DEPARTMENT OF HIGHER EDUCATION

A SURVEY OF HEREDITARY BLINDNESS AT THE SCHOOL FOR THE BLIND AT WORCESTER

by

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P R E F A C E

Very little attention has been devoted to the problem of hereditary blindness in the Republic of South Africa.

Based on superficial observations the necessity of research of this nature was realized and subsequently a preliminary investigation at the School for the Blind at Worcester was initiated to establish the needs for a comprehensive research programme, and a possible solution to the problem of hereditary blindness.

This survey is the first of its kind in the Republic of South Africa and conclusive information has come to light, indicating the importance and necessity of further research and procedures to combat blindness and impaired vision.

Prior to the investigation the Bureau considered it necessary to nominate authorities representing certain institutions and professions, to assist the Bureau in an advisory capacity in the task outlined.

The advisory committee comprises of the following members:

1. Dr. P.M. Robbertse Chairman
   Director, National Bureau of Educational and Social Research.

2. Prof. M.H. Luntz
   Department of Ophthalmology, University of the Witwatersrand, Johannesburg.

3. Dr. H. Meyer
   Department of Ophthalmology, University of Pretoria, Pretoria.

4. Dr. E. Epstein, representing the Council for the Prevention of Blindness.

5. Prof. J.D.J. Hofmeyr
   Department of Genetics, University of Pretoria, Pretoria.

6. Dr. S.H.F. Schiller, representing the Department of Health.

7. Mr. V.H. Vaughan
   University of South Africa, Pretoria.

8. Mr. N.H. Gaum, representing the South African National Council for the Blind, Pretoria.

9. Mr. D. Louw, representing the Department of Social Welfare.

Bureau Staff:

Mr. J. Op't Hof,
Research officer responsible for the research and compilation of the report.

I would like to express my sincere gratitude to the members of the Advisory Committee for their valued co-operation and interest which made the investigation possible.

We are profoundly indebted to Dr. J.M.P. Geerthsen of the University of Pretoria for his active interest and assistance in the evaluation of the combinations of eye conditions and the interpretation thereof.

This report does not provide an answer to the problem of hereditary blindness but delineates the direction and importance of the necessary research to attain a further step in reaching this aim.

B.M. Robertse.

DIRECTOR
NATIONAL BUREAU OF EDUCATIONAL AND SOCIAL RESEARCH

January, 1968.

C O N T E N T S

TABLES

CHAPTER 1

INTRODUCTION

1.1 THE PURPOSE OF THE RESEARCH

In 1963 the National Bureau of Educational and Social Research was requested by the South African National Council for the Blind to investigate the hereditary causes of blindness. This request was motivated by the observation that blindness had occurred in certain families for several generations and the assumption that eye defects in the majority of pupils at the School for the Blind at Worcester can be attributed to hereditary factors. The necessity for research of this nature is emphasized by the fact that certain eye conditions progressively deteriorate and eventually result in blindness.

In an attempt to comply with the above-mentioned request the purpose of the investigation is to ascertain in which cases hereditary factors caused blindness and impaired vision.

1.2 THE GROUP INVESTIGATED

The investigation was limited to the pupils who had attended the School for the Blind at Worcester since its establishment in 1881 up to 1965. This step was taken due to the practical problems involved in representing such an investigation as a sample of all the blind people in the country. In addition the investigation would serve as a pilot survey to a comprehensive survey in the future. The group mentioned above consisted of 1386 individuals of whom the ages at the time of enrolment ranged from 2 to 25 years.

Inmates of the old-age home and workshop for the blind at Worcester were included in the group investigated, as some of them had been pupils of the above-mentioned school and interviews could conveniently be arranged. Of this group 31 individuals had not attended the School for the Blind but were nevertheless included, as information concerning these individuals was readily obtainable by means of interviews.

This implies that the 1386 pupils of the School for the Blind which were included in the investigation were supplemented by 31 non-pupils. The total thus was 1417 individuals.

These individuals came from all parts of the Republic of South Africa including South West Africa, while a few were foreigners.

1.3 THE METHOD OF THE INVESTIGATION

The most important sources of information for this investigation were the files kept on each pupil at the School for the Blind. This information was supplemented by interviewing the staff of the School; pupils who were present at the time of the investigation; old pupils in the old-age home and workshop for the Blind, and parents and/or relatives of approximately 200 pupils who are resident in the vicinity of Worcester, Montagu, Wolseley, Touwsrivier, Robertson, Villiersdorp, De Doorns, Ceres, Cape Town, Stellenbosch, Somerset Strand, Velddrif, Clanwilliam, Morreesburg and Vredendal.

A specially designed questionnaire was used to obtain the information concerning the cause of the individual's eye condition, and family data. It was possible with the aid of the 1417 questionnaires and with information concerning family histories obtained mainly from interviews with adults, to compile 100 pedigrees of relative significance. These pedigrees were used to determine as far as possible to what extent hereditary factors are responsible for blindness and impaired vision.

For purposes of control a register was compiled containing the name of each pupil, his admission number to the School for the Blind, his family number and other information including the connection, if any, between families in which blindness occurred.

CHAPTER 2

EYE CONDITIONS, DEGREE OF VISION AND OTHER MEDICAL INFORMATION

2.1 GENERAL

In this Chapter attention is drawn to the different types of eye conditions responsible for blindness and impaired vision of the group investigated.

A brief consideration of the sex of the individuals investigated, the age at onset of eye trouble and age at onset of blindness, will supply further information as to the nature of the data dealt with.

2.1.1 Sex of Individuals

The sex distribution of the group investigated is given in Table 2.1

TABLE 2.1

SEX OF INDIVIDUALS

Sex	Number	Percentage
Male	855	60.34
Female	561	39.66
TOTAL	1,417	100.00

An outstanding feature of Table 2.1 is the three to two ratio of males to females. It would be interesting to determine whether blindness and impaired vision are subject to a sex predilection or whether parents were reluctant to enrol their daughters at the School.

2.1.2 Onset of the Eye Trouble

In Table 2.2 the age at which the onset of the eye trouble occurred in the different age groups is given for each eye separately.

