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Ehlers-Danlos Syndrome: Variable Expression in Sibs

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Ehlers-Danlos Syndrome (EDS) is a genetically determined disorder of connective tissue. It was first described by Van Meekeran in 1682(1). The condition derives

its name from reports by Edvard Ehlers, a Danish dermatologist and Henri Danlos, a French physician. Though, more than 1000 cases have been reported in world literature, current interest centres around the recognition of increasing heterogeneity in EDS(2). We report here two sibs with EDS who had different severity of manifestations.

Case Report

A ten-year-old girl born of a non-consanguineous marriage was brought to us

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with history of easy bruising and delayed wound healing since birth. On examination, she had abnormal facies in the form of widely spaced eyes and wide nasal bridge and soft and hyperextensible skin. There were non-healing gaping wounds with fish-mouth appearance on the extensor aspect of right elbow, shin of right tibia and right ankle. Over both the knees, she had the characteristic cigarette-paper or papyraceous scars (*Fig. 1*) which were corrugated by fine wrinkles. In addition, the so called molluscoid pseudotumors were found at the heel and elbows. Palpation of thighs revealed small, irregular, firm cysts commonly referred to as spherules. All the joints were hyperextensible. The knees and elbows could be extended past 180° while the fingers could be extended beyond 90° from the palmar plane (*Fig. 2*). Systemic examination of eyes and dentition did not reveal any abnormalities. She did not have varicose veins or musculoskeletal deformities. Skin biopsy showed disorganized collagen fibres.

Although there was no positive family history of similar complaints we examined the family members and other relatives. Her only sib, one 7-year-old male had mild hyperextensible skin and mild hypermobility of joints. There were no other features of EDS seen.

Discussion

EDS comprise a heterogeneous group of connective tissue disorders with extensible skin, connective tissue fragility and hypermobile joints. The syndrome has been categorized into eleven types based on clinical features, their severity and mode of inheritance(3,4). The exact abnormality in biogenesis of the collagens has been identified in four varieties and in case of EDS IV an abnormal gene locus has been determined.

In this report, the elder sib had all the signs of EDS II (mitis type) including hyperextensible skin, dystrophic scarring, easy bruising, joint hypermobility, connective tissue fragility, spherules and

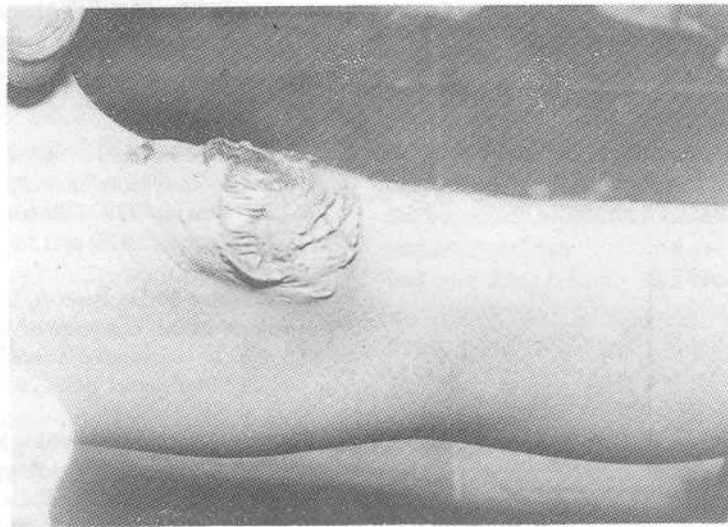


Fig.1. Cigarette-paper or papyraceous scar.

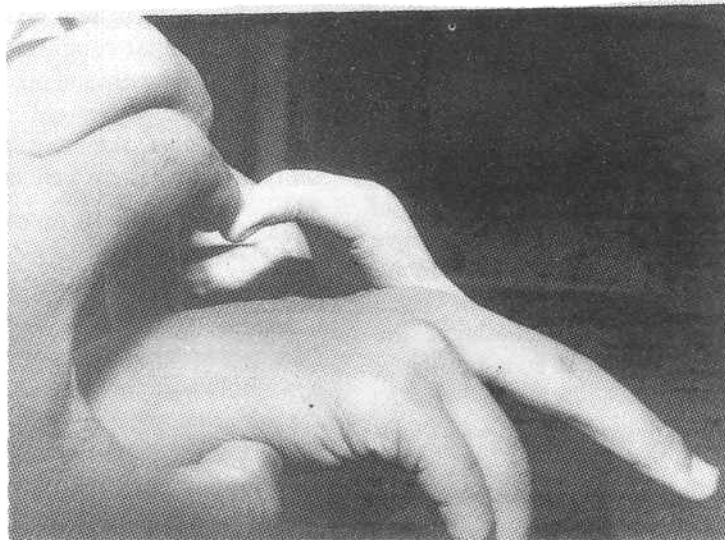


Fig. 2. Hyperextensibility of fingers.

molluscoid pseudotumors. In comparison, the younger sib had hyperextensible skin and hypermobile joints, only to a mild extent (EDS II) so as to escape the notice of parents. There were no other affected relatives. EDS II is inherited as an autosomal dominant trait. The basic defect leading to manifestations has not been determined. This type has manifestations which are similar to but are less severe than those in EDS I (gravis). Though EDS is known to have wide clinical variability, cases in the same family are known to exhibit considerable similarity(2). In contrast these sibs had marked differences in severity of manifestations. Autosomal dominant inheritance seems unlikely as none of the other family members had any manifestations of EDS. Such cases are explained by new mutation or variability of phenotypic expression.

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