An Ophthalmic Survey at the Worcester School for the Blind

D. SEVEL, Head, Department of Ophthalmology, and S. SHOCHET; formerly Senior Registrar, Department of Ophthalmology, Groote Schuur Hospital, Observatory, Cape Town.

SUMMARY

An ophthalmological survey undertaken at the Worcester School for the Blind showed that 75.8% of the students required admission because of hereditary or congenital abnormalities. The importance of genetic counselling of the older children in this closely knit social environment is essential.


An ophthalmological survey was undertaken at the Worcester School for the Blind. The purpose of the investigation was to ascertain the causes of blindness that warranted admission of the students and then to re-evaluate these aetiological factors in the light of our present knowledge.

During February 1970, Dr S. Shochet examined 253 pupils at the Worcester School for the Blind. This examination included a detailed general history and full ophthalmological examination and photography where indicated. This survey is based on the analysis of these findings.

The students at the school are divided into the following groups:

(i) those who are educated through the medium of Braille;
(ii) those who are partially sighted and are educated through the use of special visual aids;
(iii) a mentally retarded group;
(iv) a deaf, blind group; and
(v) a professional and vocational training group.

The 253 pupils examined in this survey belonged to the first 4 groups. They came from all parts of the Republic of South Africa, South West Africa and from Rhodesia and the neighbouring Portuguese territories.

The division at the school into the various groups is not determined solely by the visual acuity of the pupils. They are reclassified from one group to the other depending on intelligence and variations of the visual acuity.

Subdivision of the groups was as follows:

Braille students ........................................... 142
Visual-aided students .................................. 102
Deaf-blind group ........................................ 9

Only 41 (16.2%) of the 253 students were totally blind, i.e. they were unable to perceive light with either eye. There were 150 males (59.29%) and 103 females (40.71%). The 3:2 ratio of males to females is similar to the ratio found in a survey of the blind done in England and Wales in a group aged 1-20 years.

EYE CONDITIONS FOUND

The over-all analysis of the eye conditions is shown in Table I.

<table>
<thead>
<tr>
<th>Condition</th>
<th>No. of cases</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Anophthalmos</td>
<td>5</td>
<td>1.98</td>
</tr>
<tr>
<td>Microphthalmos</td>
<td>6</td>
<td>2.37</td>
</tr>
<tr>
<td>Corneal dystrophy</td>
<td>5</td>
<td>1.98</td>
</tr>
<tr>
<td>Keratoconus</td>
<td>10</td>
<td>3.56</td>
</tr>
<tr>
<td>Aniridia</td>
<td>1</td>
<td>0.39</td>
</tr>
<tr>
<td>Buphthalmos</td>
<td>13</td>
<td>5.14</td>
</tr>
<tr>
<td>Congenital cataracts</td>
<td>53</td>
<td>20.95</td>
</tr>
<tr>
<td>Persistent hyperplastic primary vitreous</td>
<td>1</td>
<td>0.39</td>
</tr>
<tr>
<td>Retinoblastoma</td>
<td>6</td>
<td>2.37</td>
</tr>
<tr>
<td>Retinal dystrophy</td>
<td>30</td>
<td>11.86</td>
</tr>
<tr>
<td>(tapetoretinal degeneration)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Macular dystrophy (degeneration)</td>
<td>38</td>
<td>15.02</td>
</tr>
<tr>
<td>Albinism</td>
<td>16</td>
<td>6.32</td>
</tr>
<tr>
<td>Myopic degeneration</td>
<td>18</td>
<td>7.11</td>
</tr>
<tr>
<td>Retinal detachment</td>
<td>9</td>
<td>3.56</td>
</tr>
<tr>
<td>Retrolental fibroplasia</td>
<td>7</td>
<td>2.77</td>
</tr>
<tr>
<td>Choroidoretinitis</td>
<td>7</td>
<td>2.77</td>
</tr>
<tr>
<td>Optic atrophy</td>
<td>25</td>
<td>9.88</td>
</tr>
<tr>
<td>Trauma</td>
<td>6</td>
<td>2.37</td>
</tr>
<tr>
<td>Ophthalmia neonatorum</td>
<td>4</td>
<td>1.58</td>
</tr>
<tr>
<td>Phthisis bulbi (causes unknown)</td>
<td>9</td>
<td>3.56</td>
</tr>
<tr>
<td>Enucleations (causes unknown)</td>
<td>2</td>
<td>0.79</td>
</tr>
</tbody>
</table>