TABLE 2.2

AGE AT ONSET OF EYE TROUBLE

Age Group	Right Eye	%	Left Eye	%	Total	%
Under 1 year	661	46.65	661	46.65	1,322	46.65
1 - 6	384	27.10	384	27.10	768	27.10
7 - 12	90	6.35	90	6.35	180	6.35
13 - 18	30	2.26	30	2.26	60	2.26
19 - 30	17	1.06	17	1.06	34	1.06
31 - 42	0	0	0	0	0	0
43 - 54	0	0	0	0	0	0
55 - 66	0	0	0	0	0	0
67 and older	0	0	0	0	0	0
No information	235	15.58	235	15.58	470	15.58
TOTAL	1,417	100.00	1,417	100.00	2,834	100.00

One distinct feature as observed from Table 2.2, is that the onset of the eye trouble occurred mainly (46.65%) at birth or shortly after. The percentages for the different age groups hereafter decrease until no individuals are included in the age groups of 31 years and older. One reason for the latter phenomenon is probably that individuals who experience the onset of eye trouble later in life, do not attend the School for the Blind.

Another deduction made from Table 2.2 is that there is no difference in the age at the onset of eye trouble for either the right or left eyes. This does not exclude the possibility that differences occur at the onset of eye trouble in the right and left eyes within a category.

2.1.3 The Onset of Blindness

In Table 2.3 the age at Onset of blindness in the different age groups is given for each eye separately.

TABLE 2.3

AGE AT ONSET OF BLINDNESS

Age Group	Right Eye	%	Left Eye	%	Total	%
Under 1 year	465	32.82	465	32.82	930	32.82
1 - 6	425	29.99	425	29.99	850	29.99
7 - 12	179	12.63	179	12.63	358	12.63
13 - 18	77	5.43	77	5.43	154	5.43
19 - 30	27	1.91	27	1.91	54	1.91
31 - 42	13	0.92	13	0.92	26	0.92
43 - 54	8	0.56	8	0.56	16	0.56
55 - 66	3	0.21	3	0.21	6	0.21
67 and older	3	0.21	3	0.21	6	0.21
No information	217	15.31	217	15.31	434	15.31
TOTAL	1,417	100.00	1,417	100.00	2,834	100.00

A comparison of Table 2.2 and 2.3 reveals that the figure for the onset of blindness under one year (32.8%) is considerably smaller than the figure for the onset of eye trouble under one year (46.65%).

From the age groups it is observed that the onset of blindness occurred more often in the higher age groups than is the case for onset of the eye trouble. This could mean that many cases of blindness can be prevented if the patients are discovered at an early stage and successfully treated.

2.2 CLASSIFICATION OF EYE CONDITIONS ACCORDING TO SITE AND TYPE OF AFFECTION

In the following Sections a classification of eye conditions according to site and type of affection is given with regard to the eyeball in general, conjunctiva, cornea, lens, uveal tract, retina, optic nerve, vitreous and site and type indefinite. Each of these sections is also represented by a section number in brackets to which reference will be made later on when explaining combinations of eye conditions. Each condition in the different sections has a corresponding index figure to facilitate analysis of data. The index 0 in the different sections represents the number of individuals who were not affected in that section.

2.2.1 Eyeball in General (Section 1)

The number of individuals whose eyeballs in general were affected appears in Table 2.4.

TABLE 2.4

EYEBALL IN GENERAL

Index		Frequency	%
01	Glaucoma (excluding infantile)	4	0.28
02	Myopia with detachment of retina	4	0.28
03	Myopia, detachment of retina not specified	77	5.43
04	Panophthalmitis and acute endophthalmitis	0	0.00
05	Albinism	11	0.78
06	Anophthalmos, excluding surgical	5	0.35
07	Hydrophthalmos (infantile glaucoma)	34	2.40
08	Microphthalmos	42	2.96
09	Aniridia	3	0.21
10	Coloboma, any part, excluding surgical	8	0.56
11	Multiple congenital anomalies	0	0.00
12	Other congenital anomaly specified	492	34.72
13	Congenital anomaly not specified	197	13.90
14	Disorganized eyeball (atrophic globe, phthisis bulbi)	35	2.47
15	Other general affection of eyeball specified	137	9.67
16	General affection of eyeball, not specified	368	25.97
	TOTAL	1,417	100.00

From Table 2.4 it is observed that congenital anomalies, specified (34.72%) and not specified (13.90%) were most common and myopia (an abnormal oval shape of the eyeball), (5.43%), and microphthalmos (shrunken eyeball), (2.96%), appeared rather frequently. It is also apparent that only three cases (0.21%) were found with aniridia (absence of iris) which is often a dominant condition.

2.2.2 Conjunctiva (Section 2)

The number of individuals suffering from affections in the conjunctiva (membrane that lines the eyelids and covers the eyeball in front) appears in Table 2.5.

TABLE 2.5

CONJUNCTIVA

Index		Frequency	%
1	Purulent ophthalmia of newborn (ophthalmia neonatorum)	42	2.96
2	Purulent ophthalmia other than newborn	0	0.00
3	Other affection of conjunctiva, specified	6	0.42
4	Affection of conjunctiva, not specified	6	0.42
0	None	1,363	96.19
	TOTAL	1,417	100.00

It is apparent that 2.96 per cent of the group investigated suffered from ophthalmia neonatorum (inflammation of the conjunctiva). This was mainly attributable to infectious diseases which are, however, nowadays effectively dealt with.

2.2.3 Cornea (Section 3)

In the following table the number of individuals who suffered from various corneal affections are recorded.

TABLE 2.6

CORNEA

Index		Frequency	%
01	Interstitial keratitis	5	0.35
02	Phlyctenular keratitis, including keratoconjunctivitis	1	0.07
03	Ulcerative keratitis, including trachoma	1	0.07
04	Sclerosing keratitis	21	1.48
05	Keratitis, other type specified	3	0.21
06	Keratitis, type not specified	46	3.25
11	Corneal dystrophy, degeneration	0	0.00
07	Keratomalacia	11	0.78
08	Keratoconus	35	2.47
09	Other affection of cornea specified	10	0.70
10	Affection of cornea, not specified	5	0.35
0	None	1,279	90.26
	TOTAL	1,417	100.00

It is seen from Table 2.6 that keratitis (opacity of the cornea) types were most common and that Keratoconus (2.47%) occurred rather frequently. Most of the keratitis types were most probably caused by infectious diseases which are nowadays under control.

2.2.4 Lens (Section 4)

In Table 2.7 the occurrence of individuals with various lens defects are noted.

TABLE 2.7

LENS

Index		Frequency	%
1	Cataract	182	12.84
2	Dislocated lens	5	0.35
3	Other affection of lens specified	11	0.78
4	Affection of lens, not specified	1	0.07
0	None	1,218	85.96
	TOTAL	1,417	100.00

It is observed from Table 2.7 that cataracts (opacity of the lens) were most common (12.84%). Most of these cases probably are secondary manifestations of other primary conditions. The cases in which a dislocated lens occurred (0.35%) are probably due to the syndrome known as homocystinuria.

