Recent advances in paediatric ophthalmology

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Contributed by request and dedicated to Sir Stewart Duke-Elder

In the preface to Volume VI of the "System of Ophthalmology", which is concerned with Ocular Motility and Strabismus, Sir Stewart Duke-Elder makes a comment which is so characteristic of his style: "as years pass, more and more super-specialties appear within our specialty and more and more of our colleagues climb above the golden clouds to revel in their favourite hobby, seeking to do little else". It is obvious that the emergence of paediatric ophthalmology as a separate discipline creates a further super-specialty, but there is much good in this because, as he also states: "the concentration of interest allows significant advances to be made". Indeed, it is not difficult to seek to justify the inclusion of paediatric ophthalmology as a super-specialty because of the rapid accumulation of knowledge of many ophthalmic disorders which are unique to children, or which present in childhood in a peculiar way, and also because of the increasing knowledge that many general paediatric diseases have distinctive ocular features which constitute essential elements in the assessment of each condition as a whole.

There are many topics which are worthy of mention in a discussion of the advances in paediatric ophthalmology in recent years: the role of the nucleoprotein deoxyribonucleic acid (DNA) in the chromosomes as the "blue print" for the perpetuation of each living cell with its individual characteristics; the error in the replicating process which results in the absence or disorder of an indispensable enzyme within the cell so that there is the occurrence of one of the many recognized inborn errors of metabolism; the aspiration method in the surgical treatment of congenital cataract with a maintenance of the anterior chamber throughout the operation; the distinctive features of the three main forms of amblyopia—strabismic, stimulus deprivation, and anisometropic; the peculiarities of anomalous retinal correspondence which is so commonly a feature of esotropia in childhood; and many others. It is necessary, however, to restrict this discussion because of the space available, and the vexed and complex problem of suspected blindness in early childhood is chosen as a suitable topic.

Suspected blindness

The problem of suspected blindness in infancy and early childhood, that is, at an age when the child is unable to co-operate adequately in any form of subjective test, has beset ophthalmic surgeons for countless years, but more recently the application of various electrodiagnostic procedures has helped greatly to reduce the problem. It should be noted that in this context the term "blindness" is used in a special sense and denotes a state of impaired vision which determines the need eventually for the child to be dependent on a form of education which does not involve the use of sight, and it is different, therefore,

from true blindness which implies a loss of all visual function. It is certainly a matter of great importance to achieve a precise assessment of the underlying disorder at the earliest possible stage because in certain cases therapeutic measures may be of value provided they are carried out before the condition is too far advanced. An early assessment is also of almost equal importance in certain other cases in which no form of treatment is available because of the necessity for the parents to become conditioned as rapidly as possible to the vital part which they have to play in the early years of their blind child. Inevitably, it is difficult for them to overcome the emotional stress which quite naturally engulfs their whole beings in such a distressing situation, but the future happiness of the blind child, and indeed of the whole family, is dependent on a speedy period of adjustment. This is fostered in no small way by the early realization of the parents that the verdict of blindness is based on sound judgement which has been reached only after a careful and detailed examination of the child; frequently it is of value to obtain a confirmation of the conclusion from a colleague, but every effort should be made to avoid the trailing of the afflicted child from clinic to clinic, and sometimes even from country to country, in the forlorn hope of finding some miraculous cure.

It is necessary in all cases to carry out the usual routine methods of clinical assessment: a history of the pregnancy, labour, and neonatal period; a history of the parents' observations which led to the impression of blindness; an account of the child's progress with regard to any illnesses and the timing of the classical landmarks which characterize various stages of infancy and early childhood and which provide information about the general physical and mental development; and, a thorough examination of the eyes. It is unfortunate, however, that the main item of interest—the extent of the visual function at the time of the examination and its likely extent in years to come—is susceptible only to indirect methods of determination, but considerable help is obtained from certain specialized tests which in some cases are essential before forming any precise conclusion.

Optokinetic nystagmus

Optokinetic nystagmus is a visuo-motor reflex which can be elicited in normal circumstances at an even earlier age than the light fixation and following reflexes. It occurs in response to a target which excites visual interest by producing moving contours in the visual panorama, such as a rotating drum which has alternate broad black-and-white stripes on its surface. This is not a recently recognized phenomenon; it was first described in the earlier part of the 19th century by Purkinje, who observed the peculiar jerky movements of the eyes in a crowd of people in Vienna who were watching a procession of cavalry. He recognized its physiological significance because the only other logical explanation was the occurrence of some neurological disaster which had struck suddenly and simultaneously a large number of people in Vienna! The application of this phenomenon to infants, however, is more recent and it represents an important diagnostic test because optokinetic nystagmus can be induced at an early stage of life provided there is a reasonable form of visual function and provided also that the infant is fairly attentive at the time of the examination. It is, therefore, a useful screening device because a positive optokinetic response is an indication that the child is not blind, and its consistent absence provides confirmatory evidence of a marked impairment of visual function. Furthermore, it may be applied in a more sophisticated way by the use of the electro-oculogram (which records the standing electrical potential of the eye) to measure the changes in the positions of the eyes during the test and by the use of graduated stimuli so that a quantitative measurement of the visual acuity may be determined at an early age.