Anophthalmos

It is rare that the optic vesicle does not develop (at 4 weeks) at all after the formation of the forebrain. While an eye may not be observed or felt clinically, histological examination of the orbit frequently reveals the presence...
of an eye vestige. Clinical examination of a patient with anophthalmos usually shows the eyelids to be well formed, i.e. the lashes, tarsal glands and puncta are present. The orbit is shallow but lined with conjunctiva. A functional lacrimal system is usually found, as are extraocular muscles with normal innervation.

In this series we found 5 students with anophthalmos, 2 were bilateral and 3 were unilateral.

Microphthalmos

Microphthalmos is defined as a small eye and follows on involution of the primary optic vesicle. It is often associated with failure of the embryonic fissure to close. The clinical condition may vary from a rudimentary vestige of the globe, to a small but otherwise normal eye.

In this series none of the microphthalmic eyes were normal. There were 6 students (2.37%) with microphthalmos and all were bilateral.

Corneal Dystrophy

These are corneal opacities which are bilateral and develop in apparently otherwise normal eyes. The condition often occurs in more than one member of a family. There is no evidence of associated corneal inflammation or corneal vascularization. These opacities may occur at birth, but more frequently develop later in life.

In this survey 5 students (1.98%) were considered possible cases of corneal dystrophy; in 2 of these there was a familial history of similar involvement.

Keratoconus

The cornea becomes conical as a result of an axial thinning of its stroma. This condition is usually bilateral and severe visual impairment results from the development of a high degree of irregular myopic astigmatism. Keratoconus usually presents clinically at about the age of puberty. All the children who were found to have keratoconus were admitted to the school before the age of puberty.

We found 10 children (3.97%) at the school with keratoconus and half of these had associated retinitis pigmentosa. The association of retinitis pigmentosa with keratoconus is well described in the literature.

Aniridia

Although the name implies a complete absence of the iris, this is not strictly correct, as there is usually some remnant of the iris root. Familial cases are transmitted by autosomal dominant inheritance with a high degree of penetrance. The association of non-familial aniridia with Wilms's tumour has been described, and it has been suggested that all cases of non-familial aniridia should be investigated for the presence of Wilms's tumour. The only case of aniridia in this survey did not have a Wilms's tumour.

Buphthalmos (Congenital Glaucoma)

Buphthalmos results from obstruction to the outflow of aqueous through a maldeveloped drainage angle. The obstruction occurs at birth or within the first few years of life. As a result of the plasticity of the young globe, the raised intraocular pressure causes the eye to enlarge. It then resembles an ox eye. The eye is photophobic, lacrimation occurs, the corneal diameter is enlarged, the cornea becomes oedematous and the optic disc is pale and cupped.

There were 13 students (5.14%) with buphthalmos in this survey. Dislocation of the lens was found in 4 cases but it was not possible to establish whether this was the cause or the effect of the buphthalmos.

Early recognition of the condition followed by goniotomy under direct vision appears to be the most satisfactory treatment for controlling the intraocular tension.

Congenital Cataracts

Any opacity of the lens is defined as a cataract. For the purpose of the survey only those cataracts which were present at birth or were noticed soon thereafter and which were the primary cause of poor vision were included under the heading.

There were 53 students (20.9%) with congenital cataracts—by far the greatest single cause of blindness at this school. The aetiology of the congenital cataracts was obscure in 35 cases, was familial in 12 children, and rubella was the cause in the remaining 6.