2.2.5 Uveal Tract (Section 5)

Table 2.8 represents the number of individuals affected by the different conditions in the uveal tract.

TABLE 2.8

UVEAL TRACT

Index		Frequency	%
1	Iritis, Iridocyclitis or uveitis	3	0.21
2	Kerato-iridocyclitis or sclero-iridocyclitis	4	0.28
3	Choroiditis	7	0.49
4	Chorioretinitis	40	2.82
5	Sympathetic ophthalmia	6	0.42
6	Other affection of iris, ciliary body or choroid specified (including neoplasm)	120	8.47
7	Affection of iris, ciliary body or choroid, not specified	0	0.00
0	None	1,237	87.30
	TOTAL	1,417	100.00

It is apparent from Table 2.8 that iris atrophy (degeneration of the iris, which was the other affection of the iris) occurred in 8.47 per cent of the cases. Chorioretinitis (2.82%) occurred rather frequently.

From Table 2.8 it is apparent that other affections of the iris, ciliary body or choroid specified, occurred in 8.4 per cent of the cases. This percentage represents mostly iris atrophy and individual cases of an affection of the ciliary body. Posterior choroidal degeneration is not included in this percentage but is included with that of tapetoretinal degeneration in the following section since it was found to occur with the latter in virtually all the cases. It is also seen that Chorioretinitis (2.82%) occurred rather frequently.

2.2.6 Retina (Section 6)

The number of individuals affected by various retinal conditions is given in Table 2.9.

TABLE 2.9

RETINA

Index		Frequency	%
1	Retinopathy, including retinitis and lesions of retinal vessels	13	0.92
2	Retrolental fibroplasia	6	0.42
3	Detachment of retina	30	2.11
4	Tapetoretinal degeneration and allied conditions including retinitis pigmentosa	150	10.59
5	Macular degeneration	190	13.41
6	Neoplasm of retina	12	0.58
7	Other affection of retina specified	59	4.16
8	Affection of retina, not specified	0	0.00
0	None	957	67.54
	TOTAL	1,417	100.00

From this table it is seen that tapetoretinal degeneration and allied conditions (10.59%) and macular degeneration (13.41%) were most common. In virtually all cases of tapetoretinal degeneration the choroid was also degenerate.

The occurrence of retrolental fibroplasia (0.42%) was most probably due to the incorrect use of oxygen applied to those individuals after they were born.

2.2.7 Optic Nerve, Optic Pathway, and Cortical Visual Centres (Section 7)

The incidence of individuals, whose optic nerve was affected is given in Table 2.10 according to the different conditions.

TABLE 2.10

OPTIC NERVE, OPTIC PATHWAY AND CORTICAL VISUAL CENTRES

Index		Frequency	%
1	Optic nerve atrophy, optic neuritis or papilledema	280	19.76
2	Other affection of optic nerve specified	209	14.75
3	Affection of optic nerve, not specified	6	0.42
4	Lesion of optic pathway or cortical visual centres	1	0.07
0	None	921	65.00
	TOTAL	1,417	100.00

Table 2.10 indicates that optic nerve atrophy (19.7%) occurred most frequently followed by a "pale optic disc", (which was the other affection of the optic nerve specified (14.75%). A pale optic disc is most probably also an atrophic optic nerve and should be considered an identical condition.

2.2.8 Vitreous (Section 8)

The frequency of individuals affected with the various vitreous (internal fluid of the eyeball) conditions is given in Table 2.11.

TABLE 2.11

VITREOUS

Index		Frequency	%
1	Vitreous haemorrhage	0	0.00
2	Other affections of vitreous specified	3	0.21
3	Affections of vitreous not specified	1	0.07
0	None	1,413	99.72
	TOTAL	1,417	100.00

It is evident from Table 2.11 that relatively few individuals suffered from an affection of the vitreous humor.

2.2.9 Site and Type Indefinite or not Reported (Section 9)

Table 2.12 represents the number of individuals in which the site and type of the affection is indefinite or not reported.

TABLE 2.12
SITE AND TYPE INDEFINITE OR NOT REPORTED

Index		Frequency	%
1	Amblyopia ex anopsia	3	0.21
2	Amblyopia uni- or bilateral, not explained	4	0.28
3	Congenital nystagmus (only to be used if classification elsewhere is impossible)	17	1.20
4	Evidence insufficient for diagnosis	0	0.00
5	No report on site and type of affection	369	25.97
0	None	1,024	72.34
	TOTAL	1,417	100.00

As indicated by Table 2.12 nystagmus (1.20%) occurred most frequently and in 25.97 per cent of the cases there was no report on the site and type of affection.

In the following table (Table 2.13) the total percentage of persons suffering from conditions of the different components of the eye can be seen.

TABLE 2.13

AFFECTED COMPONENTS OF THE EYEBALL

	Percentage
Conjunctiva	3.81
Cornea	9.74
Lens	14.04
Uveal Tract	23.29
Retina	32.46
Optic Nerve	35.00

These percentages serve only as a relative measure of comparison and do not add up to 100 per cent because some persons suffered from affections in more than one part of the eye.

It is seen from Table 2.13 above that defects of the retina, choroid and optic nerve are the main causes of blindness and impaired vision. It will be remembered that a defective choroid is often associated with a retinal defect.

2.3 SYNDROMES OBSERVED

During the visit to the School and Workshops for the Blind the following syndromes were observed:

(a)	Sturge Weber	1 case
(b)	Dysplasia oculo-dento digitalis	1 case
(c)	Acrocephalosyndactylia (Apert's)	1 case
(d)	Dysostosis cranio facialis (Crouzon's)	3 cases

2.4 ADDITIONAL CONDITIONS OBSERVED

In many instances the pupils at the School revealed additional conditions of medical interest, e.g.

Skin Acne
Mitral Stenosis
Poliomyelitis
Sunken Chest
Haemorrhage
Undescended Testicles (Many cases)
Purpura

Scoliosis
Nerve Disease
Epilepsy
Cleft lip and/or Palate
Hydrocephaly
Menorrhagia
Gingivitis
Clubfoot
Anaemia
T.B.
Asthma
Virus (No. 4)
Pharyngitis
Adipose Gynandrism
Roundworm Infection
Atitis
Sinusitis
Status Dysraphygus
Uremia
Deafness

The correlation between these conditions and the different eye conditions must still be established and further investigation is necessary.

Many of the ailments mentioned above could be the primary cause of the different eye conditions.

Due to incomplete records the I.Q.'s of many pupils were not available. This is a disadvantage because in many instances the low I.Q. in conjunction with a certain eye condition forms part of a syndrome.

