COMMUNICATIONS

THE EYES IN MONGOLISM

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Langdon Down in 1866 postulated that certain groups of idiots gave indications of a regression from one racial type to another. In stating that "a very large number of congenital idiots are typical Mongols" he gave a name to one particular group. Langdon Down was influenced by the appearances of the face and especially by the shape of the palpebral fissures. His clinical observations were shrewd but his racial comparisons were very superficial and unfortunate.

For centuries mongolism and cretinism had been considered to be the same abnormality, but during the latter half of the last century many contributions from distinguished observers enabled the two conditions to be separated.

Although mongolism is now recognised as a definite clinical entity it is most important to remember that there is no single diagnostic sign. Like all syndromes its recognition depends on a
The number of stigmata, each of which varies between individuals. Tredgold (1947) stated: "Many ordinary aments and even normal individuals possess one or more of the peculiarities which go to make up mongolism. It is the combination which is characteristic."

The appearances of the eyes were among the first abnormalities to be recorded, and in this small region there are several characters which may assist diagnosis in doubtful cases. The purpose of this paper is to review these signs and extend the descriptions to cover many aspects only briefly reported in the past.

Growth

One of the most important changes in mongolism is the retardation of growth affecting almost the whole of the body. This feature was recognised by Séguin in 1866, and has received considerable attention from most writers. The deficiency is well described by Benda (1946) in his book "Mongolism and Cretinism." He reviews many aspects, and emphasises that although at birth the mongoloid baby is little smaller than normal, within the earliest years of life the difference becomes very striking, so that the mongoloid always remains considerably retarded. Many observers consider that there is a tendency to retention of foetal characters throughout life in mongolism. As the mongoloid is so much slower than the normal in development, numerous foetal and infantile characters are outgrown later, but many disappear if given sufficient time. In the past very few of these patients lived for many years, but with extended control of so many infectious diseases, and the great improvement in institutional life, the mongoloid now attains a much greater age.

In the present series of 67 patients examined, 40 were over 25 years of age and the oldest was over sixty.

The Skull

Four skulls of adult mongoloids were examined in the museum at Leavesden Hospital. They were prepared by Dr. R. M. Stewart when he was physician-superintendent. Their ages were as follows—case 68, aged 31 years; case 69, aged 35 years; case 70, aged 42 years; and case 71, aged 52 years.

Although these patients had lived well into adult life, all the skulls were remarkably similar in showing features which normally disappear during the second or third years. Abnormalities were prominent where marked changes occur with normal growth after birth.

All skulls had persistent metopic (frontal) sutures. The foramen lacerum medium was widely open in three skulls (Fig. 1, Cases
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No. 69, 70 and 71). In these the foramen ovale communicated freely with the foramen lacerum medium, while the foramen spinosum was not quite cut off. Foramina of Huschka were present in all tympanic plates.

In the accessory nasal sinuses development was very limited. The small maxillae and orbital ridges are obvious in the frontal and lateral photographs. This lack causes the flattened appearance of the face so well seen in life. In contrast with the other nasal sinuses, the ethmoid air cells were well developed in every skull (Fig. 4, Case No. 71).

The nasal bones varied in size but were always smaller than normal; sometimes they were almost absent (Fig. 2). In life the height of the nasal bridge is very variable. Usually it is flattened, but quite a number are of practically normal elevation.

The general thinness of the skull bones has been recorded by other observers (Greig, 1927; Benda, 1946). It was conspicuous in these specimens, and the orbital plates were very thin. The superior and inferior orbital fissures were very wide.

The characteristic shortness of the sagittal diameter is clearly visible in the photographs. It gives each skull a flattened and broadened appearance. In this shortening the orbits are somewhat reduced in depth, but their shallowness is not as obvious as expected.

The antero-posterior orbital axes of normal skulls are inclined to each other at an angle of about 45 degrees. In mongolism, because of the relatively greater diminution of the antero-posterior axis compared with that from side to side, the orbital axes are inclined at about 75 degrees. This follows because the apices of any orbits are very close to their medial walls, and as these measuring points move forwards the angle between the diverging axes increases. The angles measured in the four skulls were as follows: in case 68, 70 degrees; in case 69, 70 degrees; in case 70, 75 degrees; in case 71, 80 degrees. In all the skulls the horizontal orbital axes definitely sloped downwards and outwards as is normally found in European skulls. Although Whitnall's orbital tubercle could be readily identified as being higher than the attachments of the medial palpebral ligament, any change in relationship between the orbital bones could not be found as an underlying cause.

The cribriform plate was deeply sunken between the orbital roofs, giving a more oblique slope to the upper parts of the medial orbital walls and indicating considerable disturbance of growth in this region.

The skulls contain so many peculiarities that it is difficult to separate the secondary effects from those of a more fundamental nature.
The floors of the skulls of one normal and four mongoloid adults. The mongoloid skulls show general bony thinning, reduced antero-posterior diameter (brachycephaly) and absent frontal sinuses. The foramen ovale communicates with the foramen lacerum medium in Cases No. 69, 70 and 71.
The mongoloid skulls show persistent metopic (frontal) sutures, wide orbital fissures, very small nasal bones and maxillae. All horizontal orbital axes slope downwards and outwards.
Case No. 69, age 35 years.  
Case No. 71, age 52 years.  
Normal European adult.  
Case No. 70, age 42 years.  
Case No. 68, age 31 years.

**Fig. 3.**

Lateral views of the skulls of one normal and four adult mongoloids. The mongoloid skulls show smallness and flattening of the facial structures.
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The medial wall of the left orbit of mongoloid No. 71, showing well developed ethmoid cells.

**Fig. 4.**

The shape of the palpebral apertures has attracted attention for many years, and it was their appearance which was especially noted by Langdon Down when he introduced his Mongol hypothesis. This region has been frequently described, but some aspects still require a correct interpretation. Normal Europeans have the outer canthus slightly higher than the inner, and occasionally this difference may be accentuated sufficiently to give the appearance of a definite slant. From a large number of photographs of members of the true Mongolian races, Gifford (1928) found that the slope of their palpebral fissures was no greater nor any more frequent than in his own non-Mongolian patients. He concluded that the slant eye of the true Mongolian is a myth, and that instead the narrow or slit-eye is more characteristic.

Komoto (1892), writing of Japanese, stated that an epicanthus exists physiologically among them, and this gives an appearance of obliquity as it passes downwards and across the inner angle.

The eyes of the true Mongol have an almond shape due to the widest part being towards the inner angle; with the long palpebral aperture narrowed and tapered laterally. This is quite different from the mongoloid. The outstanding features of the mongoloid
palpebral apertures are obliquity and shortness (Figs. 5, 7 and 9). Less commonly narrowing is also present. The obliquity may be very pronounced, but occasionally it is absent. There is often some asymmetry between the two sides of the face.

The characteristic shortness of the palpebral apertures has been frequently recorded. The combination of obliquity and shortness

Fig. 5. Case No. 26, aged 20 years. Typical mongoloid palpebral apertures showing obliquity upwards and outwards, shortness, and even arch of upper lid margins.

Fig. 6. Case No. 40, aged 32 years. Mongoloid adult showing persistent epicanthus. The upper lid margins are evenly arched although the palpebral apertures are horizontal.

Fig. 7. Case No. 50, aged 39 years. Showing very short sloping palpebral apertures and flat nasal bridge, usual appearance without epicanthus.

Fig. 8. Case No. 50. Demonstrating loose skin over nasal bridge. Stretching from below produces folds like epicanthus.

quickly draws attention to this region of the face. Often the palpebral apertures are described as narrow, but narrowing is not particularly common unless there has been an underlying infective process. Trachoma has been the commonest cause because it produces ptosis, or swollen lid margins from chronic discharge.

The curve of the upper lid is worthy of description. Normally the upper lid margin has its highest point at the junction of the inner and middle thirds. The mongoloid upper lid has a much more gradual and even curve so that its highest point is at the
centre. This peculiarity in shape is seen whether the palpebral aperture is horizontal or oblique. The even arch is occasionally seen in normal adults, and is fairly common in those conditions involving disturbed growth of the head.

The cause of the obliquity of the palpebral apertures has received explanations that are unsatisfactory. Van der Scheer (1918)

![Image of eyes](image_url)

Case No. 69, aged 35 years.  
Case No. 71, aged 52 years.

Case No. 70, aged 42 years.  
Case No. 68, aged 31 years.

Fig. 9.

The palpebral apertures of the four mongoloid adults whose skulls are shown in Fig. 2.

The slope of the palpebral apertures is in the opposite vertical direction from that of the horizontal orbital axes.

considered the cause was dwarfism or absence of the nasal bones, the frontal bones sending processes downwards to make up the defect, carrying with them the covering tissues. Although the four skulls examined all had nasal bones smaller than normal, no downgrowths of the frontal bones could be seen. If the obliquity were dependent on the smallness of the nasal bones it should be present in many normal babies. This is not so, and European babies show no more obliquity than the adults.

From numerous X-rays of young mongoloids, Benda (1946) reported the presence of egg-shaped orbits with a slope of the orbital axes upwards and outwards. He inferred that the bones, cause the palpebral apertures to follow the same direction. But the four adult skulls here reported all had orbital axes that inclined definitely downwards and outwards, as is normal in Europeans.
Photographs of these patients taken during life show the presence of oblique palpebral apertures in all (Fig. 9). The slopes are undoubtedly in the opposite vertical direction from those of the orbital axes.

In the museum of the Royal College of Surgeons of England, an examination was made of the faces of 25 foetuses of gradually increasing development from 2 months to birth. A similar examination was made of 20 foetal skulls of the fifth month and later.

At the second foetal month the eyes are directed laterally, and the apertures between the developing lids slope markedly upwards and outwards. In the following month the eyes develop a more frontal direction, and the lid edges become apposed, although the line of fusion is clearly visible. These lines on each side assume a more horizontal direction, so that by the fourth month, when the eyes are almost directed forwards, the lids are inclined only slightly upwards and outwards. By the fifth month the eyes have reached their frontal positions and the lid lines are horizontal. The eyes and palpebral apertures maintain these relations throughout life.

During the fifth month, when the palpebral apertures have already reached their permanent stations, the horizontal orbital axes are still directed upwards and outwards. Their inclination likewise changes with growth. By the sixth month the slant has almost disappeared, and at the seventh month the alignment is straight. At birth most orbital axes slope downwards and outwards, but some are still horizontal. The change continues during postnatal development, and is conspicuous in almost all European skulls.

The upward slope of the orbital axes observed by Benda appears to be a retardation of development so commonly seen in mongolism, but in time the anomaly is corrected. As the slope of the palpebral apertures and that of the orbital axes change at completely different times during development, and as they are in the opposite vertical direction in adult mongoloids, the two must be independent. The cause for the peculiarities in the eyelids is much more likely to be in the skin itself rather than in the underlying bony configuration.