Retinoblastoma

Retinoblastoma is a malignant tumour of the eye arising from immature retinal cells. Different foci of tumour growth may arise independently in one or both eyes and the condition usually presents clinically by the age of 3 years. It is bilateral in about 25% of patients. The hallmark of diagnosis is a leucocoria (cat's eye reflex) or a squint. The mode of inheritance is by an autosomal dominant gene with variable penetrance. There were 6 children (2.37%) with retinoblastoma in this survey.

An interesting observation with regard to the intelligence of children with retinoblastoma was made by Williams and Frazer and Friedman who found that in schools for the blind, retinoblastoma-blinded children had a significantly higher IQ than a group of children blinded from other causes. In this survey the average intelligence quotient was 121.42 in 5 of the children (not yet estimated in the remaining child).
TAVAN-SP 54 IN ARTERIOSCLEROSIS AND HYPERLIPEMIA


Clinical Situation
Long-term medication in 46 cases of arteriosclerosis and hyperlipemia.

Treatment
These patients were divided into an ORAL group (22 patients on 2 to 3 tablets of 25 mg 3 times daily) and PARENTERAL group (18 patients on 100 mg i.m. daily) and a small CONTROL group (9 patients without pathological blood lipid changes).

Findings
In the ORAL group the esterified fatty acids were reduced by an average of 29% (maximum drop in a single case 56%). The average blood cholesterol reduction was 18%. In the PARENTERAL group, the corresponding drop was 27% and 19%. In the CONTROL group, long-term therapy did not produce significant changes.

3 cases of cardiac infarct where anticoagublion treatment had to be stopped because of haemorrhage, were put on to TAVAN-SP 54. Improvement took place and there was no recurrence.

TAVAN-SP 54 IN DEGENERATIVE AND THROMBOTIC ANGIOPATHIES


Clinical Situation
60 cases of degenerative and thrombotic angiopathies—23 suffering from coronary sclerosis (14 acute coronary thrombosis); 11 peripheral perfusion disturbances in the lower extremities; 18 leg-vein thrombosis: 8 thrombosis at other sites.

Treatment
1 to 2 injections 100 mg i.m. daily for 8 to 15 days, thereafter 100 mg i.m. every 2nd day for 3 weeks. 4 cases received 8 to 12 tablets of 25 mg each per day.

Findings
TAVAN-SP 54 represents a significant advance in the treatment of cardiovascular diseases specifically with reference to thrombosis and degenerative arteriopathies. TAVAN-SP 54 exercises a penetrating, regulatory effect in cases of hyper- and dyslipemias. In all cases it was excellently tolerated.

TAVAN-SP 54 IN CORONARY INSUFFICIENCY AND ARTERIOSCLEROSIS WITH OR WITHOUT HYPERTENSION


Clinical Situation
37 patients with coronary insufficiency and generalised arteriosclerosis with or without hypertension.

Treatment
Outpatient therapy. At first 3 x 3 tablets for 20 days then 100 mg i.m. per day for 22 to 30 days.

Findings
A noticeable effect on clinical symptoms especially after parenteral therapy, which ensures dependable results. The tablets have a relatively weaker action but oral administration is considered valuable for technical reasons.

In our opinion, TAVAN-SP 54 deserves an important position in the treatment of arteriosclerosis. It is excellently tolerated.

TAVAN-SP 54 AND ACUTE CORONARIES (THROMBOSIS, INFARCTION AND PRE-INFARCTION)


Clinical Situation
10 patients, some of them refractory to vaso-active medication (nitro-glycerine) and heparin.

Treatment
Initially, high doses of TAVAN-SP 54 were given per infusion.

Findings
In 8 cases the anginocose crisis abated within 3 to 5 days. ECG clearly confirmed success of therapy in 3 cases.

In addition to its fibrinolytic and lipolytic action, TAVAN-SP 54 shows a moderate anticoagulative action (inhibits pro-accelerin and thrombin) ... without the disadvantages of classical anticoagulation therapy.

