GENETICS AND GENODERMATOSES

AUTOSOMAL RECESSIVE CUTIS LAXA TYPE 1 – A CASE REPORT

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Background: Cutis Laxa (CL) is a rare connective tissue disorder with a prevalence of 1 case per 1,000,000 population. It is characterised by loose, sagging, redundant skin over the body. Both inherited and acquired forms are known. We report this case of Autosomal Recessive Cutis Laxa with systemic involvement.

Case synopsis: A 7 year old boy, born out of consanguineous marriage, presented with loose, sagging skin mainly over face, extremities and skin folds since birth with recurrent respiratory & systemic complaints. Patient had a characteristic senile bloodhound appearance. Chest X-ray findings included emphysema, bronchopneumonia and pulmonary hypertension. Ultrasonographic examination showed cystitis and bladder diverticula. 2D Echocardiography showed primary pulmonary hypertension. Histopathological examination with Verhoeff-Van Gieson stain, showed markedly reduced elastic fibers. Screening for congenital disorders of glycosylation by transferrin isoelectric focusing was done to rule out CDG syndrome.. Serum amino acids copper & ceruloplasmin were normal.

Discussion: The rare reported prevalence of Cutis Laxa makes this case important. Differential diagnosis includes Ehler's-Danlos syndrome and Pseudo Xanthoma Elasticum. Diagnosis of ARCL-1 was done on basis of clinical features, extra cutaneous findings and histopathological findings with the exclusion of differential diagnosis. There is no effective treatment available at present for ARCL-1. Symptomatic management for the associated complications remains the mainstay of treatment. Genetic counselling of the affected family was essential. Around 60 cases of ARCL-1 has been reported in literature so far.
We report this case of ARCL-1 for its rarity & complex systemic involvement.