In the disease known as congenital ectodermal defect, there are numerous anomalies of the epidermis and its appendages because of faulty differentiation of the epiblastic layer. They may arise from incomplete development of the surface ectoderm or its absence in circumscribed areas. The palpebral apertures may be shortened, slope upwards and outwards, and present an appearance amazingly like that seen in mongolism. Some subjects are mentally defective, but most are normal, because the anlage of the nervous system is
distinct from the cutaneous ectoderm before the defect begins (Andrews, 1947).

In dermatomyositis and scleroderma severe skin shrinkage may cause changes in alignment of the palpebral apertures to resemble those seen in mongolism.

In mongolism skin texture is rarely normal, whilst exaggerated flexures and clefts are almost constant. The dermal patterns show many interesting features. Their form is determined before the third month of foetal life (Cummins, 1936).

The surface ectoderm is probably affected very early during development, and disturbances within it may be responsible for the appearances of the palpebral apertures so characteristic of mongolism.

**EPICANTHUS**

Epicanthus has been stressed by many writers, but there is nothing very unusual in this feature. It is common among European babies, but usually disappears during the early years of growth. The persistence of epicanthus throughout life may be genetically determined due to a pure or irregular dominant gene (Usher, 1935).

Von Ammon (1860) divided epicanthus into three main groups.
1. **Epicanthus supraciliaris**—the fold arises from the eyebrow.
2. **Epicanthus palpebralis**—from the skin of the upper lid above the tarsal fold.
3. **Epicanthus tarsalis**—from the tarsal fold. To these Komoto (1920) added a further type.
4. **Epicanthus inversus**—from the lower lid, enclosing the medial end of the upper lid in a small bow.

Von Ammon considered that epicanthus appears because the skin of the face develops at a much faster rate than the bones. It is loose in the foetal and infantile stages, but when the bones develop with the aging of the child the epicanthus disappears. Chavasse (1939) stated that its disappearance is due to growth of the face, a deepening of the orbits and an increase between them. These changes are said to depend on the development of the jaws, particularly with the onset and increase of dentition. The redundant tissue becomes less, and its looseness disappears.

Epicanthus palpebralis is the most common type seen in European infants. The same form occurs in mongolism. Although epicanthus is common among mongoloid infants, one cannot accept the statement of Brushfield (1924): "Of all the stigmata of degeneration of the head this is one of the most important, the lower the grade of amentia the more certainly will it be found." The epicanthus is merely a further indication of retarded growth in
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mongolism, and with development it almost always disappears. Benda (1946) wrote that he had never seen it beyond twelve years of age. In the present series of adults it was only seen once in an obvious form (Fig. 6).

More commonly in mongolism an abnormal looseness of the skin remains in this region, so that when it is stretched from below, folds similar to epicanthus can be produced. Fig. 7 shows case No. 50 as his usual appearance with epicanthus; but with stretching the folds show prominently (Fig. 8). In mongolism the bridge of the nose generally remains flat and broad, but this feature is rather variable, and some of the mongoloids have well-developed nasal bridges.

THE LID EDGES

Young mongoloids have thin normal lid margins with sharp borders in contact with the eyeballs, and a narrower shelf between this ledge and the lashes. During adolescence the skin often becomes rougher, and this ciliary border may thicken, so that a wider ledge projects between the sharp edge and the lashes. On this skin ledge scales may collect and lead to a secondary conjunctivitis. Increase of subcutaneous fat between the skin folds frequently gives a rolled appearance to the lower lids (Fig. 7).

The eyelashes may be rather short, but are otherwise normal unless disturbed by disease. Trachoma has been the commonest cause of gross irregularities. Alopecia affecting the scalp is moderately common. Sometimes it is almost complete, and the eyelashes may be absent (Case No. 50, Fig. 7).

THE LACRIMAL APPARATUS

All the eyes watered profusely after slit-lamp examination, and tears formed readily when the children cried. There was no evidence of deficiency in tear secretion. The drainage mechanism appeared adequate, and epiphora was seen only when there was excessive tear formation, e.g., in old trachoma.

THE IRIS

The fact that there are peculiarities of the iris in mongolism has been known for many years. Many investigators in describing the widespread stigmata refer to the "speckled iris." Brushfield (1924) reported briefly on the iris appearances in 115 cases. He described two main groups, namely the "mottled or marbelled iris" and the "speckled" iris. He observed that the changes were not obvious in the brown irides. He found that approximately 50 to 60 per cent. had blue or grey irides, about 25 to 30 per cent. had brown irides, and 10 to 15 per cent. had hazel-coloured irides.
In the present series of 67 patients, whose ages ranged from one to 60 years, the following iris colours were observed:

<table>
<thead>
<tr>
<th>Colour Description</th>
<th>Per cent.</th>
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<tbody>
<tr>
<td>Blue or Grey (little stromal pigment)</td>
<td>40</td>
</tr>
<tr>
<td>Hazel (moderate stromal pigment)</td>
<td>25</td>
</tr>
<tr>
<td>Light Brown (heavy stromal pigment)</td>
<td>25</td>
</tr>
<tr>
<td>Dark Brown (dense stromal pigment)</td>
<td>10</td>
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</tbody>
</table>

The colour of any iris depends on the amount of brown stromal pigment present giving its brown colour by reflected light, and on the amount of light scattering caused by the stroma with the densely pigmented epithelium backing it (producing blue or grey colours).

The stromal pigment is seen four to five months after birth in Europeans, first in the cells on the outer iris surface. It increases in amount up to 8 years of age, and in later life diminishes again (Mann).

When a hazel iris is examined with the slit-lamp much of its green colour disappears, and the iris shows more light-brown. This is because there is much less light scattered from the focused beam than with less regular oblique illumination. The change in colour is well illustrated in the iris in Fig. 13, which appeared yellow-green by oblique illumination and golden-brown with the slit-lamp. The colour of the iris does not appear to be of great significance because its appearance normally varies with age, race, and method of examination.

There are two characteristic changes found in the iris in mongolism:

1. Poverty of the stromal fibres.
2. Peripheral speckling.

Like all stigmata of this disease, the appearances are not confined to mongolism. They are also found in normal people and in those with other diseases, but their presence in such a high proportion of mongols allows these appearances to be considered definite stigmata of this disease, and they count as supporting evidence in diagnosis.

**The Poverty of the Iris Stroma.** The peculiar, thin iris stroma has not been described previously as a characteristic of mongolism. Among the 64 patients comprising the present series, it occurred to a considerable degree in 60 (approx. 95 per cent.).

Similar to normal senile thinning it increases with age, but by contrast it is present in the youngest children. The most characteristic region to be affected is the periphery (ciliary zone). Around the pupil, most mongoloid children show almost normal
pigmentation, with well-formed stroma and collarette, but near the junction of the middle and outer thirds of the iris surface the stroma suddenly becomes much thinner, the strands diminish in number and thickness, and become very wavy. The dark posterior pigment epithelium shows plainly from behind (Fig. 13). The peripheral strands resemble minute fibres of fluffy white wool. Some show a fine pink blood-column down the centre. Only the thickly pigmented, dark brown irides fail to show this peripheral thinning (4 cases out of 64).

With increasing age depigmentation occurs together with the disappearance of some of the more central strands. A general thinning of the iris stroma then becomes obvious. Pigment remains only in association with the coarse strands, or where strands are bunched together. The light-brown or hazel iris changes slowly in colour to a dark-grey. With the depigmentation and thinning of the stroma the sphincter muscle shows clearly surrounding the pupil (Figs. 10, 11 and 12).

In one patient, case 65, aged 47, some of the strands were coarse near the pupil, but as they passed peripherally they became very thin. A few had apparently become detached from the main iris layers, and had gained a secondary attachment to the cornea. They passed forward to join the posterior corneal surface just medial to the limbus (Fig. 12).

Sometimes, as the attenuated peripheral fibres disappeared leaving the coarser strands, a scalloped design remained with the dark posterior pigment epithelium showing plainly in the depths (Fig. 11).

When coarse stroma had not been present, as in blue irides, the thinning of the fine strands formed a delicate lace pattern. Each fibre then resembled very fine wool, and the central blood column was frequently seen.

Uncommonly (2 cases, approx. 3 per cent.), stromal atrophy was very pronounced around the pupil as well as at the periphery. The pupil was not significantly altered in shape, but the posterior pigment epithelium showed as a thin, flat layer in association with the sphincter. With the slit-lamp densely pigmented clump-cells were prominent on its surface.

The Speckled Iris. The speckling is associated with the peripheral thinning, but is not dependent upon it. It is sometimes seen in normal people where the thinning appears to be absent. It is then usually not so obvious, and is very uncommon in association with other than blue irides.

In young mongoloids, just beyond the region where the stroma pigment suddenly becomes thinner, the strands tend to bunch at almost regular intervals around a circle concentric with the pupil.
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Where these strands run together, pigment collects or the strands remain thicker, thus presenting an appearance of small golden or white splashes.

This speckling was found in 56 patients in the present series (nearly 90 per cent.). Its appearance has been noted by previous observers, and it is considered a characteristic of mongolism. When the eyes are moist it gives a most attractive appearance to shining blue eyes. As atrophy advances the speckling remains much longer than the stromal pigment around the pupil, and in no case had it completely disappeared. It is not seen in the evenly pigmented dark brown irides.

Conclusions concerning the Iris. As the iris changes are found in the youngest mongoloids the appearances suggest a possible hypoplasia of the peripheral parts of the stroma. As the thinning continues throughout life, and in later years is very pronounced, an atrophic process is probably superimposed.

In any iris most of the strands consist of relatively thick-walled blood vessels. In mongolism the vascular system is remarkably hypoplastic. Structural abnormalities of the heart, narrowing of the main trunks, and inadequate, thin peripheral capillaries are very common. The hypoplasia of the iris stroma, with its peripheral thinning and fine woolly strands showing central blood columns, is in keeping with the widespread changes in the whole vascular system.

In normal people the iris becomes thinner with age, and the changes are accelerated with the onset of senility. In many ways adult mongoloids appear to deteriorate more rapidly than normal, and the advancing iris changes are perhaps merely another example of this decline.

In contrast with the stroma, the posterior pigment epithelium of the iris is avascular, develops from a different germ-layer, and is unaffected. It always appeared very deeply pigmented, and by transillumination no thin patches were seen. The ectodermal iris muscles are apparently unaffected, as the pupil reactions are not appreciably altered.

The Pupils

The pupils showed no abnormalities in size, position or shape. Their light reflexes (direct and consensual) and near reflexes (accommodation and convergence together) were brisk and appeared normal.

At the first examination many mongoloids may show somewhat enlarged pupils. The dilatation is apparently due to fear of the strange examiner and his instruments. They showed many other evidences of fear, but these disappeared as confidence was gained.
by repeated examination, and when they knew they would not be hurt.

With homatropine and cocaine the pupils dilated evenly and to a normal extent. As with normal people, the dilatation was less complete in some of the older patients.

The normality of the pupils is surprising in view of the changes in the iris stroma, but it is understandable when the limits of the hypoplasia are known.

