FAMILIAL CORNEAL DYSTROPHY*

BY

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It may be of some interest to report the history over five generations of a family suffering from corneal dystrophy. In these days of rapid movements of the population it is seldom that the members of a family as large as the one reported remain in the immediate district in which they were born.

The dystrophy is of the nodular or granular type (Duke-Elder, 1938; Francheschetti, Klein, Formi, and Babel, 1951; Franceschetti, 1954). The cases would appear to fall into the classification of heredo-familial degenerations under the heading of parenchymatous degenerations—a dominant form of granular degeneration.

The youngest patient reported by Francheschetti was 5 years old, but in the lattice-like type of degeneration puberty appears to be the more usual age of onset. In the family here reported two girls (IV, 20 and IV, 21 in the family tree) were seen at the ages of 7 and 8 years respectively. They have a type of corneal erosion which lasted for a few days only and cleared, leaving little corneal damage. This has recurred perhaps once or twice a year, but was usually quiescent again when the children were examined.

Corneal grafting was considered for II, 7, who has since died, and his brother II, 8, still has 6/24 vision in each eye and grafting has not been considered advisable. As a consequence no such biopsy of the cornea has been possible as was done in the case reported by Stocker and Holt (1955). There appears to be no association with skin lesions as occurred in the family described by Savin (1956), in which four male members suffering from corneal dystrophy of the stroma and endothelium of the cornea also had ichthyosis simplex and allergic manifestations.

McGee and Falls (1953) were unable to trace the family history further because of geographic scattering, but suggested the possibility of a recessive gene in their second case, the child of a consanguineous marriage.

The inheritance in the family reported here would appear to be fully dominant with transmission over five generations without sex predilection; it follows the same pattern as the family with the lattice-type of corneal dystrophy described by Ramsay (1957).

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FIGURE.—Pedigree chart of a family affected by corneal dystrophy.

Key to Family Tree

(1) I, 1, who was himself affected, had nine children (II, 1-9), of whom three males and three females were affected.

(2) The eldest son (II, 1), himself unaffected, had three daughters (III, 1-3), none of whom was affected; nor were any of the children (IV, 1-4) of these women affected.

(3) The second son (II, 2), himself affected, had two children, a boy (III, 4) and a girl (III, 5), both affected. They each had a child (IV, 5 and 6), both of whom were unaffected. IV, 6 had two children (V, 1 and 2), who were also thought to be unaffected, but as he has left the area this is a little uncertain.

(4) The third child, a daughter (II, 3), herself affected, had four sons and a daughter. Her eldest son (III, 6), who was unaffected, was killed in the second world war and had no progeny. The next son (III, 7) died unaffected leaving four unaffected daughters (IV, 7-10), of whom IV, 8 and 9 each have unaffected daughters (V, 3 and 4). III, 8, who was killed in the second world war, was himself affected and had one affected son (IV, 11), one unaffected son (IV, 12), and an unaffected daughter (IV, 13). The fourth son (III, 9), himself affected, was reputed to have two unaffected children (IV, 14 and 15). The youngest child, an unaffected daughter (III, 10), had no children.

(5) The fourth child, a son (II, 4), was unaffected. He was killed in the first world war and had no progeny.

(6) The fifth child, a daughter (II, 5), herself affected, had one unaffected daughter (III, 11) followed by three affected sons (III, 12-14) and one unaffected son (III, 15).

(7) The sixth child, a daughter (II, 6), herself affected, had (by her first husband) one unaffected son (III, 16), one unaffected daughter (III, 17), and one affected daughter (III, 18). By her second husband, she had three sons and two daughters, all of whom were affected (III, 19-23).

The two unaffected children of the first marriage (III, 16 and 17), had no progeny. The affected daughter (III, 18) had three sons (IV, 16-18), of whom the eldest was affected, and one affected daughter (III, 19). Of these, IV, 16 had no progeny, IV, 17 had two unaffected daughters (V, 5 and 6), IV, 18 had one unaffected daughter (V, 7), and IV, 19 had one unaffected son (V, 8).

Of the five affected children of the second marriage, the eldest son (III, 19) had two affected daughters (IV, 20 and 21), and an unaffected son and daughter (IV, 22 and 23). The next son (III, 20) had four unaffected children (IV, 24-27). The next son (III, 21) had an unaffected daughter and son (IV, 28 and 29). The fourth child, a daughter (III, 22), had one unaffected son (III, 30). The youngest, another daughter (III, 23), had two unaffected daughters (IV, 31 and 32).

(8) The seventh child (II, 7), himself nearly blind with a corneal dystrophy which probably justified a corneal transplantation, is now dead. Of his seven children (III, 24-30), five were affected; there were two sets of twins, the first pair both affected, and the second pair both unaffected.

The eldest son (III, 24), himself affected, had one affected daughter (IV, 33). The second son (III, 25), himself affected, had no progeny. The third son (III,

26), who was the elder of the first pair of twins, was himself affected and had two unaffected daughters (IV, 34 and 35). His affected twin sister (III, 27) had three sons (IV, 36-38), two of whom (IV, 36 and 37) were affected. Of the second pair of twins (III, 28 and 29), who were both unaffected, the younger (III, 29) had two unaffected sons (IV, 39 and 40). The youngest son (III, 30), himself affected, had no progeny.

(9) The eighth child (II, 8) is now 67 years of age and from him much of the family history has been obtained. His vision has never fallen below 6/36 and his dystrophy, granular in type, has tended to improve slightly without treatment during the 5 years he has been under observation, the visual acuity being now 6/24 in each eye and 6/18 partly with both eyes. He has two affected daughters (III, 31 and 32), the elder of whom has an unaffected son (IV, 41), and an affected daughter (IV, 42). The younger daughter (III, 32), herself affected, has two sons, IV, 43 affected and IV, 44 unaffected.

(10) The ninth and last child (II, 9), himself unaffected, has four unaffected children.

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