

Picture Story

A case of cutis laxa

P M G Punchihewa¹, L P C Saman Kumara²

Sri Lanka Journal of Child Health, 2002; **31**: 69-70

(Key words: cutis laxa)

This one year and six months old girl was referred to our clinic for her abnormal appearance since birth. She was born at term to non-consanguineous parents by normal vaginal delivery. The birth weight was 2.8 kg. She was the only child in the family and there was no family history of a similar disorder.

On examination, the most obvious findings were a prematurely aged appearance of the face and widespread lax skin with numerous skin folds resembling an ill-fitting suit. In addition she had other dysmorphic features such as large ears, hooked nose with everted nostrils, shaggy jaws, long upper lip and everted lower eyelids (Figures 1 & 2).



Figure 1 and 2. Patient with Cutis laxa

Her mental and physical development were normal. The joints were not hypermobile. There was no clinical evidence of cardiovascular complications such as peripheral pulmonary artery stenosis or aortic dilatation. So far she had not had any complications of the disease, such as pulmonary emphysema, bronchiectasis, pneumothorax, hernia or rectal prolapse. However, diverticula of the gastrointestinal and genitourinary tracts have not yet been excluded.

Cutis laxa is a rare hereditary disorder characterised by loose, pendulous skin which gives the afflicted child a prematurely aged appearance¹. The pendulous skin is most prominent around the eyes, face, neck and the trunk. Many infants have a hoarse cry, probably as a result of laxity of the vocal cords².

It is inherited most commonly as an autosomal recessive disorder. An autosomal dominant form has been described^{1,2}.

¹Consultant Paediatrician, ²Registrar in Paediatrics, Teaching Hospital, Karapitiya, Galle.

The autosomal dominant form of cutis laxa may develop at any age and is generally benign and mainly of cosmetic significance². When it presents in infancy, it may be associated with intra-uterine growth retardation and delayed closure of fontanelles. Affected males may be impotent and have infantile genitalia and scanty body hair.

In contrast, those with the more common recessive form of the disease are susceptible to severe complications such as genitourinary and gastrointestinal diverticula, diaphragmatic hernia and emphysema leading to cor pulmonale and death in the first few years of life^{2,3}.

The pathogenesis of cutis laxa is not well understood, but increased rate of enzymatic degradation of elastin has been demonstrated. Reduced elastase inhibitor levels and also decreased elastin messenger RNA levels in fibroblasts of affected individuals support this finding³.

Recent studies have shown that lysyl oxidase, a copper dependent enzyme, is important in the synthesis and cross linkage of elastin and collagen. Therefore, low levels of copper could lead to diminished elastin synthesis. However, only a few patients with cutis laxa have demonstrated low serum copper levels³.

Histologically, elastic tissue is reduced throughout the dermis with fragmentation, distension and clumping of elastic fibres. Multiorgan involvement is as a result of defective supporting structure^{1,2}.

Ehlers Danlos syndrome has been confused with cutis laxa but the features of the two disorders differ considerably. The skin in Ehlers Danlos syndrome is hyperextensible and snaps back into place when stretched unlike in cutis laxa¹.

Acquired forms of cutis laxa, which are less common than the congenital forms, have been reported following febrile illnesses, inflammatory skin diseases such as lupus erythematosus, urticaria, C3 C4 deficiency, hypersensitivity reactions to penicillin and also in infants born to mothers receiving penicillamine^{2,3}. It is thought that inflammatory cells or their mediators might damage elastic fibres³.

Though there is no effective medical treatment for this disorder, plastic surgery may improve the appearance of the face but dermal laxity may recur³. Avoiding cigarette smoking is beneficial to prevent progression of the pulmonary disease³. In the absence of pulmonary emphysema, the prognosis is reasonably good but pulmonary function should be assessed regularly anticipating complications.

References

1. Rudolph A M editor, Pediatrics. Norwalk; Appleton & Lange 1987; 803-4.
2. Behrman RE editor. Nelson Textbook of Pediatrics. Philadelphia; Saunders 1992; 1660-1661.
3. Molis T. Cutis laxa (Elastolysis). www.emedicine.com/derm/topic93.htm (accessed on 16/05/2002).