CHAPTER 3

ETIOLOGY AND COMBINATIONS OF EYE CONDITIONS, DEGREE OF VISION AND OTHER MEDICAL INFORMATION

3.1 INTRODUCTION

In this chapter due consideration is given to the etiology of eye conditions discussed in the previous chapter. In addition, the occurrence of the different combinations of eye conditions as they appeared individually was calculated. The chapter concludes with a brief discussion on the degree of vision recorded for the individuals investigated and a note on the medical treatment they received.

3.2 ETIOLOGY OF EYE CONDITIONS

The importance of the etiology in the manifestation of certain eye conditions cannot be over-emphasized. In observing the data on the etiology of eye conditions further insight into the importance of hereditary causes of blindness and impaired vision is gained.

An etiological classification of eye conditions will therefore be considered according to infectious diseases, external causes, and other causes. The percentage occurrence for these categories is set out in Table 3.1.

TABLE 3.1

ETIOLOGICAL CLASSIFICATION OF EYE CONDITIONS

Categories	Frequency	%
Infectious diseases	140	9.88
External Causes	101	7.13
Other causes	814	57.45
Etiology undetermined or not specified	362	25.54
Total	1417	100.00

It is thus seen that in 25.54 per cent of the cases the etiology was undetermined or not specified. In the remaining number of cases 57.45 per cent, were classified under "other causes".

3.2.1 Infectious Diseases

The number of cases in which various infectious diseases were responsible for eye conditions, as found in the group investigated, is represented in Table 3.2.

TABLE 3.2

INFECTIOUS DISEASES

Disease	Frequency	%
Diphtheria	0	0.00
Gonorrhea	37	2.61
Measles	10	0.71
Meningitis	30	2.13
Scarlet Fever	2	0.14
Septicaemia	1	0.07
Smallpox	0	0.00
Syphilis prenatal	9	0.64
Syphilis postnatal	1	0.07
Syphilis, pre- or postnatal not specified	4	0.28
Trachoma	0	0.00
Tuberculosis	2	0.14
Typhoid Fever	0	0.00
Rubella	9	0.64
Onchocerciasis	0	0.00
Toxoplasmosis prenatal	1	0.07
Toxoplasmosis postnatal	0	0.00
Toxoplasmosis, pre- or postnatal not specified	3	0.21
Brucellosis	0	0.00
Leprosy	0	0.00
Other infectious disease, specified	10	0.71
Other infectious disease, not specified	21	1.48
Sub-Total A	140	9.90
None	1,277	90.10
TOTAL	1,417	100.00

It is seen from the table above that meningitis (2.13%) and venereal diseases (especially gonorrhea) were the most common diseases. Most of these diseases are however, nowadays effectively dealt with.

3.2.2 External cause

The incidence of different external causes of eye conditions is given in Table 3.3.

TABLE 3.3

EXTERNAL CAUSE

External cause	Frequency	%
Occupational activity	4	0.28
Military activity	1	0.07
Household activity	5	0.35
Play or sport other than occupational	25	1.76
Traffic or Transportation other than occupational	6	0.42
Birth process	3	0.21
Surgical or Medical procedure	15	1.06
Act of Violence	0	0.00
Other external cause specified	4	0.28
External cause not specified	38	2.68
Sub-Total B	101	7.13
None	1,316	92.87
TOTAL	1,417	100.00

A total of 101 cases was found in which the cause of the eye condition was of external origin. As regards Table 3.3 it is noted that play or sport other than occupational, is the most important (1.76%) external cause responsible for defective sight.

3.2.3 Causal Agent

The incidence of the causal agents responsible for defective sight in the 101 cases above is shown by Table 3.4.

TABLE 3.4

CAUSAL AGENT

Causal Agent	Frequency	%
Chemical causing burn	0	0.00
Radiation	0	0.00
Other agent causing burn	5	4.95
Firearm using explosive	11	10.89
Fireworks	0	0.00
Other explosive	4	3.96
Airgun or slingshot	2	1.98
Other sharp or pointed object	1	0.99
Blow or Fall	18	17.82
Foreign body in eye	4	3.96
Methylalcohol	6	5.94
Dinitrophenol	6	5.94
Lead	6	5.94
Quinine	0	0.00
Dysoxygenation	8	7.92
Other causal agent specified	8	7.92
Causal agent not specified	22	21.78
TOTAL	101	100.00

The most important agents in external causes of defective sight in the 101 cases are, firearms using explosives (10.89%) and blows or falls (17.82%). The incidence of dysoxygenation (7.92%)(disturbance in the saturation with oxygen) is however nowadays greatly reduced after discovering that it was responsible for retrolental fibroplasia.

3.2.4 Other Causes

As regards "other causes", due consideration is given to factors like general diseases and prenatal influences not classified elsewhere. The occurrence of these factors is indicated in Table 3.5.

TABLE 3.5

OTHER CAUSES OF EYE CONDITIONS

General Diseases not Elsewhere classified	Frequency	%
Anaemia or other blood disease	1	0·07
Diabetes mellitus	1	0.07
Nephritis or other kidney disease	2	0.14
Vascular disease (including arteriosclerosis and other cerebro-vascular lesions) ..	1	0.07
Multiple sclerosis	0	0.00
Intracranial neoplasm	10	0.71
Other disorder of central nervous system specified	0	0.00
Disorder of central nervous system not specified	7	0.49
Complication of pregnancy	1	0.07
Nutritional deficiency	1	0.07
Sarcoidosis	0	0.00
Other general disease not elsewhere classified not specified	5	0.35
General disease not elsewhere classified not specified	0	0.00
Sub-Total C	29	2.05
Prenatal Influence Not Elsewhere Classified		
Genetic origin established	369	26.04
Genetic origin probable	253	18.21
Other prenatal influence not elsewhere classified, specified	0	0.00
Prenatal influence, not specified	158	11.15
Sub-Total D	785	55.40

Etiology Undetermined or Not Specified

Causes unknown	92	6.49
Evidence insufficient for diagnosis	5	0.35
No report on etiology	265	18.70
Sub-Total E	362	25.54
Grand Total A + B + C + D + E	1,417	100.00

The most outstanding features in Table 3.5 are that genetic origin of eye conditions was established in 26.04 per cent of the group investigated and in 18.21 per cent of the cases genetic origin was probable. These frequencies include those individuals who had relatives with a similar eye condition and where the eye condition is considered to be inherited according to the literature (3). Consanguinity was also taken into account where the eye condition is of the inheritable type. Intracranial neoplasms (0.71%) and disorder of the central nervous system (0.49%) occurred relatively often. In 25.54 per cent of the cases the etiology was undetermined or not specified.

