THE ARCUATE LENS OPACITIES OF MONGOLISM AND CATARACTS OF SIMILAR APPEARANCE *

BY

RONALD F. LOWE

Melbourne, Australia

IN 1934 Mann described six patients showing abnormalities of the iris and lens due to the persistence of capsulo-pupillary vessels. In some she actually demonstrated the persisting vessel, but more often it had atrophied after having produced the various deformities. There was always a gap or notch in either the ectodermal or the mesodermal part of the iris, sometimes in both, and evidences of persistent pupillary membrane were found either as tags from the lesser circle or as pigmented spots on the anterior lens capsule. In the lens there was usually a marginal notch and often an opacity localized to the region of the coloboma. Some of the opacities resembled small white arching bands within the lens (Fig. 1)[†], while others were gross sector-shaped cataracts (Fig. 2)[†].

Sometimes after facial blows a small arc of opaque fibres may be seen near the equator of a lens. The trauma may be indirect yet sufficient to disturb those cells dividing at the time. Clear fibres may be superimposed so that gradually the opacity becomes further from the surface. Slit-lamp examination much later in life allows the position of the opaque cells to be correlated with the size of the lens at the time of the injury (Fig. 5).

Another condition in which curved lens opacities are found is mongolism. During the examination of the lenses of 52 mongoloids previously reported (Lowe, 1949) I found eight with characteristic cataracts which I termed "arcuate opacities". Some of the mongoloid arcuate opacities presented appearances similar to those described by Mann (1934, 1938) but considerable differences were also evident.

The purpose of this paper is to extend the description of the mongoloid arcuate opacities. They are the most distinctive of the various types of cataract found in mongolism.

Arcuate Opacities in Mongolism

The simplest form of arcuate lens opacity shows as a small white splash deeply within the lens. Such a cataract can be seen

* Received for publication March 7, 1950.

† Figs 1 and 2 are reproduced from I. Mann (1937) "Developmental Abnormalities of the Eye". Cambridge University Press.

FIG. 1.—Curved lens opacity caused by persistence of a capsulo-pupillary vessel. Colobomata of the choroid, lens, suspensory ligament, and iris occur as associated defects (Mann).

FIG. 2.—Large sector cataract caused by persistence of a capsulo-pupillary vessel, the strand of which can be seen in the colobomata of the lens and iris (Mann).

FIG. 3.—Thin arcuate lens opacity in mongoloid. Case No. 54, aged 40 years. (a) Appearance with loupe. (b) Opacity seen by diffuse illumination with the slit-lamp microscope. (c) Optical section of lens with the slit-lamp showing opacity embracing the plano-convex areas in the deepest layers of the foetal nucleus.

FIG. 4.—Two separate series of arcuate lens opacities in mongoloid. Case No. 53, aged 40 years. (a) Appearance with loupe. (b) Appearance with high magnification with the slit-lamp.

FIG. 5.—Curved lens opacities due to trauma found in one eye of a woman aged 29 years. History of being struck upon the eye by a cricket ball at age 12 years. Headaches and poor vision since 15 years. Slit-lamp shows opacities in outer layer of adult nucleus corresponding with age 12 to 15 years.

FIG. 6.—Arcuate lens opacities seen in mongoloids. (a) Case No. 34, aged 27 years. (b) Case No. 61, aged 44 years. (c) Case No. 58, aged 43 years. Two concentric opacities are separated by a thin layer of clear lens fibres. (d) Case No. 36, aged 41 years. (e) Case No. 17, aged 11 years. (f) Case No. 16, aged 11 years.

In (e) and (f) the arcuate opacities have developed into dense sector cataracts.

by means of a loupe and focal illumination (Fig. 3 (a))* . It forms an arch from the axis, curving around the equator of the foetal nucleus in the position that the periphery of the lens had attained at that stage of development. The smallest arcuate opacities are very narrow and relatively thin (Fig. 3 (b)). With the slit-lamp they are found just outside the plano-convex areas in the deepest parts of the foetal nucleus. They may be practically flat-surfaced or have a central rib—probably from the remains of the vessel. They then resemble minute curved white leaves near the centre of the lens.

Sometimes the opacity may be duplicated, the more superficial band being separated from the deeper (Fig. 6 (c)). Occasionally clear lens may cover the deep opacity and the two layers be more widely separated. In other cases several opaque strata are separated by zones of clear lens fibres. The more superficial opacities do not exactly conform to the deeper shapes, because within different layers there may be slightly varying amounts of clear and opaque lens fibres. Superficial arcuate opacities always occur within the same sector of the lens as those seen more deeply. Arcuate opacities are not found in the outer layers of a lens in the absence of opacities beneath them within the foetal nucleus.

A whole sector of a lens may appear opaque without any apparent differentiation into distinct layers. The opacity then extends from near the centre of the lens as a wedge. Usually only one sector is involved but no definite restriction occurs. Fig. 4 shows two different arcuate opacities within the same lens. Involvement of the whole circumference would result in a dense central cataract if only the cells within the foetal nucleus became opaque, or in a total cataract if no clear lens fibres formed as the lens increased in size. In no instance was there observed any coloboma of a lens, any displacement indicating disturbance of the zonule, or any sector defect of the iris or the pupil. In these respects the arcuate opacities of mongolism are very different from those attributed by Mann (1934, 1938) to the persistence of capsulo-pupillary vessels.

The other cataracts found in mongolism, namely opaque Y sutures and flake opacities, may or may not be present in the same lens that shows an arcuate opacity.

