A clinical quiz that turns heads

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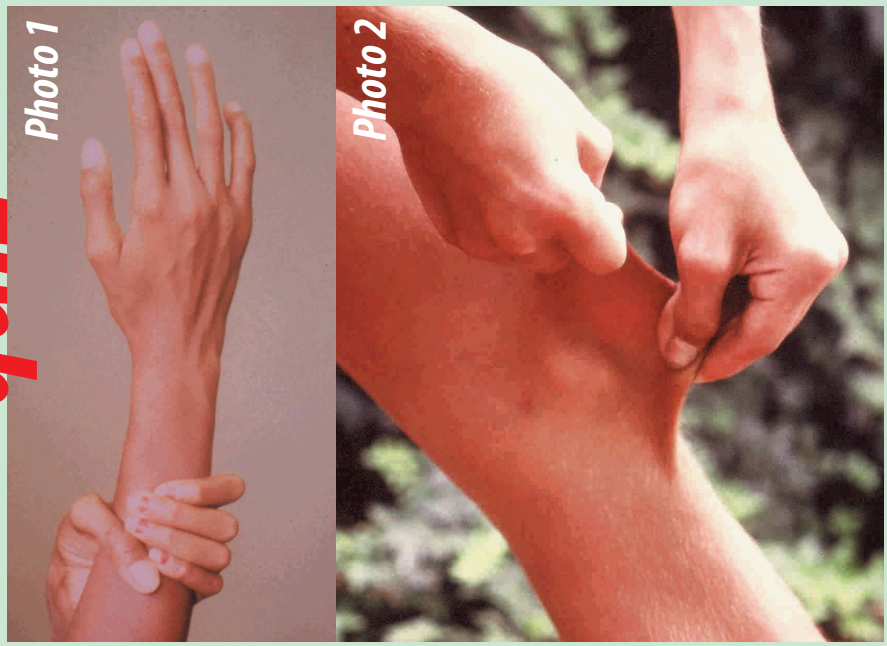
This column is aimed at developing your clinical acumen. A clinical quiz will alternate with a short discussion of a clinical sign. You are invited to send us requests for future topics and to provide photographs of clinical signs for the quiz section. Kindly send a fax or e-mail with your requests and 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.

These healthy persons came for routine medical examinations before taking up new employment. What are the most likely diagnoses (Photos 1 and 2) ?

quiz



Answer

ANSWER PHOTO ONE:

Lax ligament syndrome (a collective noun), with Marfan's syndrome the most likely condition. Marfan's syndrome is an autosomal, dominantly inherited condition, or can occur as a new mutation in 20 % of cases. It affects about 1 in 10000 of the population, but with a considerable phenotypic variation. The basic lesion is the production of poor quality fibrillin (elastin), due to a gene defect on chromosome 15.

For confirmation of the diagnosis two out of three major systems should be affected:
1. Heart: Mitral valve prolapse, Aortic aneurism (with or without dissection)
2. Eye: Lens dislocation (usually associated with varying degrees of myopia).
3. Skeletal:Tall thin body build, long arms (span> body height),legs and fingers; Scoliosis, and pectus deformity.

Due to the low quality elastin the following manifestations may in addition be present: Disc prolapse, spontaneous pneumothorax, joint dislocations, striae, and excessive keloid formation.

Advice to be given to the patient:

It is essential to treat even mild hypertension (avoid aneurism rupture) with beta-blockers. An annual cardiac ultrasound should be done, in order to plan aneurism surgery if necessary.

Strenuous physical activity and contact sport like boxing or rugby may cause retinal detachment or lens dislocation. Repeated over stretching of joints may lead to osteo arthritis later in life.

OTHER CONDITIONS WITH LAX LIGAMENTS

Homocystinuria has the same morphologic appearance as Marfan's, but is inherited recessively. They often suffer from mental retardation, glaucoma, myopia, and multiple thrombo embolic events. High doses of Pyridoxine (250 to 1200 mg per day) often controls the thrombo embolism. If not controlled, life long anticoagulation should be considered.

ANSWER PHOTO TWO:

A Marfanoid appearance with extremely lax skin, hypermobile joints, and abnormalities of blood vessels causing easy bruising of the skin is most likely due to the Ehlers- Danlos syndrome. Eleven different types, with varying genetic inheritance patterns, can occur. Clinically mitral valve prolapse, hernias, pes planus, and scoliosis are common. Repeated dislocations may lead to degenerative arthritis.

Other rare marfanoid conditions include Stickler syndrome, Beal's syndrome, and Shprinzen-Goldberg syndrome.