3.3 COMBINATIONS OF EYE CONDITIONS

It is known that many people suffering from blindness and impaired vision are afflicted with more than one eye condition at the same time.

The importance of calculating the occurrence of the various combinations of eye conditions was realized and subsequently attention is drawn to these results.

In the procedure, use was made of the index number representing each eye condition referred to earlier on and the absolute frequency of each eye condition in the different sections. This information will subsequently be imparted before explaining the system of combinations.

SECTION 1

EYEBALL IN GENERAL

Index		Frequency
01	Glaucoma (excluding infantile)	4
02	Myopia with detachment of retina	4
03	Myopia, detachment of retina not specified .	77
04	Panophthalmitis and acute endophthalmitis ..	0
05	Albinism	11
06	Anophthalmitis, excluding surgical	5
07	Hydrophthalmos (infantile glaucoma)	34
08	Microphthalmos	42
09	Aniridia	3
10	Coloboma, any part, excluding surgical	8
11	Multiple congenital anomalies	0
12	Other congenital anomaly specified	492
13	Congenital anomaly not specified	197
14	Disorganized eyeball (atrophic globe pthisis bulbi)	35
15	Other general affection of eyeball specified	137
16	General affection of eyeball, not specified.	368
	TOTAL	1,417

SECTION 2

CONJUNCTIVA

Index		Frequency
1	Purulent ophthalmia of newborn (ophthalmia neonatorum)	42
2	Purulent ophthalmia other than newborn ...	0
3	Other affection of conjunctiva specified .	6
4	Affection of conjunctiva, not specified .	6
0	None	1,363
	TOTAL	1,417

SECTION 3

CORNEA

Index		Frequency
01	Interstitial keratitis	5
02	Phlyctenular keratitis, including kerato-conjunctivitis	1
03	Ulcerative keratitis, including trachoma	1
04	Sclerosing keratitis	21
05	Keratitis, other type specified	3
06	Keratitis, type not specified	46
11	Corneal dystrophy, degeneration	0
07	Keratomalacia	11
08	Keratoconus	35
09	Other affection of cornea specified	10
10	Affection of cornea, not specified	5
0	None	1,279
	TOTAL	1,417

SECTION 4

LENS

Index		Frequency
1	Cataract	182
2	Dislocated lens	5
3	Other affection of lens specified	11
4	Affection of lens, not specified	1
0	None	1,218
	TOTAL	1,417

SECTION 5

UVEAL TRACT

Index		Frequency
1	Iritis, iridocyclitis or uveitis	3
2	Kerato-iridocyclitis or sclero-iridocyclitis	4
3	Choroiditis	7
4	Chorioretinitis	40
5	Sympathetic ophthalmia	6
6	Other affection of iris, ciliary body or choroid specified (incl. neoplasm)	120
7	Affection of iris, ciliary body or choroid not specified	0
0	None	1,237
	TOTAL	1,417

SECTION 6

RETINA

Index		Frequency
1	Retinopathy; including retinitis and lesions of retinal vessels	13
2	Retrolental fibroplasia	6
3	Detachment of retina	30
4	Tapetoretinal degeneration and allied conditions including retinitis pigmentosa	150
5	Macular degeneration	190
6	Neoplasm of retina	12
7	Other affection of retina specified	59
8	Affection of retina, not specified	0
0	None	957
	TOTAL	1,417

SECTION 7

OPTIC NERVE

Index		Frequency
1	Optic nerve atrophy, optic neuritis or papilledema	280
2	Other affection of optic nerve specified	209
3	Affection of optic nerve, not specified	6
4	Lesion of optic pathway or cortical visual centres	1
0	None	921
	TOTAL	1,417

SECTION 8

VITREOUS

Index		Frequency
1	Vitreous haemorrhage	0
2	Other affections of vitreous specified	3
3	Affections of vitreous not specified	1
0	None	1,413
	TOTAL	1,417

SECTION 9

SITE AND TYPE INDEFINITE OR NOT REQUIRED

Index		Frequency
1	Amblyopia ex anopsia	3
2	Amblyopia uni- or bilateral, not explained	4
3	Congenital nystagmus (only to be used if classification elsewhere is impossible)	17
4	Evidence insufficient for diagnosis	0
5	No report on site and type of affection	369
0	None	1,024
	TOTAL	1,417

As mentioned before the Index 0 represents the number of people not affected in the particular section. In the combinations of eye conditions the indices 4 and 5 of section 6 are both grouped under the index 5, because these two conditions often overlap in their manifestation and are sometimes difficult to distinguish clinically.

Similarly the indices 1 and 2, Section 8 are grouped under the Index 1. The frequency of each combination of eye conditions was compiled and compared with an expected value. This expected value is based on the independent association of the conditions in the combination including the index 0, calculated from the absolute frequency of each index in the combination expressed as a portion of the total.

An example will be given to illustrate the system. Referring to the fifth example in Table 3.6 the series with the index combination 12, 0, 0, 0, 0, 0, 1, 0, 0, respectively represents the sections 1, 2, 3, 4, 5, 6, 7, 8, 9. The index 12 (Section 1) represents congenital anomaly specified.

The index 0 is registered for each of the Sections 2, 3, 4, 5, 6, as well as for 8 and 9. The index 0 indicates that no apparent abnormal eye condition is recorded for the section in question. Section 7 with index 1 represents optic atrophy. Forty-seven individuals were found with this combination whereas 73.3411 were expected on the basis of independent occurrence of the indices. The chi-square (X^2) value, with one degree of freedom, as determined for each combination is used as a

relative measure to compare different combinations and hence determine to what extent the components in the combination are associated or not.

The following assumptions are made when the observed frequency of a combination is compared with the expected value.

(a) When the frequency of a combination is compared with its expected value and found to be approximately the same, it is concluded that the conditions in that combination, including the index 0, are independent and associated by chance or incidentally.

(b) When the frequency of the combination is greater than the expected value, it is concluded that the conditions in the combinations including the index 0, occur together more often than would be expected by chance. This could imply that these conditions may be mutually dependent, or that certain conditions are usually the result of others.

(c) When the frequency is less than the expected value, then conditions, including 0, occur less frequently together than would be expected by chance. A more frequent occurrence with other conditions, whatever they may be, is then postulated.

Two hundred and forty-five combinations of eye conditions were found. Only those combinations with a chi-square (X^2) value greater than 3.84 (one degree of freedom) and an observed frequency greater than 2 are considered, as a discussion on all the combinations will be too lengthy a procedure with little significance. This initial elimination was conducted to reduce the total number of combinations to those which are most significant with regard to the difference between observed and expected value.

