PRESENTATION OF A PEDIGREE OF AN IRANIAN FAMILY WITH TWO MEMBERS WITH CUTIS LAXA AUTOSOMAL RECESSIVE TYPE I

H. POUR-JAFARI*, Ph.D., M.G, AND A. SARIHI, ** Ph.D.

From the Departments of *Genetics and **Physiology, Medical School, Hamadan University of Medical Sciences, Hamadan, I.R. Iran.

ABSTRACT

Congenital cutis laxa is an exceptional condition. No large scale pedigree has been reported from Iran. We report a family with 106 members with two members affected with cutis laxa.

Our cases were two patients (male and female) with pre- and postnatal growth retardation, cutis laxa, characteristic facies and other manifestations which proved that they were affected with cutis laxa. Their family history was studied and a large pedigree was drawn up.

Based on the findings in their pedigree pattern, in addition to clinical and pathological studies, one can say that cutis laxa in this family is autosomal recessive. We also showed obligate carrier members in the family.

Recent studies have shown that cutis laxa is a heterogeneous group of conditions both clinically and genetically. Autosomal dominant, autosomal recessive, X-linked and also acquired forms have been reported. Our study indicates that our case is an autosomal recessive type I. We discussed the pedigree that covers five generations.


Keywords: Abnormalities, Cutis laxa, Hereditary diseases.

INTRODUCTION

Goltz et al. described affected brothers and suggested recessiveness because of other reported instances of affected sibs as well as parental consanguinity.1 Different studies have shown that cutis laxa is a heterogeneous group of conditions both clinically and genetically. Autosomal dominant,2 autosomal recessive,3 X-linked,4 and also acquired forms5 have been reported.

Our research group recently worked on a case of cutis laxa that is alive, and her dead brother who was affected too. Her hanging skin, giving the appearance of premature aging and her other clinical, para-clinical and genetic features were studied. Based on clinical manifestations and laboratory findings, we found that our cases are affected with CCL (congenital cutis laxa type I).6 We studied their family through different approaches. The aim of the present study is to determine the mode of transmission of the mutant gene through our case family.

CASE REPORT

Our primary case was a 14-year-old Iranian girl affected with congenital cutis laxa (Fig. 1). We studied her clinical manifestations, laboratory findings and also her family history. She had pre- and post-natal growth retar-
Cutis Laxa in an Iranian Family

Fig. 1. Photographs of our patient’s face, hands and feet (a 4-year-old girl with congenital cutis laxa).

Fig. 2. Pedigree of the kindred analyzed. Blackened symbols indicate individuals affected with cutis laxa. Semiblackened symbols indicate obligate carriers. IV-27 is the propositus.

Indeed, complete clinical and family studies and skin biopsy with standard histology, orceine staining and histomorphometric analysis of the collagen and elastic fibers of the dermis were performed to prove that our case was a case of congenital cutis laxa. Her family history showed that she had another sib that based on his hospital information was affected with similar disease. He had died at the age of two years. We drew up their pedigree containing four generations and 106 members.

Based on the following observations, it seems that the pedigree pattern of cutis laxa in the subject family is autosomal recessive:
REFERENCES