The Interpupillary Distance. Brushfield (1924) found that the average interpupillary distance in 84 mongoloids was 5.2 cm. in males and 5.4 cm. in females. The mean distance between the pupils among unclassified mental defectives was 4 cm. Probably most of the subjects were children.

In the present series the average interpupillary distance of 17 fully grown adult, non-squinting mongoloids was 55 millimetres, with extremes of 52 and 60. These distances are in keeping with those found by Brushfield. These readings are only approximate because the eyes waver, and the method of measurement is a pair of calipers extending from the edge of one pupil to the corresponding point on the other pupil. The eyes are set about 5 millimetres closer than in normal adult Anglo-Saxons, but the general stature and head size of the mongoloids are smaller.

During growth the interpupillary distance and the skull breadth do not extend evenly, and the increase of each does not necessarily occur at the same time. In mongolism the growth of the skull in breadth is much less affected than in other directions. Sometimes the interpupillary distance seems to lag behind the increase in the interparietal diameter, and the eyes then appear set abnormally close together.

Lens Opacities

Ormond (1910) was the first to describe the typical lens opacities of mongolism. He examined 28 cases and found lens changes in nineteen. Subsequently (1912) he extended his series to 42 patients, finding 25 with some form of cataract. All his investigations were conducted with oblique illumination, and he observed that some of the opacities were very thin, being invisible if the light were not sufficiently oblique. He described most of the cataracts as a dot variety in lamellar form. Some of his illustrations show Y-shaped sutural opacities, although he did not describe them as being related to these structures. He also found an incomplete variety existing as a single circumscribed opaque mass in one meridian of the lens. He observed that the opacities did not reach to the periphery of the lens. Contrary to the findings in the present investigations, his illustrations show them to be thickest.
Fig. 10. Iris of mongoloid No. 43, aged 35 years. Blue iris showing white speckling, fine stroma, posterior pigment epithelium and sphincter.

Fig. 11. Iris of mongoloid No. 47, aged 37 years. Thin grey iris showing white speckling, and very pronounced atrophy of stroma at periphery (ciliary region).

Fig. 12. Iris of mongoloid No. 65, aged 47 years. Thin grey iris showing advanced stromal atrophy and thick strands becoming very thin towards the periphery.

Fig. 13. Iris of mongoloid No. 39, aged 32 years. Hazel iris, thickly pigmented around the pupil where the stroma is coarse, showing characteristic thinning of the stroma towards the periphery.
Fig. 14. Lens of mongoloid No. 54, aged 40 years. (a) Appearance with loupe showing flake opacities and small deep arcuate opacity. (b) The arcuate opacity as seen by diffuse illumination with the slit-lamp microscope. (c) Optical section of lens with the slit-lamp, showing the arcuate opacity in the deepest layers of the foetal nucleus and flake opacities in the infantile and adult nuclei.

Fig. 15. Lens of mongoloid No. 33, age 26 years. (a) Appearance with loupe showing numerous flake opacities and axial stellate cataract. (b) Optical section showing stellate splashes in the same layers as the other flakes (infantile and adult nuclei).

Fig. 16. Lens of mongoloid No. 53, aged 40 years. (a) Appearance with loupe showing flake and arcuate opacities. (b) Appearance with high magnification with the slit-lamp. The arcuate opacities have been reduplicated several times. The peripheral coronary splashes have a pallisade appearance and the small flakes are superficial to the deep arcuate opacities.
FIG 17. Lens of mongoloid No. 32, aged 26 years. (a) Appearance with loupe showing Y sutural cataract and peripheral splashes. (b) Magnified view of Y suture cataract and flakes. (c) Optical section showing Y suture cataract in the deepest layers of the foetal nucleus, with flakes of different colour and size localised to the infantile and adult nuclei.

FIG 18. Lens of mongoloid No. 48 aged 37 years. (a) Appearance with loupe showing numerous small blue flakes, especially towards the periphery. (b) Optical section without magnification showing flakes in distinct bands in the infantile and adult nuclei. (c) Optical section with slit-lamp microscope showing different sized flakes thickly spread in the infantile and adult nuclei. The Y suture opacity is only seen with high magnification.
FIG. 19. Lens of mongoloid No. 41, aged 32 years. (a) Appearance with loupe showing small flakes and posterior polar cataract. (b) Optical section without magnification showing flakes within the adult and infantile nuclei, fine opacities in the foetal Y suture and posterior polar cataract. (c) Magnified view of feathery Y suture cataract extending in depth throughout the foetal nucleus. (d) Denser opacities in the less regular sutures of the infantile and adult nuclei. (e) Magnified appearance of flakes showing different colours and bizarre forms, thickest towards the periphery.
at the axis. From the absence of these opacities in very young mongoloids he concluded that they were of late development.

Van der Scheer (1919) reported 60 cases examined for opacities. He found cataracts in 36 of them and confirmed Ormond's observation of their absence in mongoloids under eight years of age. He divided the opacities into three types:

1. Punctate, macular and floccular opacities.
2. Snow-flake cataract (cataracta punctata disseminata).
3. Y-shaped opacities, which he recognised as being related to the sutures in the lens of the new-born.

Ormond and Van der Scheer were the only investigators to describe more than a few cases in any series. Jeremy (1921) reported a further case, which probably had bilateral dense lamellar cataracts. Leeper (1912) published a series that is strange in having characters identical in every way with those reported by Ormond (1910). Brushfield (1924) mentioned three cases, but gave no details of the appearances. Ormond and Van der Scheer conducted their examinations without the aid of the slit-lamp. With this instrument and the increased knowledge that had been built up by its use it became possible to extend the observations and more accurately to localise the layers involved.

Koby (1924) examined one patient, and Goulden (1928) reported three more.

The Present Investigation. In all, 52 patients were examined. All were examined with oblique illumination and binocular loupe, (x3) and with their pupils fully dilated. Thirty-one were further examined with a slit-lamp. For the oblique illumination an electric ophthalmoscope was used after removal of the lens head. This could be readily moved in different directions and held so that the light was thrown into the lens almost tangential to its surface. The extreme obliquity was necessary to see the thin flake opacities when they were few in number. If the light was too direct it passed through the flakes, and unless they were relatively numerous they were easily missed.

The mongoloid patients soon lose their fear of the instruments, but one cannot expect their eyes to be held steady for more than a few seconds. With the slit-lamp one has to chase the continually moving eyes, or at times wait for the lens to return to the focused position.

The 31 patients examined with the slit-lamp varied in age from 20 to 51 years. The slit-lamp was not used for children, as it was considered that little extra information would be gained for the greatly increased difficulties. The oldest patient, aged 60 years, was unable to be moved from bed, and some were unsuitable subjects because of trachomatous pannus.
Types of Cataract. The cataracts were found to fall into 4 main groups.

1. Arcuate opacities—commencing during the formation of the foetal nucleus.

2. Sutural opacities—mainly affecting the anterior Y sutures of the foetal nucleus, but occasionally showing in later sutures.

3. Flake opacities—characteristically within the infantile and adult nuclei.

4. Various congenital cataracts with individual differences.

The Arcuate Opacities. The arcuate opacities are the earliest to develop, as they show plainly within the foetal nucleus. The simplest form is seen as a small white arc deeply within the lens (Fig. 14, Case No. 54). Such a cataract appears to have been formed in relation to an abnormal capsulo-pupillary vessel during early foetal life.

The capsulo-pupillary vessels begin to regress during the fourth foetal month, and their disappearance is complete at birth or a little later (Mann, 1937).

The smallest arcuate opacities are quite narrow and relatively thin. They arch around the equator of the early layers of the foetal nucleus, and extend radially anteriorly and posteriorly. They may be practically flat-surfaced, or have a central rib—probably from the remains of the vessel. They then appear like tiny curved white leaves deep in the lens. Such appearances suggest a cause that has acted for a short time but caused no permanent damage to the growing cells.

Sometimes the injury is more severe so that all the lens fibres laid down in the affected region become opaque. A large, white sector-shaped opacity then develops extending from the foetal nucleus right out to the periphery. In these cases no coloboma of the lens was found in the affected part, but considerable lenticular astigmatism was evident.

If the disturbance that causes an arcuate opacity were to act over the whole of the circumference of the foetal lens instead of being localised to one sector, a lamellar (zonular) cataract would form. The lens cells might recover and lay down clear lens fibres again, or they might continue to produce opaque fibres showing as a completely white lens.

At other times the developing cells in the affected region appear to recover from the initial disturbance and lay down clear lens fibres, but apparently they do so with reduced vitality. Some future upset, possibly a metabolic disturbance or an intercurrent illness, causes the production of opaque fibres again for a short time, with recovery and clear fibres again later. In this way the original opacity may be reduplicated several times within the lens.
THE EYES IN MONGOLISM

at different depths between the foetal nucleus and the surface (Fig. 16, Case No. 53).

One arcuate opacity was found in a child of 11 years. It was very dense and appeared likely to have been present at birth. These cataracts are the earliest type to be seen in mongolism, and could be expected to be found at any age. In the present series of 52 cases (103 eyes) they were detected in 8 patients (11 eyes).

The Sutural Opacities. The Y sutures of the foetal nucleus are recognisable at 8·5 weeks of foetal development (35 mm.), and this pattern persists until just before birth, when the adolescent (infantile) nucleus begins to form with 4 rays instead of three (Mann, 1928).

In the present series 14 patients had opacities of the Y sutures. In practically all, the changes were bilateral. The anterior upright Y was always clearly identifiable (Fig. 17, Case No. 32), but the posterior inverted Y was always less distinct. The limbs of the anterior Y always showed plainly, but on the very few occasions when an opacity was present in the position of the posterior Y it showed as a faint irregular blur.

The youngest patient to show a sutural opacity was aged 11 years. Two children aged 13 and one aged 17 showed similar opacities. In these young patients the sutural opacities were extremely delicate and would have been missed had a special search not been made for them. For the most part, similar cataracts were quite obvious in adults. They therefore gave the appearance of developing within the foetal sutures some years after the sutures were laid down, and of thickening very slowly to become obvious only many years later.

It is to be expected that slit-lamp examination of young children would reveal some of early appearance, because in some of the adults their presence was not recognised with the loupe, and only found with the slit-lamp (Fig. 18, Case No. 48). With the loupe, when these opacities are very fine they appear like tiny strands of cobweb. When denser they are blue, and the little Y is easily seen. The slit-lamp shows that they extend for varying depths within the foetal nucleus. They are usually linear in form (Fig. 17), but sometimes they have a feathery appearance as the narrow beam moves across (Figs. 18 and 19). A few showed the more superficial parts like strings of small green beads. (Fig. 19).

Only four cases showed definite opacities of more superficial sutures. They usually appeared as very fine grey lines extending irregularly from the axis at varying depths within the infantile nucleus.

Flake Opacities. Vogt (1921) described wreath-shaped or coronary cataracts and found them in 25 per cent. of normal
adults. In the present series of 52 patients, opacities of this type were found in forty-five. If the patients under 14 years of age are excluded, flake opacities were found in 43 out of 47 (92 per cent.).