TAVAN-SP 54 promises excellent results especially in the pre-infarction stage even in serious and therapy-resistant cases. It provides a non-toxic and effective maintenance therapy to combat "ischemic coronaryitis" and recurrence of infarction.
Tapetoretinal Degeneration

There were 30 children (11.86%) who were diagnosed as suffering from tapetoretinal degeneration. Their fundal pictures showed marked attenuation of the vessels, pale optic discs and pigmentary disturbances. Of these children, 14 had the typical bone corpuscle pigmentation of retinitis pigmentosa; 3 suffered from bilateral deafness and 2 had associated renal disease. The association of deafness with retinitis pigmentosa is well described. There was one case of Laurence-Moon-Bardet-Biedl syndrome. The boy is mentally retarded, grossly obese and has hypogenitalism. He had supernumerary toes which have been amputated.

Macular Degeneration

Macular lesions showed a great variety of morphological types as evidenced by varying degrees of pigment disturbance.

In our survey 38 children (15.02%) were diagnosed as having macular degeneration. All of them had bilateral abnormalities. While central vision was lost in these patients, peripheral vision was retained.

In 50% of these cases there was familial history of similar involvement. Sixty-five per cent of this group are being taught through the medium of Braille.

Retinal Detachment

There were 9 children (3.56%) with retinal detachment in this series. In 4 students the retinal detachments were associated with pathological myopia and in 3 with aphakia; this indicates that the retinal detachment was not the primary cause but followed on concomitant ocular pathology.

Retrolental Fibroplasia

This condition was diagnosed in 7 children (2.77%). The disease occurs in premature infants who have been exposed to high oxygen concentrations soon after birth. The effect on the retinal vessels is dependent on the abnormally high blood oxygen levels and the subsequent relative hypoxia. Retinal neo-vascularization and vitreous haemorrhage result. The retina becomes detached and may be seen as a white mass lying behind the lens. This usually becomes evident at the age of about 3 months. The toxic effect of oxygen on premature children was first discovered in 1951 and it was interesting to note that only 1 child in this group was born after 1954.

Optic Atrophy

In this condition the optic disc is pale and the conducting function of the optic nerve is reduced to a varying degree.

In this survey 25 children (9.88%) were diagnosed as having optic atrophy. The aetiology of the disease could be ascertained in only 10 of these patients: 5 probably were due to familial optic atrophy, 2 were a result of hydrocephalus, 1 was due to a cerebral tumour, 1 followed on tuberculous meningitis and 1 was due to Crouzon’s disease (craniofacial dysostosis).

Albinism

Albinism is a condition in which pigment deficiency results from an inborn error of protein metabolism. There is a lack of the enzyme tyrosinase which converts 3,4-dihydroxyphenylalanine (dopa) into melanin. Because of the absence of pigment there is light scatter within the eye resulting in poor vision, and in addition the maculae are pathological. These patients usually have a coarse nystagmus.

In our study 16 children (6.32%) were suffering from albinism. Of these, 2 patients had complete albinism, i.e. a total lack of pigment in the hair, skin and eyes, and 14 had incomplete albinism, i.e. only the eyes showed a lack of pigment. Only 12.5% of these children were being taught through the medium of Braille, and as a group their visual acuity was the least affected.

Myopic Degeneration

As a result of the lengthening of the anteroposterior axis of the eye degenerative changes occur in the posterior segment of the globe. The condition is usually associated with high degrees of myopia, i.e. usually more than −6.0 dioptres. Progressive myopia is usually an autosomal dominant inherited condition.

In this survey 18 children (7.11%) were diagnosed as having myopia with retinal degeneration.

Ophthalmia Neonatorum

This is a bilateral conjunctivitis, usually mucopurulent and frequently suppurative. The condition manifests at birth or within the first few weeks of life. The common organisms responsible for the disease are Neisseria gonorrhoea and staphylococci. In the majority of cases the newborn child is infected during its delivery through the infected genital tract.