Those combinations with an expected value approximately equal to the observed value in which the X^2 is less than 3.84 are excluded, because the observed conditions in the combination could then have occurred by chance.

Those combinations occurring only once are also excluded because such a combination could have occurred incidentally.

By means of this procedure 245 combinations were reduced to 41 which will be considered in the following tables.

First of all those combinations with an observed frequency less than the expected value will be considered in Table 3.6.

TABLE 3.6

OBSERVED FREQUENCY OF COMBINATION LESS THAN EXPECTED VALUE

Index	Combination	Observed Frequency	Expected Value	X^2
1.	3,0,0,0,0,0,0,0,0	3	21.6419	16.0577
2.	3,0,0,0,0,0,1,0,0	3	11.4781	6.2622
3.	7,0,0,0,0,0,0,0,0	3	9.5561	4.4979
4.	8,0,0,0,0,0,0,0,0	3	11.8046	6.5670
5.	12,0,0,0,0,0,1,0,0	47	73.3411	9.4606
6.	12,0,0,0,0,5,0,0,0	26	49.0775	10.8516
7.	12,0,0,0,6,0,0,0,0	2	13.4038	9.7022
8.	15,0,0,0,0,5,0,0,0	3	13.5661	8.2295

It is seen from the Table above that the most significant difference between the observed frequency and the expected value are examples 1 and 6. As regards the combination observed in the 26 cases in example 6 of the following can be assumed.

(a) Diagnoses were incomplete.

(b) Congenital tapeto-macular degeneration was a primary condition without an associated or secondary condition.

(c) The associated or secondary affections to congenital tapeto-macular degeneration expected had a later manifestation.

This method of analysis also applies to the other combinations in Table 3.6.

The significance of analysis in these cases lies therein that the individuals found with these combinations, could be re-examined to see if expected associated conditions are present and if not, then to certify that the combinations in Table 3.6 represent primary conditions.

Table 3.7 represents the combinations of eye conditions where the observed frequency is greater than the expected value.

TABLE 3.7

OBSERVED FREQUENCY OF COMBINATIONS GREATER THAN EXPECTED VALUE

Index combination	Observed Frequency	Expected Value	X^2
1. 3,0,0,0,0,5,1,0,0	25	4.0736	107.5005
2. 3,0,0,0,0,7,1,0,0	21	0.7069	582.5575
3. 8,0,0,1,6,5,1,0,0	6	0.0321	1109.5274
4. 8,0,4,0,0,0,0,0,0	4	0.1935	74.8808
5. 8,0,9,1,0,5,1,0,0	3	0.0090	994.0090
6. 12,0,0,0,0,0,0,0,2	3	0.3967	17.0838
7. 12,0,0,0,0,0,0,0,3	11	1.6863	51.4410
8. 12,0,0,0,0,1,1,0,0	3	0.9952	4.0386
9. 12,0,0,0,0,5,1,0,0	181	26.0292	922.6541
10. 12,0,0,0,0,6,0,0,0	9	1.7321	30.4961
11. 12,0,0,0,4,0,1,0,0	12	2.3696	39.1393
12. 12,0,0,0,4,5,1,0,0	4	0.8410	11.8659
13. 12,0,0,0,6,5,1,0,0	9	2.5230	16.6276
14. 12,0,0,0,6,7,1,0,0	3	0.4378	14.9951
15. 12,0,0,1,0,0,0,0,0	41	20.6460	20,0661
16. 12,0,0,1,0,5,1,0,0	14	3.8862	26.3210
17. 12,0,0,1,6,0,0,0,0	5	2.0012	4.4937
18. 12,0,4,1,6,0,0,0,0	3	0.0328	268.4230
19. 12,0,6,1,6,0,0,0,0	4	0.0718	214.9130
20. 12,0,11,0,0,0,0,0,0	4	0.5397	22.1857
21. 12,1,6,1,0,0,0,0,0	3	0.0222	399.4276
22. 12,1,0,0,0,0,0,0,0	13	4.1535	18.8420
23. 13,0,0,0,0,0,0,0,0	197	55.3695	362.2788
24. 14,0,0,1,6,0,0,0,0	3	0.1423	57.3889
25. 15,0,0,0,0,0,0,0,3	4	0.4661	26.7934
26. 15,0,0,0,0,3,0,0,0	6	1.1970	19.2721
27. 15,0,0,0,0,5,1,0,0	19	7.1950	19.3687
28. 15.0,0,0,4,0,1,0,0	3	0.6550	8.3954
29. 15,0,0,0,5,0,0,0,0	4	0.1852	78.5782
30. 15,0,0,1,0,3,0,0,0	6	0.1787	189.6336
31. 15,1,6,0,6,0,0,0,0	3	0.0039	2301.6962
32. 15,1,0,0,0,0,0,0,0	6	1.1481	20.5042
33. 16,0,0,0,0,0,0,0,0	368	103.7124	673.4771

In Table 3.7 the difference between the observed frequency and expected value is most significant in examples 2, 3, 5, 9, 18, 19, 21, 31. It is also seen that the combination of conditions in example 9 (congenital tapeto-macular degeneration and optic atrophy) occurred most often.

When the observed frequency of combinations of eye conditions is greater than the expected value as in Table 3.7 a different interpretation is necessary. In the ninth example (Table 3.7) for instance, the combination of the congenital conditions, tapeto-macular degeneration and optic atrophy occurred far more often with a frequency of 181 than the expected value of 26.0292, resulting in a X^2 value of 922.6541 (one degree of freedom) which makes the difference very significant.

It is thus postulated that the conditions in this combination should occur more frequently with other conditions by chance or incidentally. Reasons for the observed phenomenon could be that:

(a) Diagnoses were incomplete, but this is unlikely.
(b) The conditions in the combination are mutually dependent, or one is usually secondary to the other via a single or dependent cause (gene(s)).
(c) The conditions have independent separate causes (genes) which are genetically linked.

This method of interpretation also applies to the other combinations in Table 3.7.

In these cases the results are especially significant in that they could serve as an aid in future diagnoses of the eye, e.g. when tapeto-macular degeneration in a patient is found, specific attention should be paid to the optic disc to determine whether optic atrophy which is expected, occurs. Thus the possibility of the condition remaining undetected, is eliminated to a certain extent.

3.4 DEGREE OF VISION AND MEDICAL TREATMENT

It is known that many people considered as blind are not totally blind, but still have the use of their eyes to a certain extent, and that the outcome of medical treatment depends to a large degree on the amount of vision available.