Examination with oblique illumination and binocular loupe shows two types of flakes. The commonest form consists of fine blue or smoky-grey minute flakes (snow-flake cataract of Van der Scheer). In young mongoloids they are very sparse. The pupils must then be dilated, as they are commonest somewhat internal to the equator, and the axial region remains free until they become relatively numerous. They are readily seen in most adults.

The other form of flake cataract is found as equatorial, white or light-brown radial spokes. They resemble the more usual coronary cataracts, but they are mostly lanceolate in outline and rarely club-shaped. Often they arch around the equator of the affected nucleus. Sometimes they are closely packed and have a palisade appearance involving certain sectors of the lens (Fig. 16). The peripheral splashes rarely extend towards the axis, but sometimes they do so, particularly in the posterior layers.

The small flakes and peripheral splashes are usually associated, but in very varying proportions. Arcuate opacities, sutural cataracts and flake opacities may be seen within the same lens.

With the slit-lamp the flake opacities are localised mainly within the infantile and adult nuclei. Very occasionally they extend more deeply and may be seen with the Y-shaped opacities within the foetal nucleus. Rather more often they may be found superficially, gradually thinning as the outer layers of the cortex are reached.

When a narrow beam from a slit-lamp is shone into a typically affected lens, and the result observed with the naked eyes, the affected zones stand out very prominently as anterior and posterior bands within the lens. A lamellar appearance is then presented (Figs. 15, 18 and 19).

With the binocular microscope, the flake opacities are seen to be very much more numerous than previously expected, and to assume extremely variable forms. Many appear like tiny blue or white snowflakes varying in size up to about one quarter of a millimetre. They are often thicker at the edges, and some appear as bizarre rings. The thicker opacities are more opaque, suggesting small drops of paint (Fig. 19).

Dust-like particles are very numerous. Possibly fine crystals are also present, because occasionally the beam is reflected as a tiny, brilliant, red or green light like a minute spangle.

The equatorial splashes are larger, but localised within the same nuclear layers. Infrequently they may extend from the axis, giving a stellate appearance to the cataract (Fig: 15).

The colour of the flake opacities may be blue, grey, green, white
or light-brown. The colour probably depends on the different ways that the light is dispersed. By their appearances they may be classed as forms of coronary cataract.

These opacities are as a rule sharply localised within the infantile and adult nuclei, rarely being found deeper or more superficially. The deeper layers of the affected bands show no denser collections and no larger particles than the later-formed more superficial layers.

As the flakes are rarely found near the lens surface, irrespective of age, it appears that they gradually form after the lens fibres are laid down, but once they have reached their distinctive thickness and shape, little further change occurs.

In their investigation the youngest patient showing them was aged 9 years, but the children were not examined with the slit-lamp. As the flakes are occasionally seen within the foetal nucleus of adults, some infants would probably show their presence by slit-lamp examination. Although they are very sparse in children they are almost always seen in adults. They bear no direct relationship to age. The oldest patient, Case No. 67, aged 60, showed few of them and they were most numerous in Case No. 49, aged 38 years (see Table No. III).

Hess (1893) showed that the anatomical basis for coronary cataract is lacunae between the lens fibres. The spaces are filled with homogeneous or finely granular coagula that stain deeply with haematoxylin. According to Vogt the opacities consist of fluid, and the form of the opacity depends upon its location in the concentric zones, i.e., if the vacuoles lie peripherally, club-shaped opacities result, whilst axially located vacuoles cause disc or ring shapes. Within the fluid there may be cellular debris, crystals, protein or lipoid particles. The thickness and fluid or particulate contents determine their optical effects upon the light beam and their different colours.

The flake and sutural opacities are probably of the same nature but in different location, whereas the arcuate opacities are of another character. It is important to emphasise that the appearances of the flakes, being purely optical phenomena, give no indication of their underlying cause.

The presence of large numbers of flakes is consistent with adequate vision, for owing to their thinness they obstruct little light. When they were very numerous the fundus could not be seen with an electric ophthalmoscope, and visual acuity must have been considerably reduced. The central opacities have a very unfavourable affect on vision.

As the older patients require relatively little vision for their interests, extremely few of them need any surgical attention. Even
those with dense central cataracts are able to manage in limited surroundings.

**Uncommon Congenital Cataracts.** The presence of mongolism does not preclude the formation of better-known types of congenital cataract. Five eyes were thus affected in three patients. Case No. 16, aged 11 years, had a typical dense right disc-shaped cataract. Thick bilateral lamellar cataracts were present in Case No. 56, aged 42 years. In childhood his lenses had been needled, but the results were poor. Typical posterior polar cataracts from the imprint of the hyaloid artery were found in each eye of Case No. 51, aged 39 years (Fig. 19).

These cataracts may be directly associated with mongolism or due to an independent cause. Some congenital cataracts can be genetically determined, their presence in a mongoloid being purely fortuitous. It is not known whether any disease can be responsible for both mongolism and congenital cataract (e.g., rubella).

**Similar Cataracts in Other Diseases.** The flake and dust opacities are usually considered to be similar to the cataracts occurring in other diseases (so-called endocrine cataract). Suggestive similarities are found in many cases with myopathy (e.g., myotonia atrophica) but the myotonic cataracts have some differences.

Several patients with myotonia atrophica were examined with the slit-lamp. In every case the cortex was involved more than the deeper layers. Compared with mongolism these appearances suggest that the opacities form more quickly after the fibres are laid down. The faster progress in myotonia is also shown in the usually rapid advance to mature total cataract. Many myotonics require operation to restore vision, as the loss is so severe. Few, if any, mongoloids require surgery, as most of the opacities progress little.

Twelve cretins aged between 26 and 61 years were examined. Most of these patients were receiving thyroid, but their clinical appearances were typical of congenital sporadic cretins. Arcuate or sutural opacities were absent. In eight cretins flake opacities were seen with the slit-lamp, but they were much less numerous than those seen in mongoloids of similar age. The flakes were visible with the loupe in four cretins. The most frequent site for the flakes was the adult nucleus, but sometimes fine specks were seen in the deeper parts of the cortex. Eight of the cretins showed a very marked zone of disjunction between the cortex and the adult nucleus reminiscent of a very thin sheet of mica. This appearance was not seen in mongolism.

Pituitary infantilism may cause lens opacities, and one patient was examined. She was aged 29 years, but had the appearance of a girl about fourteen. Unfortunately she had only one eye, the
other having been lost from an injury. The opacities were minute flecks just visible with reflected light from an electric ophthalmoscope. With the slit-lamp numerous very fine particles were seen in the deepest layers of the cortex and outer zones of the adult nucleus. Scattered flecks extended forwards into the more superficial parts of the cortex. A few peripheral coronary splashes were present. The appearances were not like those seen in any mongoloid, but more like those found in some of the cretins. A further resemblance was the presence of a band of white specks just outside the embryonic nucleus. These central specks were seen in three of the cretins, but in only one of the mongoloids, and he was reported to have suffered from rickets in infancy. The flecks were more numerous than in the anterior axial embryonic cataract (Vogt).

These lens appearances may be important in view of the suggestion by Benda that mongolism is due to a hypopituitarism. This isolated observation does not support such a view, but it would be interesting to investigate the lenses of more patients with pituitary deficiency diseases.

Conclusions concerning the Cataract. The lens is an exceptional organ in that none of the cells formed during its development is normally destroyed or replaced. As it is a clear structure, any opacity occurring within it from the earliest weeks of the embryo to the last days of post-natal life can be localised in time. The earliest cells are at the centre, and the later-formed fibres are superimposed like the layers of an onion. The time at which the different layers form is known from histological and slit-lamp examinations.

The developing lens separates from the surface ectoderm about the 10 mm. stage (4 weeks+) (Mann, 1937), and the primary lens fibres begin to form a few days later. The earliest fibres remain as a clear zone at the centre of the lens.

A disturbance of the formation of the primary lens fibres is said to cause a disc-shaped cataract (Collins, 1908). One patient (Case No. 16, aged 11 years) showed a typical disc-shaped cataract of his left lens. This might indicate abnormal development at the very early stage of 5 to 7 weeks. Unfortunately our knowledge of disc-shaped cataract is not precise, and as this type of cataract was seen in only one eye in the present series, its occurrence with mongolism might have been fortuitous.

The most distinctive cataracts in mongolism are the arcuate opacities. They were seen in one-sixth of the patients investigated. They are extremely rare in normal people or in those with other diseases. Similar opacities have been reported in association with coloboma iridis where an abnormal capsulo-pupillary vessel caused
extensive developmental defects (Mann, 1934). In mongolism these associated disturbances of other parts of the eye are lacking. The deepest layers of the arcuate opacities are found encircling the early secondary lens fibres at the same level as the most internal parts of the Y sutures. This corresponds with the 35 mm. stage (8-5 weeks) of foetal life. At this time the vascular capsule of the lens is well formed, and presumably the developing lens fibres depend upon the vessels for their nutrition. A few weeks later the vascular capsule begins to regress, and the lens then obtains its nutrition by fluid exchange from more distant sources. This change allows light rays to be unimpeded in their passage to the retina.

Abnormal vessels during development would be most likely to cause defects in lens structure during the period when they are supplying the lens fibres directly, and the fibres most susceptible to damage would be those dividing at that time. The arcuate opacities are examples of this disturbance being caused by the presence of abnormal capsulo-pupillary vessels. They provide proof that in mongolism the peripheral vessels may show abnormalities as early as 8-5 weeks. This is in keeping with developmental defects seen in other parts of the mongoloid vascular system, e.g., heart. The arcuate lens opacities are different from the other distinctive cataracts found in mongolism, as they are probably due to opaque lens fibres and not to opacities between the fibres.

In mongolism the sutural and flake cataracts are much more common than the arcuate opacities, but they are also found in normal people and in those with other diseases. Their incidence is unusually high in mongolism. The peculiar shapes of the sutural and flake cataracts are due to fluid and débris between the lens fibres. As their appearances are purely due to their optical properties, there is no justification for assuming that they are caused by the same factors that produce similar appearances in other diseases. They are often thought to be caused by those endocrine disturbances which are so extensive in mongolism. The cause is almost certainly a metabolic peculiarity, but its nature is unknown. A satisfactory explanation will be provided when more is known of the biochemistry and physiology of the lens. Until then the account must be purely descriptive.

The Intra-ocular Fluids

By clinical examination no abnormalities were detected in the aqueous. The intra-ocular pressure always appeared normal to palpation, and no evidence of glaucoma was seen. The vitreous was always free of developmental structures as observed with the
THE EYES IN MONGOLISM

ophthalmoscope. The only pathological changes were those associated with myopia, when vitreous floaters were commonly seen.

THE FUNDUS OCULI

The optic disc varied no more than would be expected in the examination of a random sample of the general population. Many of the discs appeared pronouncedly pink, like those commonly seen in children. Possibly it was this appearance that gave some of the older writers (Oliver, 1891) the impression that neuro-retinitis was common, but there is nothing to support such an interpretation.

