HEREDITARY CILIARY AND SUPERCILIARY HYPOTRICHOSIS OF A DOMINANT CHARACTER*

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CONGENITAL lack of lashes and eyebrows, whether total or partial, is seldom encountered. Associated at times with other developmental defects, mostly ectodermal, it may appear in conjunction with general alopecia, faulty dentition, and/or cataract formation (Duke-Elder, 1952). In the so-called Ullrich-Dohna syndrome, it has been seen in combination with multiple ocular malformations, strabismus, micrognathia, trigonocephaly, and hypogenitalism (Ullrich and Fremerey-Dohna, 1953; Weyers, 1954; Leffertstra, 1956); the disease is not hereditary. In one instance, at least, it has also been observed as part of a complex syndrome consisting of retinitis pigmentosa, opacities of the lens, anisocoria, oligophrenia, and some degree of somatic hypotrophy (Sánchez Salorio, 1955); the affection was transmitted according to a simple recessive pattern.

Its early occurrence in cases of hyperkeratosis follicularis spinulosa decalvans (Siemens, 1926) has likewise been recorded (Wessely, 1929); in this condition, which shows a sex-linked recessive mode of inheritance (Holthuis, 1943; Jonkers, 1950), fine, superficial punctate opacities of the cornea have been described, which, according to Franceschetti, Rossano, and Jadassohn (1956), Franceschetti, Jaccottet, and Jadassohn (1957), and Sendi (1957), occur primarily and not as a consequence of the scarcity of cilia.

As an almost isolated anomaly, ciliary alopecia is still less common, since, as far as we know, it has been observed but once (Traquair, 1912), in a father and son, in whom not only the eyelashes but the intermarginal area of the lower lid was absent.

On account of its considerable rarity, we feel justified in presenting the pedigree of a family in which the defect was strictly localized to the eyebrows and lashes and appeared in three generations (Figure). All the members of this family except the affected subject in the first generation were seen personally.

Case Reports

Case 1 (III, 4), an alert and well-developed 17-year-old girl, was said to have almost completely lacked cilia and brows from birth. She had had good vision until recently, when some difficulty in perceiving distant objects had been noticed.

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FIG.—Pedigree showing dominant inheritance of congenital lack of cilia and supercilia in three generations.

On examination, no lashes proper, but only scanty, extremely fine, downy, blond hairs were present at the upper lid margin; on the lower lid even these lanugo-like, rudimentary cilia were completely absent, but the margins were normal in all other respects. The eyebrows were also almost entirely missing, but the scalp exhibited a luxuriant hair growth, brown-black in colour, as did the other normally hairy parts of the body.

The eyes and their adnexa were otherwise normal. The visual acuity was 20/20 in each eye with correction -4.5 D sph., -2 D cyl., axis 0° in the right eye, and -3.5 D sph., -2.5 D cyl., axis 5° in the left.

No other physical abnormalities were present.

Inasmuch as the defect appeared to be due to a germinal disturbance, the patient's relatives were traced in order to establish whether it was sporadic or hereditary.

Case 2 (III, 5), a 15-year-old boy, younger brother of Case 1, in whom the eyebrows and lashes had always been absent, had complained of poor distant vision since the age of 13.

The clinical picture was exactly the same as in Case 1. In addition, a moderate degree of acne vulgaris was present on the face, back, and chest; no signs of hyperkeratosis follicularis were detected.

The visual acuity was 20/20 in each eye with correction -1.75 D sph.

Case 3 (II, 5), a 39-year-old woman, mother of Cases 1 and 2, gave a similar history of congenital lack of lashes and eyebrows and of some shortsightedness since puberty. She had been afflicted for the last few months with involutional melancholia and had entered a mental home.

The anomaly in this case was identical in all respects with that in the children. The media and fundi were normal, and, although the refraction could not be adequately determined, it was ascertained that a myopia of 2–3 D existed, because the retina could be seen clearly only by the interposition of such a concave lens.

No skin disorders nor other physical anomalies were present.

The father of Cases 1 and 2 was examined and found normal.

When the remaining sibs in the second generation (II, 1, 2, 3, and 4) were studied, two elder brothers of Case 3 (II, 2 and 3) were found to present the same defect but without myopia.

The other affected member of the pedigree, the maternal grandmother of Cases 1 and 2 was dead, but it was satisfactorily established that she had presented an identical picture. The grandmother and her husband were first cousins, but no information concerning her brothers and sisters or parents could be obtained.

Comment

The occurrence of congenital underdevelopment of the lashes and eyebrows in three generations makes it highly improbable—to say the least—that the anomaly might be due to environmental factors. The facts demonstrate that it is a genetically determined, regularly dominant trait. Although the simultaneous appearance of other disorders, such as slight myopia in Cases 1, 2, and 3, and mental deficiency or disease in Case 3, may be coincidental, it may indicate a polyphenic action of the pathogenic gene or a disturbed genotypic milieu.

Summary

A pedigree showing the transmission of a hereditary, dominant type of congenital ciliary and superciliary hypotrichosis is presented.

REFERENCES

- DUKE-ELDER, S. (1952). "Text-book of Ophthalmology", vol. 5, p. 4667. Kimpton, London.
- FRANCESCHETTI, A., JACCOTTET, M., and JADASSOHN, W. (1957). *Ophthalmologica (Basel)*, **133**, 259.
- , ROSSANO, R., JADASSOHN, W., and PAILLARD, R. (1956). *Dermatologica (Basel)*, **112**, 512.
- HOLTHUIS, P. (1943). *Ophthalmologica (Basel)*, **106**, 325.
- JONKERS, G. H. (1950). *Ibid.*, **120**, 365.
- LEFFERTSTRA, L. J. (1956). *Ibid.*, **132**, 204.
- SANCHEZ SALORIO, M. (1955). *Arch. Soc. oftal. hisp.-amer.*, **15**, 496.
- SENDI, H. (1957). "Quelques cas de Keratosis follicularis spinulosa decalvans (Siemens)". Thèse No. 2475. Imprimerie La Sirène, Geneva.
- SIEMENS, H. W. (1926). *Arch. Derm. Syph. (Berl.)*, **151**, 384.
- TRAQUAIR, H. M. (1912). *Ophthal. Rev.*, **31**, 138.
- ULLRICH, O., and FREMERIEY-DOHNA, H. (1953). *Ophthalmologica (Basel)*, **125**, 73 and 144.
- WESSELY, K. (1929). *Klin. Mbl. Augenheilk.*, **83**, 335.
- WEYERS, H. (1954). *Z. Kinderheilk.*, **74**, 468.