An investigation of this nature would be of more value if the considerations above are included, and subsequently attention will be drawn to factors, such as visual acuity, field of vision and treatment received by the group investigated.

3.4.1 Visual Acuity

The visual acuity of the group investigated, ranging from 0 to 6/6, is indicated in Table 3.8 for each eye separately.

TABLE 3.8

VISUAL ACUITY

Visual Acuity	Right Eye	%	Left Eye	%
0	334	23.57	318	22.44
1/60	327	23.08	350	24.70
2/60	161	11.36	154	10.87
3/60	115	8.12	110	7.76
4/60	36	2.54	36	2.54
5/60	32	2.26	41	2.89
6/60	62	4.38	61	4.30
6/36	21	1.48	19	1.34
6/24	7	0.49	6	0.42
6/18	7	0.49	4	0.28
6/12	5	0.35	5	0.35
6/9	1	0.07	1	0.07
6/6	0	0.00	2	0.14
No information	309	21.81	310	21.86
Total	1,417	100.00	1,417	100.00

An outstanding feature, that is apparent from this table, is that the visual acuity differed in the right and left eye. However, it was the experience that the difference per individual was very limited.

In approximately 21 per cent of the cases no information concerning the visual acuity was obtained.

It is further also observed that only approximately 23 per cent of the cases were totally blind in the right or left eye, and that the majority of cases still retained a certain degree of vision. This fact might have some bearing on the teaching or training potential of these individuals.

3.4.2 Field of vision

The degree of field of vision (the area of visual perceptors

functioning in the eye ball), found in the group investigated, is given in Table 3.9 for both the right and left eye.

TABLE 3.9

FIELD OF VISION

Field of vision	Right Eye	%	Left Eye	%
Nil	334	23.57	318	22.44
Less than 25%	135	9.53	142	10.02
25% - 50%	340	23.99	347	24.49
Central Scotoma	27	1.91	28	1.98
Hemianopia	14	0.99	11	0.78
More than 50%	234	16.51	239	16.87
No Information	333	23.50	332	23.43
Total	1,417	100.00	1,417	100.00

From this table it can also be deduced that approximately 23 per cent had no vision in the right or left eye at all. This percentage coincides with the findings in the previous table. It is also seen that in the remaining cases, a certain degree of vision was present at the time of the diagnoses which coincides with the degree of vision found in the previous table. This finding indicates the importance of preventing progressive deterioration of eye conditions at an early stage.

3.4.3 Treatment Received and Results

The number of individuals who had received either medical or surgical treatment and the result thereof is given in Table 3.10.

TABLE 3.10 (a)

TREATMENT RECEIVED

Type of Treatment	Frequency	%
Medical	38	2.68
Surgical	227	16.02
Unknown	1,152	81.30
Total	1,417	100.00

TABLE 3.10 (b)

RESULTS OF TREATMENT

Results of Treatment	Frequency	%
Beneficial	112	42.26
Non-beneficial	149	56.23
Uncertain	4	0.61
Total	265	100.00

It is seen from table 3.10 that out of the 265 cases who had received medical or surgical treatment only 112 cases benefited. It should however, be kept in mind that these cases are chronic eye conditions for which treatment is seldom effective.

More information concerning the unknown cases, which are in the majority, would be of considerable value.

3.4.4 Prognoses

Table 3.11 illustrates the frequencies of prognoses at the time of the diagnoses.

TABLE 3.11

PROGNOSES

Prognoses	Frequency
Stationary	147
Improvement	26
Uncertain	1,244
Total	1,417

It is seen that in only 26 cases improved eyesight is possible and that in the majority of cases the prognosis is uncertain. More information on the uncertain number of cases is a necessity.

CHAPTER 4

FAMILY PEDIGREES AND RELEVANT OBSERVATIONS

4.1 INTRODUCTION

In this chapter consideration is given to observations concerning relatives of the individuals in the group investigated, regarding consanguinity and miscellaneous findings.

4.2 CONSANGUINITY

The occurrence of consanguinity reported in the families of the group investigated is indicated in Table 4.1.

TABLE 4.1

CONSANGUINITY

Consanguinity	Frequency
Yes	125
No	524
Uncertain	768
Total	1417

As seen, only 125 cases where consanguinity occurred in the family, were reported in the files. However many more are suspected as the experience from personal interviews and compiled family pedigrees indicated. It was also the experience that in some of the cases where consanguinity was denied in the files, the contrary was proved.

In the completed family pedigrees where consanguinity could not be established, the surnames of ancestors were often identical.

As such, consanguinity is not the cause of blindness or any defect for that matter, but it is instrumental in bringing together the defective genes or factors responsible for the defect.

4.3 INFORMATION CONCERNING RELATIVES

Virtually no information concerning the relatives of the pupils was on record at the School for the Blind. In a few instances the maiden name of the pupil's mother was stated and only in exceptional cases was mention made of relatives suffering from blindness or impaired vision. This limitation hampered the investigation considerably, and interviews were the only means by which such information could be obtained.

4.3.1 Miscellaneous Findings

The following observations were made from the information obtained in family pedigrees of the group investigated.

(a) In some families where blindness and impaired vision occurred, relatives were suffering from defects such as deafness, epilepsy and low I.Q.

(b) It was found that many of the old pupils were related to present pupils of the school.

(c) In many families the blindness or impaired vision of brothers and sisters was attributable to different eye conditions. An undiagnosed eye condition should not be assumed to be identical to a brother's or sister's recognized defect.

(d) It was interesting to note that in certain families the children of married cousins were devoid of any defect (known) but that eye defects occurred in the next generation.

(e) Cases were found where the blind pupil's parents were related and not blind, but where blindness had occurred in the previous generation. This then could be a case of a dominant condition with incomplete penetrance.

(f) Only a few cases were found where both parents were blind and suffering from the same condition as the child.

(g) In certain families cousins were suffering from exactly the same eye condition.

(h) Cases were found where the occurrence of an eye condition was omitted in one or two generations.

(i) In some families certain eye conditions revealed a dominant mode of inheritance with a reasonable measure of penetrance but with variable expression. In such cases blindness occurred in successive generations but with variable severity.

(j) Cases were discovered where no blindness or consanguinity occurred in the family of the pupil.

(k) In some cases a member of a family may have attended the School for the Blind while brothers or sisters suffering from the identical eye condition managed when aided, to attend a normal school.

(l) Cases were found where both parents were blind with all, some, or none of the children afflicted with blindness.

(m) A few cases were found where brothers and sisters were suffering from an identical eye condition.

(n) Two cases were found where blindness could be sex-linked; additional information would be required however, to confirm the finding.

