Sjögren-Larsson Syndrome in a Turkish Family

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Sjögren-Larsson syndrome, first described by Sjögren in 1956, and then jointly with Larsson in 1957, is known to occur in different populations throughout the world (Blumel, Watkins, and Eggers, 1958; Richards, 1960; Link and Roldan, 1958; Zaleski, 1962; Heijer and Reed, 1965; Selmanowitz and Porter, 1967). The condition appears to be a clinical and pathological entity characterized by ichthyosiform erythroderma, spastic diplegia, and mental retardation. Sjögren-Larsson syndrome has also been observed in Turkey (Drs. Koloğlu and Renda, personal communications). The present report, to our knowledge, is the first description of a Turkish family with the clustering of such cases.

The Family

The Akar family is of Turkish origin, having lived in the southern villages of central Turkey, near Konya.

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The parents come from a large kindred where marriages between blood relatives are common, they themselves being first cousins. They, however, are healthy and free of any one of the signs characteristic of the syndrome. No one else in the family is known to be so affected.

The family have lived in Ankara for some years.

The propositus, E.A., born in 1962, is the first female child of a sibship of three who all proved to be affected. The mother and father were 22 and 24 years old, respectively, when she was born, and there was no history of abortion or stillbirth. When first seen she was still unable to walk or to speak, and her skin had been ichthyosiform since birth.

On physical examination she was generally found to be in good health; but she could not walk, and even standing was impossible without support. Her height was 91 cm. and weight 15.2 kg. Growth was uneventful but pes equinovarus developed, apparently secondary, and subnormality became evident. Her speech consisted of only a few words. The degree of mental retardation precluded intelligence tests. Fig. 1 shows the three affected sibs together.

FIG. 1. The three sibs with the Sjögren-Larsson syndrome. Their legs are bandaged because of biopsy.

The skin was ichthyosiform (Fig. 2), with the exception of head, face, and genitalia, and sweating occurred only on the face, palms, and soles. No abnormality was detected in other organs and systems. Pyramidal signs were apparent, there being increased muscle tone, and the reflexes were slightly increased. She was calm and smiling but obviously retarded in mental development.

FIG. 2. Skin appearance of E.A., the oldest of the sibs. The scar over the deltoid is due to tuberculin testing.

Laboratory investigations, including radiography, revealed normal findings. Phenistix test strips (Ames Co.) for phenylketonuria were also normal and a normal amino acid pattern was reported for urine. The skin condition was confirmed microscopically and gastrocnemius muscle biopsy showed a mild degree of atrophy. Karyotyping from peripheral blood leucocytes revealed no abnormality. The patient was discharged from hospital without any specific treatment.

B.A., the second female child of Akars, was born in 1963 and affected similarly. She too could not walk or stand up, and she was obviously retarded in mental development; though her general health was reasonably good (Fig. 1). Her karyotype was normal.

B.A., a male, 1½ years old at the time of examination,

is the last sib with spastic diplegia, mental retardation, and congenitally ichthyosiform skin. No other abnormality was found. It seemed that he was less severely affected than his older sisters, though judgement was necessarily subjective.

Discussion

The Akar family with three affected children born to related parents confirms the hypothesis that the Sjögren-Larsson syndrome is due to a rare single autosomal gene (see the references), affected individuals being homozygous. Affection of all three sibs can be easily explained as a chance phenomenon, for the parents are apparently heterozygotes for the gene. They, however, are free of the signs of disease, and this would be expected as the gene appears to be completely recessive. Minor abnormalities seen in some heterozygotes (Selmanowitz and Porter, 1967) have not been observed by us.

To relate the Akar kindred living at Konya villages to any documented family or ancestry is impossible; but there is no doubt that they are of Anatolian extraction. This supports the suggestion that the mutation to the abnormal allele has occurred several times and in different populations (Blumel *et al.*, 1958). On the other hand, the mutation could have occurred many generations ago, even before the ancestors reached Turkey, and, there being so many consanguineous marriages, cases could have occurred in previous generations.

Summary

A Turkish family with three children is reported. All three sibs, two female and one male, are affected with the Sjögren-Larsson syndrome, an autosomal recessive condition. The parents who are first cousins are normal. Investigations did not disclose any metabolic defect underlying the disease.

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