Small congenital crescents and pigmented arcs were fairly common, but von Szily has shown that such conditions occur in 25 per cent. of normal people.

Typical myopic crescents were found in those with high myopia. They were associated with the usual fundus changes, i.e., thinning of the choroid, large choroidal vessels clearly seen, patches of atrophy and pigmentation.

The macula was carefully examined when it was not obscured by lens opacities. The foveal reflex was constantly present, and ophthalmoscopic appearances always suggested that it had differentiated anatomically.

Apart from fundus degenerations associated with myopia, anomalies were rare. Case No. 29, an extremely low-grade ament, showed an incomplete choroido-retinal coloboma extending downwards and outwards from the right optic disc, which had an eccentric pit in the same direction. Case No. 67, aged 60 years, had typical gyrate atrophy associated with high myopia. The clear-cut, atrophic, circular patches extended around both discs and maculae. Fixation had been lost with the right eye.

VISUAL ACUITY

Very few mongoloids can be tested for visual acuity by the usual clinical methods. Most of them never learn to read, although many more could be taught if special attention were given to their slow development. Only 5 of the 67 patients could read Snellen's Test Chart (see Table No. II).

The best visual acuity recorded was 6/12 right and left eyes in a male, aged 34 years, I.Q. 34, who had only a few lens flakes. By retinoscopy he had almost no refractive error. For reading he brought the type to a distance of about 6 inches from his eyes, to gain the benefit of larger retinal images, and then read J.1. The remaining four patients had less clear vision, which improved little with attempted correction after refraction. Subjective testing was
impossible, because they could not grasp the significance of "better or worse" when lenses were changed.

The attainment of 6/6 vision is a conditioned reflex which depends on appropriate stimulation and reinforcement (Chavasse, 1939). The development of such a conditioned reflex is subject to many hazards in mongolism. Some of the commonest may be mentioned. The reception of stimuli by the appropriate sensory end organs at the maculae may be disturbed by congenital lens opacities (arcuate, lamellar, posterior polar), or the retinal image may not be clearly focused, producing insufficiently defined stimuli (congenital myopia, astigmatism, nystagmus). Sufficient anatomical development, awareness and docility are further requirements for the development of any conditioned reflex (Pavlov). All are variable and in some cases inadequate in mongolism. In later life the onset of strabismus, developmental lens opacities, myopic degeneration and trachomatous pannus cause further visual loss.

The lower grades of ailments will be found to have very poor visual acuity. Some can only fixate objects momentarily, and ocular nystagmus is pronounced. Others move their eyes from place to place with apparent inability to concentrate on any particular object. The higher grades will develop interest and much better visual acuity, but with increasing age the risks are multiplied by developing lens opacities and high myopia.

The visual acuity of mongoloids is seldom good, but their requirements are so limited in their homes and institutions that, even with grossly reduced vision due to dense lens opacities, very few require special attention for these defects. For those who are to receive special schooling a full ophthalmological examination should be conducted early, to determine the presence of lens opacities or to correct any refractive error (particularly myopia).

**COLOUR VISION**

Nine patients were found to have sufficient intelligence and lenses clear enough to be tested for ordinary colour vision. No scientific methods were used, but a series of 20 pairs of coloured wools were presented for naming and matching. Pastels and common confusion shades were included. In no case was any significant defect suspected. Sometimes the wrong wools would be chosen in an attempt at quickness, but on their refusal the right colour would be found. For those who could name the colour the correct word was always given.

The colour vision of mongoloids is clinically normal in those who have sufficient mental development for colour to be of significance.
**THE EYES IN MONGOLISM**

**Refractive Condition**

With mydriasis by homatropine and cocaine, retinoscopy was performed on 35 patients ranging in age from 5 to 60 years. An electric retinoscope was used, together with spherical and cylindrical lenses in a trial frame. The power and axis of any astigmatic correction could then be recorded as accurately as possible. The results are summarised in Table No. 1. The astigmatism could not always be determined very accurately owing to irregularity from lens opacities, trachomatous pannus, or head and eye movements. In the table these cases are indicated by the "U".

The 35 cases may be divided into 2 groups:

1. Those with physiological variations from emmetropia.
2. Those with high myopia.

Twenty-two patients (approximately two-thirds) showed refractive errors which may be considered as physiological variations of emmetropia. As in any similar series of normal people, there is a tendency for the refractive errors to vary little from emmetropia. Astigmatism was not very high, and its axes were usually symmetrical in the two eyes. Sometimes arcuate opacities caused the astigmatism to be less regular. In this group the vitreous and fundi were normal.

Thirteen patients (approximately one-third) were high myopes. By focusing the equators and the posterior poles of the fundi the myopic refraction was seen to be mainly axial. Vitreous floaters, choroidal-retinal thinning and degeneration, and bilateral convergent strabismus were all common findings in this group, but retinal detachment was not seen. The myopia is no different from that in the general population, but its incidence is very much greater. The simple, non-progressive, uncomplicated myopia must always be differentiated from the sinister type with its fundus degenerations and visual catastrophes. It is the latter severe myopia which is found in such a large number of mongoloids.

The aetiology of high myopia is still uncertain. Its frequency among some races (Jews, Japanese), its repeated appearance in some families, and its association with some hereditary diseases (non-progressive night blindness) all indicate its genetic background. But its inheritance is not simple, and has been confused by the failure to separate the different types. Possibly its occurrence is conditioned by multiple factors. The effect of environment in foetal life is not known. The very high incidence of severe myopia in mongolism raises many interesting speculations, but difficulties immediately arise because of our profound ignorance concerning myopia in general.
### Table I—Showing the refractive errors determined objectively in 35 patients with Mongolism

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<th>Left Eye</th>
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</tr>
<tr>
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</tr>
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<td>11</td>
<td>CAT</td>
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</tr>
<tr>
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<td>12</td>
<td>+4</td>
<td>irreg</td>
</tr>
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</tr>
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### High Myopes

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<tr>
<td>9</td>
<td>-2'0</td>
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<td>High myope in view of age. R. myopic crescent. Frequent rolling twitches</td>
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<td>—</td>
<td>—</td>
<td>Typical myopic fundi without atrophy</td>
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<td>-20'0</td>
<td>?</td>
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<td>Myopic crescents, thinned choroid. Ocular nystagmus L. converg. squint</td>
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<td>135</td>
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<td>45</td>
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<td>-6</td>
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<td>—</td>
</tr>
<tr>
<td>34</td>
<td>-20</td>
<td>—</td>
<td>-20</td>
<td>—</td>
<td>—</td>
<td>—</td>
<td>Bilateral convergent squint</td>
</tr>
<tr>
<td>35</td>
<td>-8</td>
<td>—</td>
<td>-10</td>
<td>—</td>
<td>—</td>
<td>—</td>
<td>Typical myopic fundi without extensive atrophy. Trachomatous pannus R. &amp; L.</td>
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<td>-5</td>
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<td>Bilateral convergent squint with abduction weakness R. &amp; L.</td>
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<td>-15</td>
<td>—</td>
<td>-10</td>
<td>—</td>
<td>—</td>
<td>—</td>
<td>L.C.C.S. about 20° Wearing undercorrecting glasses</td>
</tr>
<tr>
<td>52</td>
<td>-15</td>
<td>—</td>
<td>-8</td>
<td>—</td>
<td>—</td>
<td>—</td>
<td>Bilateral convergent squint. Myopic crescents</td>
</tr>
<tr>
<td>57</td>
<td>-10</td>
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<td>-8</td>
<td>—</td>
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</tr>
<tr>
<td>61</td>
<td>-20</td>
<td>—</td>
<td>-15</td>
<td>—</td>
<td>—</td>
<td>—</td>
<td>Bilateral convergent squint with severe abduction weakness. Fundus changes</td>
</tr>
<tr>
<td>67</td>
<td>-12</td>
<td>—</td>
<td>-11</td>
<td>—</td>
<td>—</td>
<td>—</td>
<td>Bilateral convergent squint. Gyrate atrophy R. &amp; L.</td>
</tr>
</tbody>
</table>
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TABLE II

Showing the visual acuity and refractive errors of 5 mongoloids capable of reading Snellen’s test charts

<table>
<thead>
<tr>
<th>Case No.</th>
<th>Age</th>
<th>I.Q.</th>
<th>V.R.</th>
<th>V.L.</th>
<th>Unaided</th>
<th>Refraction</th>
<th>With Glasses</th>
</tr>
</thead>
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<tr>
<td>37</td>
<td>30</td>
<td>73</td>
<td>6/24</td>
<td>6/18</td>
<td>J.1</td>
<td>-1'0</td>
<td>+2'0</td>
</tr>
<tr>
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<td>35</td>
<td>34</td>
<td>6/12</td>
<td>6/12</td>
<td>J.1</td>
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<td>0'25</td>
</tr>
<tr>
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<td>37</td>
<td>52</td>
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<td>6/36</td>
<td>J.2</td>
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<td>6/60</td>
<td>6/60</td>
<td>J.1</td>
<td>-4'5</td>
<td>-1'0</td>
</tr>
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</table>

The evidence indicates that, in the main, the refractive conditions of the eyes in mongolism are determined by the same factors which influence them in normal people. The genetic background is probably important, but in some subjects the relationship between myopia and other mongoloid changes is obscure.

Mongolism is a disease in which there is great retardation of growth of many parts of the body. In some organs (e.g., limbs) genetic factors determining size appear to be unable to produce their characteristic effects in the presence of mongolism. In the eyes, however, myopia is associated with excessive growth. The other organ that is characteristically large in mongolism is the tongue.

NYSTAGMUS

Nystagmus occurring in mongolism was recorded in 1899 by Sutherland and subsequently by numerous writers—Muir (1903), 5 in 26 cases; Ormond (1912), 5 in 42 cases; Brushfield (1924), “seldom.” They made no attempt to determine its cause or to describe its different types, but merely noted its presence.