(o) A dominant mode of inheritance is possible in cases of

(i) Retinoblastoma (Tumor arising from retinal germ cells);

(ii) Tapetoretinal degeneration;

(iii) Myopia, without detachment of the retina, with myopic degeneration and optic atrophy;

(iv) Retinal detachment;

(v) Infantile glaucoma (intra-ocular pressure).

(p) In most cases the eye condition followed a recessive mode of inheritance.

(Records of pedigrees to illustrate the cases mentioned above are available on request).

CHAPTER 5

SUMMARY, GENERAL DISCUSSION AND RECOMMENDATIONS

5.1 SUMMARY

This chapter concludes the report on hereditary blindness with a brief summary of the contents, followed by a general discussion and recommendations submitted.

5.1.1 Introduction

Based on observations it was assumed that in many cases hereditary factors were most probably the cause of blindness and impaired vision suffered by individuals who had attended the School for the Blind at Worcester since its establishment in 1881, to 1965. This motivated an investigation to determine in which cases hereditary factors played a role.

A total of 1417 individuals of whom 97.8 per cent had attended the School for the Blind at Worcester was included in the group investigated. A questionnaire, designed for the purpose, was completed for each person. Information concerning the cause of the eye conditions and family data was obtained from personal files at the School for the Blind and from personal interviews.

5.1.2 Eye Conditions and Other Medical Information

For the group as a whole the sex of the individuals was considered. Here a near three to two ratio of boys to girls was found.

The age at onset of eye trouble was found to occur mainly (46.65% of the cases) at birth or shortly after birth, whereas the age at onset of blindness was comparatively predominant in the higher age groups.

In a classification of eye conditions according to site and type, it was found that, considering the eyeball in general congenital anomalies were most common, and as regards the different components of the eyeball, defects of the optic nerve, retina and choroid were the most important causes of blindness and impaired vision.

Of interest was the finding of 4 different syndromes in the group investigated. In addition to defective sight, individuals were sometimes afflicted with additional anomalies such as deafness, anaemia, epilepsy and many other types.

5.1.3 Etiology of Eye Conditions

The individuals were also classified according to the etiology of the eye conditions, as regards infectious diseases (9.88%), external causes (7.13%) other causes (57.45%) and etiology undetermined or not specified (25.54%). In 26.04 per cent of the cases genetic origin was established and classified under "other causes"; and in 18.21 per cent of the cases genetic origin was probable.

5.1.4 Combinations of Eye Conditions

It was known that one person could be afflicted with more than one eye condition simultaneously and subsequently the frequencies of combinations of eye conditions were calculated which were compared with an expected value. The observed occurrence of the following primary conditions, myopia, optic atrophy and tapeto-macular degeneration without associated conditions, was most significant.

On the other hand various combinations were found which occurred far too often to be ascribed to chance. Of these the following were most significant: Congenital tapeto-macular degeneration + optic atrophy, and microphthalmos + cataract + iris atrophy + tapeto-macular degeneration + optic atrophy. The interpretation and significance of these combinations have been pointed out.

5.1.5 Degree of Vision

It was found that as regards visual acuity and field of vision approximately 23 per cent of the cases were blind in either the left or right eye, and that most of the other cases had some degree of vision.

5.1.6 Treatment Received

Of the 265 cases found to have undergone either medical or surgical treatment only 112 gained benefit from it. Prognoses indicating improvement was found in only 26 cases.

5.1.7 Family Pedigrees and Relevant Observations

Virtually no information concerning the relatives of the pupils was on record at the School for the Blind. However, by means of personal interviews, a total of 100 family pedigrees could be compiled. From information thus obtained cases were found where information obtained from the files of individuals

concerning consanguinity of the ancestors was not reliable.

In most cases a recessive mode of inheritance was the cause of defective sight. Dominant modes of inheritance were less frequent whereas a X-linked mode of inheritance could be possible in 2 cases. In addition to these modes of inheritance others with a variable measure of penetrance and expressivity were encountered.

5.2 GENERAL DISCUSSION AND RECOMMENDATIONS

This survey is the first of its kind and valuable information has come to light. Although the investigation cannot supply all the answers to the questions as outlined, ample material for consideration has been provided.

It is realized that a considerable amount of time and energy was spent to obtain little significant information. With a view to alleviating the task of a comprehensive investigation of this nature in the future suggestions concerning the filing system at the School for the Blind and other detailed information are given. Further measures considered as essential for the prevention of blindness and impaired vision are submitted.

5.2.1 Filing System

It is recommended that the present system of filing at the School for the Blind at Worcester be adapted to one where the files are more permanent and more easily accessible. The information on each individual should be kept in a predetermined factual sequence.

5.2.2 Detailed information

More detailed medical information regarding each pupil is a prerequisite. The necessary information should include:

(a) Cyto-genetic studies.

(b) Routine clinical and pathological analysis, e.g. of blood, urine, X-rays, encephalogram analysis and other analyses.

(c) A more detailed description of the eye condition which will include e.g. the use of an electroretinogram.

(d) Colour photographs of the individual and especially the fundus (inside the eyeball) of the eye. By this means a

record of the actual eye condition can be kept from which comparisons can be drawn and standard diagnoses maintained.

(e) Compilation and filing of family pedigrees.

It is considered that, when these procedures are followed, means of establishing the causes of blindness will be greatly improved. Two important measures which can then be applied are firstly, the prevention of the causes and secondly, the application of corrective treatment in established cases before progressive deterioration sets in.

5.2.3 Preventative Measures

Steps that could be taken in the national interest by various organizations for the prevention of blindness at this stage are the following:

(a) Information concerning all individuals suffering from blindness and impaired vision in the Republic of South Africa should be kept in a central register. This register should strictly confirm to the requirements of an effective recognised system of filing and control.

(b) The organization in control of such a register should collaborate far more extensively with active research in Ophthalmology and Genetics than is experienced at present.

(c) Effective screening of infants should be conducted to detect eye conditions at an early stage to apply more effective treatment and thus arrest progressive deterioration.

(d) The detection of heterozygotes or carriers of different eye conditions is a most important factor in aiding genetic counselling.

Based on these foundations, active genetic counselling can be conducted. In addition valuable contributions to basic and applied research could be made and thus preventative measures to combat blindness and impaired vision could materialize.

5.3 CONCLUDING REMARKS

This investigation furnishes conclusive proof that further research in this field is essential. One cannot help thinking of the even larger number of individuals suffering from other

hereditary disorders such as deafness, epilepsy, mental deficiency and many other primary chronic diseases which could possibly be alleviated or prevented in many instances if the necessary facilities are provided in an institute or concern engaged in active research on human genetics.

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