

Congenital Cutis Laxa

A four-month-old male child presented with history of repeated episodes of fever, cough and fast breathing since early neonatal period. He had old man like facies with puffy eyes, prominent easily stretchable ears and everted nostril. Skin was loose and redundant, most marked over both the lower limbs. There was small paraumbilical hernia, left inguinal hernia and micropenis (**Fig.1**). Abdomen was soft and distended with hepatosplenomegaly (liver 3 cm and spleen 2 cm below subcostal margin). Anteroposterior diameter of the chest was increased with bilateral crepitations and rhonchi on auscultation. Weight of the child was 3.6 kg (< 5th percentile), length- 53 cm (< 3rd percentile) and head circumference- 35.8 cm. X-ray chest revealed bilateral emphysematous lungs. Based on clinical and radiological features, a diagnosis of congenital cutis laxa was made.

Congenital cutis laxa is a rare hereditary connective tissue disorder resulting from abnormality of elastin fibres. It is characterized by loose, redundant skin all over body associated with other extra cutaneous manifestations in form of inguinal and umbilical hernia, pulmonary emphysema, gastrointestinal and bladder wall diverticulae and pulmonary artery branch stenosis. Our patient probably had type I autosomal recessive variant of cutis laxa. Absence of similar type of illness in other family members rules out the possibility of autosomal dominant type. The condition needs to be differentiated from Ehlers-Danlos Syndrome, where skin is hyperextensible and joints are hypermobile. Other condition with cutis laxa like skin



FIG. 1 (a) Senile appearance of the face, puffy eyes and everted nostrils; (b) Loose skin folds over lower limbs, small paraumbilical hernia, right inguinal hernia and micropenis.

changes is Costello syndrome which can be differentiated by presence of hyperkeratotic changes of palms and soles, abnormally flexible finger joints and presence of papillomata around mouth and nostrils. Absence of joint laxity rules out the possibility of type II congenital cutis laxa and Ehlers- Danlos Syndrome. There is no specific treatment for this condition. Prognosis of type I congenital cutis laxa is bad and majority of children die in early childhood due to pulmonary or cardiovascular complications.

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Multifocal Cystic Osseous Tuberculosis with Lupus Vulgaris

A 6-year boy reported with multiple swellings on the dorsum of both hands and elbows and low grade fever for 6 months. He had received multiple antibiotics without any response. Examination revealed cervico-axillary lymphadenopathy with irregular soft swellings on the dorsum of both hands and elbows. Overlying skin was erythematous with ulcers having bluish thin undermined margins and serosanguinous ooze (**Fig. 1**). Investigations revealed ESR 50mm, Mantoux 15mm and normal chest



FIG.1 Erythematous skin with characteristic ulcers.