When a normal person attempts to move his eyes beyond the limits of the field of normal fixation, the ocular musculature is attempting to cause the eyes to depart beyond their normal excursions. The eyes tend to drift back from these abnormal positions into which they have been forced by the cortical demands. As the
### Table III—Showing the types of cataract found in 52 mongoloids from aged 5 to 60 years

| Case No. | 9 | 10 | 11 | 13 | 14 | 15 | 16 | 17 | 18 | 19 | 21 | 22 | 23 | 24 | 25 | 26 | 27 | 28 | 29 | 30 | 31 | 32 | 33 | 34 | 35 | 36 | 37 |
|----------|---|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|
| Age of Patient | 5 | 9 | 9 | 9 | 10 | 10 | 11 | 11 | 11 | 11 | 11 | 13 | 13 | 13 | 13 | 14 | 17 | 20 | 22 | 25 | 25 | 25 | 26 | 26 | 26 | 27 | 27 | 27 | 30 |
| Type of Examination | L | L | L | L | L | L | L | L | L | L | L | L | L | L | L | S | L | S | L | S | S | S | S | S | S | L | S | S |
| Arcuate opacities | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 2 | 0 | 0 | 0 | 0 | 0 | 1 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 2 | 0 | 0 | 2 | 0 |
| Y sutural opacities | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 1 | 0 | 1 | 1 | 1 | 2 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| Other sutural opacities | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| Scattered flakes & dots | 0 | 0 | 0 | 1 | 1 | 0 | 2 | 0 | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 2 | 1 | 1 | 1 | 0 | 0 | 1 | 1 | 3 | 2 | 1 | 2 |
| Peripheral splashes | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 1 | 0 | 2 | 1 | 2 | 1 | 0 | 0 | 1 | 1 | 1 | 2 | 2 | 1 | 1 |
| Central radiating spokes | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 2 | 0 | 0 | 0 | 0 | 0 |

| Case No. | 38 | 39 | 40 | 41 | 42 | 43 | 44 | 45 | 47 | 48 | 49 | 50 | 51 | 52 | 53 | 54 | 55 | 56 | 57 | 59 | 60 | 61 | 64 | 65 | 66 | 67 |
|----------|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|----|
| Age of Patient | 30 | 32 | 32 | 32 | 34 | 35 | 35 | 37 | 37 | 38 | 39 | 40 | 40 | 41 | 42 | 43 | 44 | 44 | 46 | 47 | 51 | 60 |
| Type of Examination | S | S | S | S | S | S | S | S | S | S | S | L | S | S | S | S | S | S | S | S | S | S | S | S | S | S |
| Arcuate opacities | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 3 | 2 | 0 | 0 | 2 | 0 | 0 | 0 | 0 | 0 |
| Y sutural opacities | 1 | 0 | 0 | 1 | 3 | 1 | 0 | 0 | 0 | 1 | 1 | 0 | 2 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 3 | 0 | 0 | 0 | 0 | 0 |
| Other sutural opacities | 0 | 0 | 0 | 0 | 1 | 1 | 0 | 0 | 0 | 1 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 1 | 0 | 0 | 0 | 0 | 0 |
| Scattered flakes & dots | 1 | 1 | 1 | 2 | 3 | 3 | 2 | 1 | 3 | 2 | 4 | 1 | 2 | 1 | 2 | 1 | 1 | 1 | 1 | 2 | 3 | 1 | 3 | 2 | 2 | 1 |
| Peripheral splashes | 3 | 0 | 1 | 1 | 1 | 1 | 1 | 1 | 1 | 2 | 1 | 2 | 2 | 4 | 3 | 2 | 2 | 0 | 0 | 3 | 3 | 1 | 2 | 2 | 1 |
| Central radiating spokes | 0 | 0 | 0 | 0 | 0 | 2 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 3 | 0 | 1 | 1 | 0 | 0 |

**Examination:**
- L = loupe.
- S = loupe & slit-lamp.
- 1 = few flakes or thin lines.
- 2 = moderate number.
- 3 = numerous.
- 4 = extremely numerous or thick.
drift occurs, it is quickly corrected by pyramidal mechanisms. The drift and check cause a form of nystagmus. This type can be elicited in 50 per cent. of normal persons on extreme forcible deviation of the eyes to one side or the other (Chavasse). It is more easily produced if there are any physical handicaps, or when the person is fatigued. If strabismus is present, nystagmus is often seen when we lead the eyes in the direction against the squint (in the direction of action of the weakened muscle). Many mongoloids do not use their lateral recti as much as normal people, the extreme examples being seen in the common bilateral convergent strabismus associated with uncorrected high myopia.

**FIG. 20.**

Brain of mongoloid No. 72, aged 27 years. Showing simplification of gyri, small pons, brain-stem and cerebellum.
Owing to the frequency of strabismus, obliquity of the palpebral apertures, and lack of keen interest in their surroundings, this so-called pseudo-nystagmus can be found very commonly in mongolism. The excursions are somewhat more pronounced than in normal people, so that this nystagmus may receive unwarranted attention unless one is aware of its nature.

A feature of mongoloid brains that was noted as early as 1890 by Wilmarth, and subsequently by many observers, is the smallness of the brain-stem and cerebellum (Fig. 20). Sometimes these structures may be only one half the normal size, and on section show considerable cellular irregularity and demyelination.

In view of these changes, nystagmus should be an almost constant feature, but this is not so. In the present series of 67 patients, if one ignores the very brief nystagmus produced on lateral version, which is so common in normal people, then most of them have steady eyes in all directions. After eliminating those with strabismus as a cause of nystagmus when the direction of gaze is led against the weakened muscle, only 9 cases remain. Of these, seven are explicable on the basis of peripheral causes (retinal or ocular nystagmus). They may be subdivided according to the probable cause of their ocular nystagmus as follows:

1. Lens opacities—5 cases.
2. Maculae at fault, high myopia—2 cases.

In mongolism the commonest causes of nystagmus (and of strabismus) are lens opacities and high myopia. The cataracts prevent clear images falling on the maculae. Uncorrected high myopia results in unfocused images. These may fall on maculae further disturbed by choroido-retinal thinning.

These defects interfere with the fixation reflex, so that control of eye posture via afferents from the maculae to the occipital cortices is eliminated, and a pendular type of nystagmus occurs. The searching movements may be constant, or increased by attempted fixation. If fixation can be brought about (e.g., small bright light) the nystagmus diminishes.

The remaining two patients are very low-grade aments, and probably have considerable aplasia or degeneration of the central nervous system. Both have nystagmus constantly in all positions (appendix No. 1). Ophthalmoscopically they have clear ocular media, and the maculae appear to have differentiated anatomically. They were the only subjects with nystagmus apparently due to central causes.

Therefore pronounced nystagmus in mongolism is not commonly due to causes within the central nervous system, but is usually due to ocular defects of which the most frequent are lens opacities and high myopia.
The Eyes in Mongolism

Strabismus

The frequent occurrence of strabismus has been noted by almost all writers on mongolism. They agree that the squint is always convergent, but very wide differences of opinion exist concerning its frequency. In 1899 Sutherland reported: "Nystagmus and squinting are often present during the first six months, but pass off as the infant grows." Ormond (1912) found 9 cases among 24 patients examined (21.4 per cent.). Brushfield (1924) said: "Every mongol examined has exhibited this in more or less degree without exception." Most publications on mongolism are written by physicians who apparently formed their opinions purely from the appearance of the eyes in relation to the nose and palpebral apertures.

The bridge of the nose is often wide and flattened in mongolism, and many babies or infants have epicanthal folds. When such a patient looks slightly to one side the cornea turned towards the nose partly disappears, so that the eye seems to be turned inwards much more than its fellow is turned outwards. A squint may be thought to be present when it is not. In any ophthalmic department children are frequently seen who have been brought along under this misapprehension. Similarly, marked obliquity of the palpebral apertures may cause deception.

The presence of strabismus can only be diagnosed when the visual axes are shown to be out of parallel alignment, and the necessary tests are not always easy to perform in very young or retarded children.

In the present investigations the cover test was used whenever possible. This was difficult in many cases because the patient would fixate any object only momentarily, and when the cover was moved the eyes would not be held steady. The more intelligent mongoloids co-operated sufficiently, but in many, reliance had to be placed upon the position of each corneal reflex.

Normal Binocular Vision. The physiology of binocular vision and its perversions leading to strabismus have undergone great advancement since most articles on mongolism were written. The development of binocular function has been admirably described by Chavasse (1939). In considering strabismus in mongolism a short review is desirable.

The attainment of fully developed human binocular vision is a complex process which is not normally achieved until the child is about 6 years old. The underlying anatomical requirements are not completed until several years after birth. Functional fulfilment depends on the development of a number of reflexes, some of which are innate, whereas others have to be conditioned by use. Chavasse described how, during evolution, the two eyes became locked by afferents from receptors outside the eyes. The primitive labyrinths were particularly concerned in the control of this early "compensatory reflex." The eyes moved conjugately, but did not have to follow the head slavishly in all its movements. To this course control was later added a fine adjustment from afferents arising in the retinae and so visual in nature—"oriental fixation." Later the eyes were able to make disjunctive movements for convergence, increasing the benefits of binocular fixation in relation to objects moving closer to the head.
The human primitive "compensatory reflex" is of such phylogenetic age that it has become unconditionally fixed and is present at birth. The relatively more recent oriental fixation and vergence reflexes are only rudimentary at birth, and if they are not used they are quickly lost. Normally these reflexes are not fully established to resemble unconditioned fixity until the child is aged about 5 years. In young babies apparently aimless eye movements are frequent, but even these "aimless" movements occur only over a restricted range. Horizontal divergence is limited to the position of rest; convergence to about 35 degrees, and vertical deviation is practically never seen. With anatomical completion of the visual pathways and increasing visual awareness, the rewards for convergence are quickly appreciated. The regarded object appears larger and stereoscopically isolated.

The convergence reflex is reinforced, and develops rapidly. These reflexes, however, do not lead to increased definition. In addition, a further series of reflexes develops associated with the slower forming accommodation process.

The accommodation reflexes are three in number (Chavasse):—

1. The primitive accommodation–convergence reflex, via the occipital cortex. A blurred or double image elicits convergence and accommodation in fixed relationship.

2. The more recent harmonic accommodation–convergence reflex via the occipital cortex. There is a physiological range of elasticity between the amount of convergence and accommodation to allow appropriate adjustments to be made.

3. The "voluntary" control from the frontal cortex via the pyramidal system. There is no control over individual muscles, so that accommodation and convergence cannot be separately reinforced. Convergence develops much more rapidly than accommodation in the early months of perception, but by the time the child is five to six years old the two have become fully co-ordinated.

Binocular Function in Mongolism. In mongolism reflex development is greatly retarded, and the visual mechanisms show considerable differences from normal. As has been mentioned previously, the exclusion of suitable retinal impulses sometimes prevents the development of the fixation reflex, and searching or ocular nystagmus ensues.

Among any group of mongoloids a conspicuous feature is the number who retain the infantile habit of converging spontaneously and irregularly. During ophthalmoscopic examination they often attempt to focus the light, and in so doing both eyes undergo extreme convergence. The same phenomenon occurs frequently during refraction attempts to look at the spectacle-frame. When they follow an object approaching the nose, convergence may be excessive and then adjusted. Sometimes one eye converges more than the other, and is then brought back into alignment.

Types of Strabismus in Mongolism. Among 67 patients examined, 22 cases of constant strabismus were found (appendix No. 2). This proportion is at least 20 times the incidence of squint in the general population. If to these are added the inconstant squints and the number who converge spontaneously or irregularly, then some form of noticeable convergent muscular anomaly was found in more than one half of the mongoloids. The deviation was always convergent and horizontal. Vertical deviation or ocular torticollis were not seen. The presence of divergent strabismus is not recorded in the literature.
The commonest causes of convergent strabismus in ordinary ophthalmic practice are muscle pareses, hypermetropia and anisometropia. Developmental myopia sometimes causes divergent strabismus, and rarely congenital myopia produces convergent squint. Less common causes of dissociation are macular disease, optic atrophy, cataract and corneal opacity.

