Atypical serum cholinesterase in a family with congenital distichiasis*

Summary. This paper describes the coexistence of genetically determined reduced cholinesterase activity and congenital distichiasis in the same family. The pedigree suggests that these two autosomal dominant diseases are segregated independently and do not show evidence of linkage.

Several genetically determined variants of human serum cholinesterase have been identified (Kalow, 1959; Lehmann and Liddel, 1969; Whittaker, 1970). Individuals homozygous for the so-called 'atypical' allele show relatively low levels of enzyme activity and are excessively sensitive to succinylcholine. They are likely to suffer prolonged apnoea when this drug is used during routine anaesthetic procedures (Evans *et al*, 1952; Bush, 1961; Baraka *et al*, 1974). Such cases are relatively uncommon in most populations.

Congenital distichiasis or the presence of two rows of eye-lashes at the lid margins is a rare entity that follows the autosomal dominant mode of inheritance (Falls and Kertesz, 1964; Robinow *et al*, 1970; Hoover and Kelley, 1971).

The purpose of the present report is to describe the coexistence of distichiasis and atypical cholinesterase in the same family.

Subjects and methods

A 7-year-old boy (II.9) (see pedigree, Fig. 1) was found to have at the inner edge of each lid margin aberrant eyelashes that were rubbing over the cornea (Fig. 2). During the surgical correction of the distichiasis under general anaesthesia, small doses of succinylcholine (0.1 mg/kg) injected intravenously produced an abnormally prolonged neuromuscular block.

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The serum cholinesterase dibucaine numbers (DN) of the patient and the other members of the family were determined according to the method described by Kalow and Genest (1957). All the members of the family were also examined for the presence of distichiasis.

Finally, all members were tested for the ABO, MNS, P, Rh, Lu^a, Le, Fy, jK, and X_g^a blood groups.

Results

The mother and 4 other members of the family (II.2, II.5, II.9, II.13) were found to have distichiasis (Fig. 1).

The results of the dibucaine numbers are shown on the pedigree (Fig. 1). According to these results, the members of the family showed a trimodal distribution. Enzyme activity was found to be low

FIG. 1.

FIG. 2.

in II.4, II.9, and II.13, normal in II.5 and II.12, and intermediate in I.1, I.2, II.1, II.2, and II.7.

Discussion

Falls and Kertesz (1964) described a syndrome inherited as autosomal dominant and consisting of different congenital anomalies, mainly distichiasis involving all four lids, chronic lymphatic oedema of the lower extremities appearing at puberty (Milroy's disease), pterygium colli or webbing of the neck, and partial lateral ectropion of both lower lids. Other congenital anomalies such as vertebral anomalies, extradural cysts, and astigmatism were associated later to the syndrome (Chynn, 1967; Bergland, 1968; Robinow *et al*, 1970; Hoover and Kelley, 1971).

To our knowledge, this is the first published instance of congenital distichiasis and serum cholinesterase deficiency occurring in the same family and the same individuals (II.9 and II.13). The pedigree suggests, however, that these two traits are segregated independently and do not show evidence of linkage. Moreover, the analysis of the blood subgroups did not reveal any linkage either to the gene of distichiasis or to that of pseudocholinesterase.

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