Electroretinopathy

In the young child this investigation requires a general anaesthetic so that the contact lens bearing the active electrode may be inserted accurately over the cornea, and the infant may be kept at rest during the period of recording. The electroretinogram (ERG) measures the action potential produced in the retina in response to light stimulation. The amplitude of the response is low at birth even in normal infants, but slowly rises to adult levels during the early months of life. In pathological conditions, the ERG response is diminished only if a large proportion of the elements in the outer layers of the retina is abnormal (about 50 per cent.). Severe dysfunction of the macular region, of the ganglion cell layer of the retina, of the optic nerve fibres, and of the optic radiations and the visual cortex is associated therefore with a normal ERG response, and the investigation has only a limited but important role in the determination of blindness. It is evident, however, that in the normal eye under photopic conditions, the ERG response is due mainly to cone activity, and this is reflected in a high flicker fusion frequency of the response, whereas under scotopic conditions, the electrical response is due mainly to rod activity and the flicker fusion frequency is low. It is possible, therefore, by varying the intensity, colour, and frequency of the light flash stimulation under both scotopic and photopic conditions of background illumination, to differentiate between those pathological conditions which cause widespread dysfunction of the cones alone, of the rods alone, or of all the receptor elements.

The ERG is of critical value in certain conditions of the retina which, despite a marked impairment of visual function, show no obvious abnormality on ophthalmoscopic examination, such as the cone dysfunction syndrome or neuroepithelial dysgenesis in which there appears to be a functional block in the transmission between the intact retinal receptors and the bipolar cells, and also in other conditions of the retina such as the tapeto-retinal degenerations, particularly the congenital form (Leber's amaurosis), in which obvious abnormal changes in the retina (attenuation of the arteries, pallor of the optic disc, and irregular pigmentary changes) are frequently delayed for some years. Indeed, little reliance can be placed on the pigmentary changes in the fundus from a diagnostic point of view in an assessment of a tapeto-retinal degeneration in early life because, unlike the classical retinitis pigmentosa, these are seldom marked so that subtle pigmentary changes are akin to those which may occur in the normal fundus, except rarely when they become aggregated in the macular area with a misleading appearance suggestive of a macular dystrophy rather than a wide-spread retinal degeneration.

Electro-oculography

The electro-oculogram (EOG) measures the standing potential of the eye and is dependent on the integrity of the pigment epithelium; its application to the precise recording of ocular movements has been discussed earlier in relation to optokinetic nystagmus. The EOG is abnormal in a tapeto-retinal degeneration so that it is complementary to the ERG, but it remains normal in a neuroepithelial dysgenesis in contrast to the ERG which is abnormal.

Cortical evoked potentials

The visually evoked response (VER) of the cerebral cortex to a stimulus may be recorded by electrodes provided an averager (essentially a computer) is used to eliminate the distracting background of the persistent electrical activity of the cortex which is unrelated to the stimulus from the specific signal. It is obvious that the nature of the stimulus is of critical importance, because it accounts for the production of the data-processing within the cortex

which in turn accounts for the electrical potentials presented to the recording apparatus by the computer. It is evident, however, that a knowledge of the data-processing is also of great importance in an interpretation of the electrical recordings, particularly in view of the peculiarity of the afferent visual pathway in that the relative sizes of the components of the visual apparatus in the retina are reversed in the visual cortex; for example, the small foveal area in the retina and yet the large area of foveal representation in the visual cortex. Furthermore, there is evidence of several cortical representations of the visual field and in each cortical representation there are cells which respond to differing features of the retinal image (Hubel and Wiesel, 1968); it seems likely that these are served by distinctive retinal ganglion cells which do not show uniform latent periods or conduction times in their nervous transmission. It follows that different forms of stimuli are liable to provoke different forms of responses, and this is endorsed by the finding that the visual cortex consists of a number of parallel channels, each of which is concerned with a different form of stimulus (Campbell and Kulikowski, 1966).

It follows that the VER is not a single entity but a variable phenomenon depending on the nature of the exciting stimulus, and this adds considerably to the difficulty of an interpretation of the changes which occur in the visual cortex in response to a visual stimulus, particularly as in disease the numerous VERs may be affected in different ways. When a single flash from a simple stroboscope is used as the stimulus there is the production of a complex type of VER, but when a simple harmonic signal consisting of a light of rhythmically fluctuating intensity is used, there is an improved form of VER (Arden, 1973), although the response appears to arise from a wider region than the striate area of the visual cortex (Regan, 1972). From a clinical point of view the use of luminance-evoked potentials provides information about the function of the afferent visual pathways, but in order to localize the response to a single cortical area it is necessary also to use a stimulus which produces a vigorous response from the cortical cells and this is achieved when the stimulus involves the features of contrast and movement, such as alternate black-and-white squares which can be illuminated in a rhythmical way so that as a black square becomes white the adjacent white square becomes black, the so-called pattern reversal stimuli (Arden, 1973).

It is obvious that the VER is of great importance in an assessment of suspected blindness when this is the result of a lesion of the afferent visual pathways, particularly when the lesion is in a supragenulate situation so that there is an absence of optic atrophy on ophthalmoscopic examination and also an absence of any anomaly of the pupillary reflexes. By this means it is possible to determine precisely the occurrence of cortical blindness, and also to avoid the mistake of regarding a child as suffering from an irreversible form of cortical blindness when the apparent absence of any visual response is essentially the result of a delayed development of the visual processes (as part of a general mental handicap), so that eventually there may be evidence of a reasonable form of visual function. It is suggested that cases of this kind should be regarded as showing a "retardation of visual perception" rather than a "cortical blindness" (Karseras and Wybar, in preparation).

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