In mongolism, judging by the changes which have been reported in the brain stem, the commonest factor responsible for the production of strabismus should be muscular paresis. Limitation of abduction is commonly seen, and appears to support this conclusion. That muscle pareses are common is refuted when the vertical muscles are considered. Not one case was found with ocular torticollis or vertical deviation of any significant degree. Among ordinary cases of strabismus vertical anomalies are relatively common.

If a sufficiently large number of mongoloids were examined, some cases with height difference would be found. The risks of muscle paresis in mongolism must be at least equal to those among the general population.

The most surprising feature was the number of cases with bilateral convergent squint associated with abduction weakness of each eye. There were 9 of these cases, and the appearances were at first very suggestive of bilateral paresis of the lateral recti. On further examination 8 were found to have high myopia. (The other patient had dense developmental cataracts, and may have been myopic before their formation.) Chavasse attributed this form of strabismus to congenital myopia. The far point of distinct focusing for uncorrected myopes is close to the eyes, so that the range of clear vision is very small. The benefits of binocular vision are readily appreciated when objects are viewed within this range. As distant objects are too blurred to excite interest, the eyes tend to remain converged, and the person so affected devotes himself to things close at hand. When any muscle remains restricted in activity, secondary changes occur within it and the surrounding fascia. After some months or years these structural alterations lead to permanent reduction of the muscle’s excursions. Contracture is typical of long-standing squint, and the differences between paretic and other types do not then exist (Chavasse). The frequency of congenital myopia increasing with development has already been shown to occur in approximately one-third of the patients with mongolism. As most of these do not receive spectacles, uncorrected high myopia has been the most common cause of constant strabismus. The other important cause of squint was found to be some form of cataract. If sufficiently clear images are not received by the maculae, binocular vision is not possible.
The necessary reflexes may be precluded by congenital cataracts, or binocular vision may be broken by opacities developing later in life.

Normal infants usually have an esophoria which gradually weakens and changes to an exophoria during adult life, the change being accentuated by the onset of presbyopia. If dissociation occurs, the type of squint is usually determined by the underlying muscle-balance of the time. In mongolism the excessive power of the medial recti continues, so that, if dissociation occurs, a convergent squint follows irrespective of age. If only one eye is affected, so that it receives distorted and embarrassing images, it will be deviated to permit the suppression of these unwelcome stimuli.

Five patients were seen in whom squint was due to lens opacities. Three of these had congenital cataracts (two arcuate, one lamellar), and two had cataracts developing in adult life.

Isolated paresis of a medial rectus is very rare, so that a divergent strabismus would be extremely uncommon. The medial recti are so strong in mongolism that the usual causes of divergent strabismus fail to operate.

Unlike many of the features of mongolism, the strabismus can be readily explained. The inconstant and variable aberrations of convergence are due to insufficient development of the harmonic accommodation-convergence reflex and to the attempts to obtain clear images by the demands from the frontal cortex.

The most important causes of constant strabismus are uncorrected high myopia and lens opacities. They produce the high proportion of constant squinters in this disease. With the other more expected causes, strabismus is so frequent as to be considered one of the stigmata of mongolism.

EXTERNAL INFLAMMATIONS

Nearly all the older writers emphasise the frequency and chronicity of extra-ocular inflammations, particularly blepharitis and conjunctivitis. Young mongoloids show thin lid margins, healthy surrounding skin, clear conjunctival sacs with white globes and clear corneae. Ocular infections are then not common. Later in life the skin tends to become thicker and rougher, and in the patients so affected conjunctival and lid margin infections increase in frequency.

When the eyes are rubbed frequently, particularly with dirty hands, maceration of the skin, blepharitis and ectropion may be produced. Mongoloids are very affectionate, and in institutions they tend to associate with each other, separating from other
patients who dislike their embraces and kisses. Infection can easily pass from one to another.

In days when efficient antiseptics were not available and institutions were less clean, severe chronic infections of the lids and conjunctivae were common. Trachoma was frequently seen with the usual complications of epiphora, chronic conjunctival discharge, entropion, trichiasis, ptosis, conjunctival shrinkage and corneal scarring. Owing to improved hygiene, sulphonamides and penicillin, infections can now be readily controlled and severe sequelae prevented.

In the present investigations trachoma was noted seven times among males and twice among the females. The youngest patient affected with it was aged 27 years. All eyes were quiescent, but some had extensive corneal pannus, conjunctival shrinkage or trichiasis. Most had chronic epiphora, and this appeared to cause a few to have blepharitis and swollen lid margins. Among the remaining 59 patients only one had active conjunctivitis, and another had blepharitis, although a few had some loose skin flakes between the lashes.

**INTRA-OCULAR INFLAMMATIONS**

Mongoloids are very susceptible to infections of all mucous surfaces, particularly of the respiratory and gastro-intestinal tracts. Pneumonia, pulmonary tuberculosis and dysentery with their complications, have all contributed heavily to a short expectation of life. With modern methods of prevention and treatment these illnesses are less powerful but still common. What tremendous possibilities for "septic foci" must exist! Yet intra-ocular inflammation is very uncommon, and in the present series evidence of it was only seen once or twice. Case No. 34, aged 27 years, showed bilateral attenuated posterior synechiae, which did not prevent the pupils from dilating fully. They probably resulted from a single very mild attack of iritis many years previously.

Case No. 61, aged 44 years, showed several irregular patches of choroido-retinal atrophy with heavily pigmented borders, perhaps due to an old mild choroido-retinitis. She was highly myopic with large complete crescents and choroido-retinal thinnings, so that the patches may have been primarily degenerative and not inflammatory. The Wassermann reaction of the blood and parallel tests were performed as a routine on admission. No positive reports occurred in the whole series.

In contrast with extra-ocular infections, intra-ocular inflammations were seen no more commonly in mongolism than might occur in the general population.
Discussion

In reviewing the peculiarities of the eyes in mongolism, fundamental defects should be differentiated from those produced secondarily. Where an understanding of normal structure and function is available, this distinction may not be difficult, but where this intelligence is lacking, separation may be impossible.

An abnormal vascular system has been shown to be the cause of arcuate lens opacities. These in turn may lead to disturbances of visual function causing ocular nystagmus or convergent strabismus. High myopia has been shown to be a common cause of bilateral convergent strabismus simulating bilateral sixth (abducens) nerve paralysis. These conclusions are possible from a knowledge of the embryology of the lens, and normal and perverted physiology of binocular vision.

It is easy to imagine other organs in which abnormal vascular supply during early development could lead to secondary structural changes, and then to superimposed functional alterations affecting distant parts of the body. The peculiarities of the palpebral apertures may be due to early developmental defects in the skin, but more extensive investigations are required in this field.

The processes involved in normal and abnormal growth are still too obscure to provide explanations for many of the structural alterations in mongolism, and small variations in the early stages of development may lead to wide differences later in life. In that way the clinical syndrome may be built up from a number of patients, but each may show considerable individual differences.

It is interesting to look for the earliest evidence of disturbance associated with mongolism. In the present series one male showed an incomplete coloboma of one eye. The foetal fissure begins to close at its central end in man at about 11 mm. stage (4 to 5 weeks) and all trace has disappeared by the 15 mm. stage (5 to 6 weeks) (Mann, 1937). The condition in this patient represents a disturbance at about 5 weeks, and it is interesting that he showed the lowest grade of amentia of all, in contrast with the high social position and intelligence of his parents. One case, however, is not enough to eliminate other causes than mongolism. Such cases are valuable because our knowledge of choroidal colobomata is much more accurate than that of disc-shaped cataract, where another case suggested a disturbance between 5 and 7 weeks.

The eyes were surprisingly little affected during the vulnerable organogenetic period up to the third month of foetal life. Cases showing anophthalmos, microphthalmos, orbital cysts or choroidal colobomata were unexpectedly rare. The earliest consistent abnormalities seen in the eyes were the arcuate lens opacities, which
THE EYES IN MONGOLISM

correspond with a foetal length of 35 mm. (8·5 weeks). Their dependence upon abnormal vessels shows that mongolism is established by this period at least. The disturbance which produces the abnormalities in the vascular system could probably determine anomalies in other developing structures. The cause of this disturbance is probably the cause of mongolism itself. Of this we are still ignorant, although many theories have been propounded.

The presence of high myopia in approximately one third of the mongoloids appears to be a most significant finding. Unfortunately our knowledge of myopia is still meagre. If severe myopia could be shown to have a high incidence among the relatives of mongoloids, this would produce good support for a germinal influence in mongolism. If such findings were lacking, the influence of the pre-natal environment would be shown to be important for some cases of myopia. The records would be valuable from either aspect. The observations are simple, as the myopic fundus picture is so easily recognised, and the focus on a direct ophthalmoscope would be adequate if retinoscopy were not available.

Tredgold describes the tissues in mongolism as having a peculiar lack of durability and vitality. Mongoloids are much slower to grow up, never reach proper maturity, and begin to decline earlier. The infants appear to be only about half their chronological age, whilst the older adults look much more than their years. The lack of tissue durability and vitality is seen very well in the vascular iris stroma. To the hypoplasia found in young mongoloids, atrophy is added later and the changes become very marked as age advances. But senile cataracts do not occur unduly early in the lens—a tissue very susceptible to sclerosis with age. In all patients up to 60 years of age, no senile cataracts were seen. Similarly, except in myopia, the retinal vessels and maculae presented no degenerative appearances.

The problem of the cause of mongolism remains unsolved. Developmental anomalies of the eyes, viscera, dermal patterns and cardio-vascular system show that the disturbance begins very early during foetal life. The embryo may be defective from maternal causes alone or from a genetic predisposition that only finds expression in a particular environment. Penrose (1931-1932) has shown statistically the importance of maternal age, and many other workers have further incriminated the foetal environment. The geneticists, while emphasising the major contribution from the environment, point to the studies on twins and the frequency of features usually influenced genetically (e.g., dermal patterns). In mongolism the importance of the environment is so definite, and dysplastic changes so widespread and little understood, that to attempt too much explanation on a genetic basis is
undesirable. It is a disease full of riddles, complicated by incomplete and inaccurate observations. Many aspects are inexplicable until more is known about normal physiology, embryology, and the occurrence of some of its features among the general population.

Summary

The eyes of 67 mongoloids varying in age between one and sixty years, were examined. The series contains 40 adults over 25 years of age.

Mongolism is a syndrome that is recognised by a number of stigmata, each of which is not separately specific for the disease. Those found associated with the eyes were short sloping palpebral apertures, evenly curved upper lid margins, convergent strabismus, nystagmus, characteristic lens opacities, speckling and peripheral atrophy of the iris stroma, and high myopia.

Four adult mongoloid skulls presented considerable dysplastic changes, particularly in the regions where growth is normally pronounced. The slope of the palpebral apertures is shown to be independent of the slope of the orbital axes.

Epicanthus is of the same type found in normal babies. It usually disappears with growth of the face.

