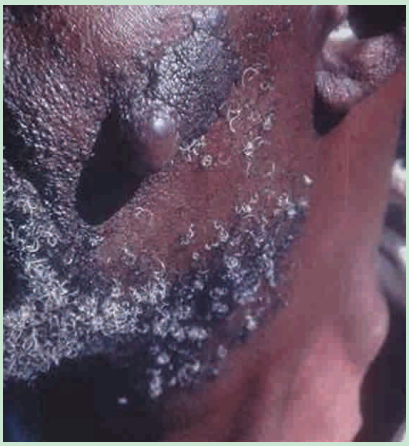
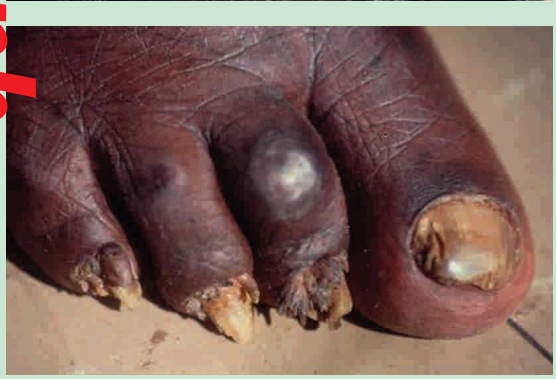
This column is aimed at developing your clinical acumen. A clinical quiz will alternate with a short discussion of a clinical sign. You are invited to send us requests for future topics and to provide photographs of clinical signs for the quiz section. Kindly send a fax or e-mail with your requests and mail high gloss photographs or a disk with high resolution (**300dpi**) **jpeg** files to us. (See contact details above) Photographs may include clinical signs, photographs of poisonous insects, plants, snakes, contaminated water or anything that may cause sickness or disease in South Africa. Kindly provide a short clinical synopsis of 100-200 words from which a quiz can be formulated.

quiz

This patient experienced frequent epileptic seizures since the onset of puberty.
What is the most likely diagnosis?
And, what other conditions are associated with this disease?







Answer

Tuberous sclerosis (Epilola or Adenoma Sebaceum). This is an inherited autosomal dominant condition that may present with a number of cutaneous manifestations.

The most characteristic are the cutaneous angiofibromas that start to appear more or less around puberty. Because they are often more abundant around the nose and the appearance at puberty they were wrongly called "adenoma sebaceum"; however they are neither adenomas nor sebaceous. The same fibromas may occur under the nail beds (very tender to pressure) or on the nail cuticles, called subungual or peri ungual fibromas. Lesions in visceral organs may lead to excessive bleeding during surgery. They are easily demonstrated on CT or MR scanning. It is assumed that the intra cerebral fibromas are the cause of late onset epilepsy.

The one lesion that may be visible at birth is a so called "ash leaf" lesion – a patch of depigmented skin in the shape of an ash leaf. (For South Africans, a willow leaf shape may be more familiar). A variation on the above is called a "confetti" lesion. A cluster of depigmented spots, each about 3mm in diameter with normal skin around the lesions. An additional lesion is called a "shagreen" patch (like shark skin). These are patches of rough, slightly raised skin, occur most often on the buttock, and less often on other places. Epilola patients generally have a slightly lower IQ compared to the national average. Like most inherited conditions, spontaneous new mutations do occur. Thus, a positive family history is present in about 60% of cases.