At birth there is absence of tears, the lymphoid tissue of the conjunctiva has not developed and the cornea is thin. There is, as a result, little resistance to this infection. Pent-up pus beneath the swollen lids causes a rapid dissolution of the cornea with disastrous blinding effects.

The condition is an ophthalmic emergency and intensive antibiotic therapy (penicillin drops, concentration 20 000 units/ml) is indicated. Systemic penicillin is advised. Frequent opening of the palpebral fissure is of paramount importance to allow drainage of the mucopurulent exudate. Prophylactic treatment still remains an important aspect of ophthalmia neonatorum. Chloromycetin oint-
ment is recommended for local conjunctival application to all newborn children. Since the advent of antibiotics, ophthalmia neonatorum has largely been eliminated as a cause of blindness.

There were 4 children (1.58%) at the school who were diagnosed as having had ophthalmia neonatorum. The youngest child in this group was born in 1953.

Deaf-Blind Group

In 1959, the Worcester School for the Blind established a special section for deaf-blind children. Highly specialized teaching is applied in an effort to develop speech. There were 9 pupils (3.56%) in this group: 4 were deaf-blind due to rubella, 3 had tapetoretinal degeneration and deafness and 2 had optic atrophy and deafness.

DISCUSSION

This survey showed that 192 students (75.8%) who required admission to the Worcester School for the Blind suffered from hereditary or congenital abnormalities. Many of these hereditary conditions are sporadic. Genetic counselling of the older children in such a closely knit social environment is obviously essential. The groups at special risk are the patients suffering from heredomacular degeneration, corneal dystrophy, tapetoretinal degeneration (retinitis pigmentosa) and the children with progressive myopia.

Patients with retinoblastoma in particular require genetic counselling because of the high penetrance of these genes. All cases of retinoblastoma presenting at the Department of Ophthalmology at Groote Schuur Hospital are assessed at the Ocular Tumour Clinic (combined ophthalmology and radiotherapy clinic). A well organized regimen of treatment accordingly is instituted. It is mandatory that the siblings be assessed at regular intervals. The importance of genetic counselling is always emphasized to the parents of the patient and also to the patient himself when he is old enough to comprehend its importance.

Of particular importance in this survey is the re-evaluation of the causes of blindness in the light of our present knowledge. A better understanding of the disease processes of the eye coupled with surgical techniques such as microsurgery have opened new and encouraging vistas.

The use of the aspiration technique for congenital cataracts may decrease the incidence of complications, such as secondary glaucoma and retinal detachment. It is now possible to view the filtration angle with greater facility and therefore goniotomy for buphthalmos has become a less hazardous procedure. With our present knowledge, retrolental fibroplasia should not be a frequent cause of blindness.

The general practitioner should be aware that rubella in the first 3 months of foetal development is still a significant cause of blindness (due to congenital cataract, iridocyclitis and macular degeneration) not infrequently associated with mental retardation, deafness (in 4 patients in this series) and congenital heart disease.

The awareness of ophthalmia neonatorum and the routine use of antibiotic ointment after the birth of a child have significantly reduced the incidence of blindness caused by this pre-1940 scourge.

The critical, brittle and significant period of ocular development is within the first 3 months of foetal life. During this sensitive phase drugs should be regarded as potentially teratogenic and therefore, if at all possible, should be avoided.

We wish to thank Mr T. Pauw, Principal of the Worcester School for the Blind, and his staff, for their whole-hearted co-operation and assistance in this survey; and Sisters J. H. B. Steyn, A. P. Kotze and M. de Wet for helping with the examination of the children.

Finally we wish to express our gratitude to the children. This project was financed by the Gratitude Fund of the Ophthalmic Department of the University of Cape Town, the Willem Goosen Trust for Ophthalmology, and the Ophthalmic Research Fund.

REFERENCES