The iris often shows thinning of the stroma peripherally, with a speckled appearance due to collections of pigment in a circle concentric with the pupil. Generalised vascular hypoplasia is suggested as the main cause. The changes are not seen in heavily pigmented brown irides.

Lens opacities are very common. Three types are described, the combination of which is characteristic for mongolism. Arcuate lens opacities develop during foetal life owing to the presence of abnormal vessels. Sutural opacities occur mostly within the anterior Y sutures of the foetal nucleus. Flake opacities are found within the juvenile and adult nuclei. The opacities form slowly, progress little and rarely require surgery.

The refractive errors of 35 mongoloids were measured. Two-thirds of them had variations from emmetropia corresponding to the general population. One third had high myopia with the usual fundus changes.

Nine patients were found to have obvious nystagmus. Of these ocular nystagmus due to lens opacities or myopia was present in 7 cases. Aplasia or degeneration of the central nervous system was probably responsible for the remaining two.

Twenty-two mongoloids (one third) had constant strabismus. It was always convergent and horizontal. Eight had bilateral squint due to high myopia. Lens opacities were the cause of 5 cases.
THE EYES IN MONGOLISM

Convergence and accommodation are frequently not co-ordinated, so that excessive convergence is common. Muscular palsies are not important causes of strabismus.

The visual acuity of mongoloids is frequently poor because of myopia, nystagmus, strabismus, lens opacities, and failure to reinforce conditioned reflexes during development.

Where possible, explanations are given for the peculiarities found. Other conditions await further knowledge of normal physiology before the abnormalities in mongolism can be fully described.

ACKNOWLEDGMENTS

The investigations were conducted from the Institute of Ophthalmology, University of London, and under the terms of a Gordon Craig Travelling Scholarship of the Royal Australasian College of Surgeons.

The research was suggested and made possible by Miss Ida Mann and Dr. W. M. McGrath. Dr. J. H. Watkin, physician-superintendent of Leavesden Hospital, provided every possible facility for the examination of the adults, and permitted access to case records and museum material. The children were examined at The Fountain Hospital, by courtesy of Dr. L. T. Hilliard, physician-superintendent.

The co-operation, enthusiasm and kindness of the above and their staffs made the work a great pleasure. The friends who have helped me in the preparation of this work are too numerous to mention individually. I must ask them to accept this collective expression of my gratitude.

APPENDIX NO. 1

CONTAINING THE CASE REPORTS OF 9 MONGOLIDS WITH NYSTAGMUS

OCULAR NYSTAGMUS

(a) LENS OPACITIES.

Case 16, aged 11 years.—R. disc-shaped cataract (no vision). Left arcuate opacity causing mixed astigmatism. Macula appears normal. Eyes parallel. Some nystagmus on fixation, coarse nystagmus to left, fine to right.

Case 51, aged 39 years.—Pendulum nystagmus on fixation, typical bilateral posterior polar cataracts from hyaloid arteries preventing central vision.

Case 53, aged 40 years.—Bilateral arcuate lens opacities and constant ocular nystagmus. Right convergent strabismus 45°. Will not fixate with right eye when left is uncovered. Low hypermetrope when retina focused with direct ophthalmoscope. When the fixated object is brought to 3 inches from the nose the excessively converged right eye swings out a little, both visual axes appear directed on the object, and the ocular nystagmus diminishes.

Case 56, aged 42 years.—Dense congenital lamellar cataracts right and left. Unsuccessful operation on right during childhood. Continual searching movements of head and eyes. If he fixates a bright light, the eyes remain steady as the nystagmus diminishes.

Case 60, aged 44 years.—Very dense bilateral developmental cataracts of central radiating type (cortical and adult nucleus), coarse rolling nystagmus increased on looking to right or left. Right convergent strabismus of variable angle. Can just see enough to find his way about in familiar places.

(b) MYOPIA.

Case 9, aged 5 years.—Very frequent rolling twitches more noticeable on looking to right or left. Eyes parallel. Media clear. Myope with fundus thinning but no obvious atrophy.
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Case 25, aged 17 years.—Uncorrected high myopia with anisometropia and chorioido-retinal thinning. Left coronary and sutural lens opacities (see further notes under strabismus). Does not maintain fixation anywhere more than a few moments. Many variable jerking movements on attempting fixation (ocular nystagmus). Nystagmus very coarse, and increased on looking to left (against left internal strabismus) reduced and finer to right.

Nystagmus due to Defective Nervous System

Case 29, aged 25 years.—Extremely low-grade amert, resistive and impulsive, even fails to recognise his parents. Comes from a high-grade family who did all that was possible, but he has remained in the lowest category. Media clear. Right and left fundus showed no abnormality, and left macula gave good reflex. No gross refractive error. Will not hold fixation of a light. Constant searching type of nystagmus in all directions. Right convergent strabismus of variable angle.

Case 47, aged 37 years.—Fundi seen well, so flake lens opacities insufficient to reduce vision, eyes parallel. Low-grade amert. Persistent, variable, coarse nystagmus greatly increased with all movements. On close fixation the eyes become steady.

Appendix No. 2

Containing the Case Reports of 22 Mongoloids with Convergent Strabismus

Accommodative, Inconstant, Variable

Case 11, aged 9 years.—Left eye converges irregularly, slight overaction of inferior obliques on each side on adduction.

Case 23, aged 13 years.—Parallel for distance, often converges evenly, but on repeated convergence sometimes develops internal strabismus and becomes monocular, can alternate. Low compound hypermetropic astigmatism.

Accommodative—constant.

Case 23, aged 13 years.—Alternating convergent squint. Hypermetropia +7-0 D. Can hold fixation with either eye but prefers fixation with left. Abduction weakness more to right than to left; probably due to secondary contracture.

Case 50, aged 39 years.—Alternating convergent squint about 20°. At times converges spontaneously. After instillation of homatropine and cocaine, eyes became parallel. Low compound hypermetropic astigmatism. Slight overaction of right inferior oblique on adduction.

Due to Uncorrected High Myopia.

Case 25, aged 17 years.—Uncorrected high myopia with anisometropia. Right myopia approximately —10 D. Both retinæ show choroïdo-retinal thinning. Left coronary and central lens opacities, right lens almost clear. Ocular nystagmus, but only attempts fixation with the right eye. Left convergent strabismus with very variable angle due to influence of frontal cortex in attempting to obtain clear vision by voluntary control over the accommodation-convergence synkinesis.

Case 34, aged 27 years.—Alternating bilateral convergent squint with limited abduction to right and left. Binocular convergence when object held closely. By retinoscopy myopia —20 D right and left, and wearing —13 D spheres right and left. Fine stretched posterior synechiae. Small arcuate lens opacities at 9 o’clock in each eye.

Case 36, aged 27 years.—Anisometropia (R.-12D. L.-5D.) and prefers fixation for distance with left eye as it has lower myopia. Bilateral convergent squint with marked abduction weakness right and left. Does not appear to become binocular at any distance. When object is close can hold fixation with either eye, but the
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non-fixing eye always remains excessively adducted. Bilateral arcuate lens opacities temporally.

Case 40, aged 32 years.—L.C.C.S. about 20°. High myope, has worn glasses many years although considerably undercorrected. Does not hold fixation long with either eye; will hold fixation a short time with the right eye but quickly gives it up. For quite close, eyes assume equal convergence and appears to fix binocularly.

Case 44, aged 35 years.—Bilateral convergent squint. Now dense central cataracts right and left. When lenses were clear very high myopia was noted with myopic degeneration of the fundi.

Case 52, aged 40 years.—Bilateral convergent squint. High myopia but no retinal degeneration. Can abduct fairly well but each eye remains converged about 30° most of the time. Can alternate until the object is very close when she appears to become binocular.

Case 61, aged 44 years.—Bilateral convergent squint. Can abduct either very little beyond the midline. High myope with some choroido-retinal degeneration, appears to become binocular when object is held very close.

Case 67, aged 60 years.—Bilateral convergent squint with abduction weakness right more than left. High myope with typical gyrate atrophy. The right eye is more convergent than the left, the atrophic patches are more extensive, and its vision is probably very poor.

DUE TO LENS OPACITIES.

Case 17, aged 11 years.—R.C.C.S. with right abduction weakness. Right arcuate lens opacity causing considerable astigmatism and leading to ambylopa ex anopia. Right fundus focuses at zero with direct ophthalmoscope.

Case 27, aged 22 years.—L.C.C.S. 30° with left abduction weakness. Only fixes light momentarily, and eyes move conjunctively in rapid change of positions. Developmental lens opacities obscure fundi to ophthalmoscopic examination.

Case 53, aged 40 years.—R.C.C.S. 45° arcuate lens opacities right and left giving very poor vision, although fundi showed no abnormality. Constant searching nystagmus. On convergence fixes with left eye and both adduct until the object is about 3 inches from the eyes. The right eye then swings out until both optic axes are directed on to the object, nystagmus diminishes, and binocular vision seems to occur.

Case 56, aged 42 years.—Variable convergent squint, visual acuity very poor owing to dense bilateral lamellar cataracts. These were needled during childhood, but the result is poor. The head and eyes undergo continual searching movements.

Case 60, aged 44 years.—Right convergent squint about 30° but variable. Has dense central developmental lens opacities. Fundi not seen.

DUE TO DEVELOPMENTAL ANOMALY OF RETINA.

Case 29, aged 25 years.—Right convergent squint of variable angle. Very low-grade ament. Partial choroidal coloboma spreading from right disc downwards and outwards, and appearing to disturb right macula. Constant searching type of nystagmus, although left eye normal in appearance.

DUE TO CORNEAL OPACITIES FROM TRACHOMA.

Case 64, aged 46 years.—R.C.C.S. 50°. Considerable trachomatous pannus and ptosis. Right pupil smaller than left. Too stupid to co-operate for further examination.

Case 55, aged 41 years.—L.C.C.S. about 45°. Extensive trachoma of left cornea preventing fundus examination. Right fundus can be seen and focused at −8 D. Fixes steadily with right but cannot fixate with left.

DOUBTFUL CASES.

Case 2, aged 1 2/12 years.—Convergent squint, probable alternator, conjugate movements appear good. Unable to investigate further.
Case 43, aged 45 years.—L.C.C.S. 45° ambylopic left eye. Compound hypermetropic astigmatism. Retinoscope shows left astigmatism more irregular than right so probably due to anisometropia.

Case 64, aged 46 years.—Bilateral convergent squint with abduction weakness right and left. Now has dense trachomatous pannus and moderately thick lens opacities. Fundi not seen, impossible to estimate refraction. May have been myopic before opacities occurred, but no records available.

Case 65, aged 47 years.—L.C.C.S. about 15° with slight weakness of left abduction. Performed cover test very well. Low compound-hypermetropic astigmatism. Some lens opacities, but fundi were seen clearly. Possibly paretic left external rectus.

Case 72, aged 27 years.—Not seen during life but case history records bilateral convergent squint. Brain shown in Fig. 20. Sections through sixth nerve nuclei at different levels showed no abnormality (Prof. R. Willis). No records of refraction, probably high myope.

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