GENETICS AND GENODERMATOSES

GENETIC ANALYSIS IN A PATIENT WITH GENERALIZED CUTIS LAXA- AN INTERESTING CASE REPORT

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Background: Cutis Laxa [CL] is a rare connective tissue disorder which causes abnormalities in elastic tissue. They can either be inherited or acquired. Acquired forms are secondary to inflammatory diseases. We report a patient with CL for whom genetic analysis was performed.

Observation: A 23 year old female patient presented with loosening of skin all over the body for 10 years. She gives history of recurrent urticarial lesions with angioedema in her childhood, after which she started noticing progressive laxity of skin which became generalized.On examination skin was loose and inelastic and on stretching the skin there was slow recoiling Based on history, clinical examination and histopathology with Verhoeff–van Gieson stain diagnosis of CL was considered. Investigations revealed no systemic abnormality.Genetic analysis for elastin gene was found to be negative. Inherited CL can be autosomal dominant, autosomal recessive or X linked.Autosomal Dominant is due to defect in elastin gene and presents at birth or at adolescence. Autosomal Recessive is the most severe form which presents at birth due mutation in fibulin gene . Acquired variants may occur at any age following inflammatory skin disease or exposure to drugs, in these cases release of elastases into the extracellular milieu by inflammatory cells may lead to proteolytic degeneration of elastic fibres and clinically presents as lax skin. Since our patient had onset of disease at adolescence, with a preceding urticarial eruption ,manifesting as loosening of skin and the genetic analysis showed no defect in elastin gene, we made a diagnosis of acquired generalized CL

Key Message: Acquired generalized CL is a rare presentation which can occur secondary to urticaria and genetic analysis can help in substantiating the diagnosis.