Cause of the Arcuate Opacities.—A most fascinating feature of ocular embryology is the formation and disappearance of the vascular capsule of the lens. During the earliest phases of development the cells receive their nutrition by diffusion from the

* For easy reference Figs 3 and 4 are reproduced from this author's article in this Journal (March, 1949, Figs 14 and 16).

uterus. With enlargement of the embryo and organ differentiation this supply becomes inadequate and blood vessels are provided.

The lens vesicle is able to separate and the posterior lens cells are able to grow forwards to form the primary lens fibres without vascular nourishment. The earliest secondary lens fibres envelop the primary lens fibres and meet to form the Y sutures (8.5 weeks) only a few days before the vascular capsule of the lens is fully developed (9 weeks). For the following two months the developing fibres are nourished by blood vessels in direct contact with the lens surface. Their nutrition is intimate and essential. But by the fourth month the posterior vascular capsule and the capsulo-pupillary vessels begin to regress (Mann, 1938). The lens is then supplied by a more distant source presumably the ciliary body.

If some of the capsulo-pupillary vessels were hypoplastic during the critical period of lens nutrition from about the eighth to the sixteenth week, it is conceivable that the cells dependent upon these vessels for their nutrition would be damaged. Those most likely to be involved would be the subjacent equatorial cells in the act of dividing and elongating at the time. Such defective cells cannot be shed, and remain throughout life to register the time of their derangement.

In mongolism the severity of the disturbance is insufficient to prevent division of the equatorial cells and thus cause a coloboma lentis, but it is intense enough to interfere with their ability to form clear lens fibres. Hence a sector opacity is produced without coloboma. If the deficiency were slight, only the lens fibres in active growth at the time of the damage need be opaque, those laid down subsequently could be clear. In this way a thin arcuate opacity would form indicating the size of the lens at the time of the disturbance. From knowledge of the times of formation of the zones of the lens, slit-lamp examination can determine when the disorder occurred.

In the lenses that show a recurrence of opaque layers interspersed with clear zones it may be supposed that the equatorial cells suffered permanent damage. When their nutrition was optimal they could lay down clear fibres, but when the favourable balance was disturbed, as by an intercurrent illness or metabolic upset, the lens fibres developing during that period would become opaque. With recovery, clear lens fibres would be formed again.

The character of any arcuate opacity therefore depends upon the degree of original damage to the equatorial cells, and upon the subsequent health of the individual.

The arcuate lens opacities associated with mongolism were found to originate deeply within the foetal nucleus arching around the plano-convex areas on the same level as the deepest Y sutures.

This situation corresponds with a lens size of about 8.5 or 9 weeks, *i.e.*, the beginning of the period of its nourishment by the capsulo-pupillary vessels.

Discussion

A hypoplastic vascular system is a characteristic feature of the syndrome known as mongolism. Postnatally this may be readily seen in the eyes by the poverty of the iris stroma, and generally by a very poor peripheral circulation. Defects of the heart and great vessels are common and indicate anomalous development, particularly about the end of the second month. The revelation of abnormal capsulo-pupillary vessels shows that defective vascular development is present in the early capillaries as well as in the heart and great vessels. This suggests that in mongolism the whole vascular tree is subject to defects in its earliest stages.

Two important concepts arise. First, the abnormal vascular system links with other developmental anomalies to show that the state of mongolism in the developing child is already determined by at least the third foetal month. Secondly, the presence of an inadequate vascular supply is a potent cause of secondary structural and functional defects, some of which help to characterize the syndrome of mongolism.

In the eyes the ultimate results of abnormal capsulo-pupillary vessels are diverse. Opacities in one sector of a lens cause gross astigmatism. If each lens is so affected, accurate fixation is impaired and a searching or ocular type of nystagmus results. Unilateral involvement may determine monocular vision, strabismus, and amblyopia. With the exception of high myopia, lens opacities are the commonest cause of strabismus in mongolism.

Other organs must be subject to similar hazards during their formation so that defects due to inadequate vascular supply should be widespread,

Unfortunately, we are ignorant so far of the cause of the vascular anomalies. This is probably the cause of mongolism itself. It is possible that numerous agents acting at the appropriate time and with suitable intensity can separately produce a similar end result. Since 1866, when Langdon Down first introduced the mongol nomenclature, innumerable theories have been propounded about the cause of this disease, but they are all unsatisfying and disappointment persists. Mongolism is a disease full of riddles made more difficult by many inaccurate observations and biased conclusions. The whole picture is still incomplete although adequate clinical material is readily available in mental hospitals. The

interpretation of many observations awaits more detailed knowledge of normal and abnormal growth processes.

Summary

Curved lens opacities may be found associated with persistence of a capsulo-pupillary vessel, after trauma, and in mongolism. Different types of mongoloid arcuate lens opacities are described and illustrated. The cause of these arcuate opacities is thought to be hypoplasia of capsulo-pupillary vessels. The position of the opacities in the lens indicates that mongolism is established by at least the third month of pregnancy. If abnormal vessels during development can cause such severe structural and functional defects in the eyes of mongoloids, it is possible that some of the other stigmata of this syndrome may be dependent on abnormalities of early blood supply.

REFERENCES

- LOWE, R. F. (1949). *British Journal of Ophthalmology*, **33**, 131.
MANN, I. (1934). *Arch. Ophthalm., Chicago*, **11**, 174.
——— (1937). "Developmental Abnormalities of the Eye". Cambridge University Press.