PATTERN OF OCULAR FINDINGS IN PERSONS WITH ALBINISM IN KENYA

Research proposal for dissertation in part fulfillment for the degree of

Master of Medicine, Ophthalmology, University of Nairobi

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DECLARATION

This dissertation is my original work and has not been presented for a degree in any other university.

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LIST OF ABBREVIATIONS AND SYMBOLS

AFEA	Albinism Foundation of East Africa
BCVA	Best corrected visual acuity
BL	Blind
CHS	Chediak-Higashi syndrome
cm	Centimetres
D	dioptre
HPS	Hermansky-Pudlak syndrome
LogMAR	logarithm of the minimum angle of resolution
NLP	no light perception
OCA	oculocutaneous albinism
OA	ocular albinism
PL	perception of light
SVI	severe visual impairment
W.H.O	World Health Organization
VA	visual acuity
v.i.	visual impairment
≥	equal or more than
≤	equal or less than
±	with or without

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ABSTRACT

Aim

The aim of this study was to establish the ocular findings in persons with albinism in Kenya.

Method

This was a school based cross-sectional study conducted in schools for the blind and integrated education programmes in Eastern and Central province. All the pupils with albinism present during the study period were examined, giving a total of one hundred and one pupils. Assessment of visual acuity and stereoacuity for each pupil was carried out. Amsler grid was used to test macula function. Orthoptic assessment was done followed by anterior segment examination using a torch, +20 dioptre loupe and a portable slit lamp. Posterior segment examination was performed using a direct and indirect ophthalmoscope. Objective refraction was performed on all the pupils. All pupils with conditions requiring management were given appropriate treatment and referrals.

Results

A total of 101 pupils were examined, 49 males and 52 females. Common ocular complaints were photophobia(91%) and reduced vision(78%). Majority of the pupils (83%) were wearing photochromic spectacles, prescribed to them by an ophthalmologist. Most of the pupils had moderate visual impairment (86%). Nystagmus (98%) and strabismus (91%) were common ocular findings. Stereopsis was absent in most pupils (93%). Amsler grid assessment was normal in most cases (79%).Common refractive errors were hypermetropic astigmatism(46%) and myopic astigmatism(28%).The commonest anterior segment finding was iris trans-illumination defects(87%). The commonest posterior segment findings were foveal hypoplasia(100%), hypopigmentation of the retina(97.5%) and optic nerve hypoplasia(97%).

Conclusion

Most of the pupils examined had moderate (86.5%) and to a lesser extent severe visual impairment (5%). Common ocular symptoms were photophobia and reduced vision. Common

ocular findings were iris transillumination defects, foveal hypoplasia, retinal hypopigmentation and optic nerve hypoplasia. A strong relationship was found between reduced vision and foveal hypoplasia, nystagmus, optic disc hypoplasia, refractive error and strabismus. However, these findings were also present in the pupils without visual impairment. Therefore, it was difficult to single out one main cause of reduced vision in these pupils. The commonest refractive errors found were hypermetropic astigmatism and myopic astigmatism. Majority of the pupils were wearing photochromic spectacles, prescribed to them by an ophthalmologist.

1.0 INTRODUCTION

Albinism refers to a group of hereditary disorders that involve an abnormality of melanin synthesis or distribution.

The term albinism comes from the Latin word *albus*, which means white, and, in 1908, Garrod first scientifically described it.[1] Clinically, albinism presents as a pigmentation abnormality of the skin, the hair, and/or the eyes. Albinism can be divided into 2 broad categories, as follows: oculocutaneous albinism and ocular albinism. Oculocutaneous albinism involves both the skin and the eyes, whereas ocular albinism mainly affects the eyes with minimal to no skin involvement.

The primary morbidity of both oculocutaneous albinism and ocular albinism is eye related. Signs and symptoms include photophobia, monocular vision, strabismus, pendular nystagmus, iris transillumination defects, reduced vision due to refractive errors, foveal hypoplasia, and abnormal decussation of the optic nerve fibers. These ocular manifestations are almost always present in both forms of albinism; however, the degree of their presentation can vary depending on the type of albinism and the racial background of the patient.

The actual number of Kenyans with albinism is unknown: 'There are no available statistics on the incidence of albinism in the country, and we are almost totally ignorant about the needs of those born with this genetic condition.'[2] There is growing evidence of social discrimination and stigmatization directed towards this population.[3,4] Along with their differences in appearance, a lack of knowledge about albinism in the community leads to such stigma.

There are still many gaps in our knowledge of visual impairment in African children. Much information has come from schools for the blind. Previous studies in east Africa reported that many children are inappropriately enrolled in these schools and taught only Braille.[5,6,7]

There is a paucity of data on the visual status and ocular morbidity in albinos in Kenya or even Africa. As a result, a lot of information is needed in order to prepare programs to address ocular morbidity in this population.

2.0 LITERATURE REVIEW

As mentioned before, not much attention has been directed towards studying ocular findings in albinos, less so in African albinos. As a result, no study has been carried out on the ocular features in albinism in Kenya.

2.1 Prevalence

Prevalence of albinism worldwide differs depending on the region. A school-based study in Zimbabwe ascertained 157 albinism cases among 772,758 primary school pupils, giving a prevalence of 1 in 4,922. Similar results (1/4,476) were found among secondary school pupils.[8]

In Kenya, the exact figures are unknown. However the AFEA estimates a prevalence ratio of 1:5,000, approximately 6,000 people countrywide[2].

In the U.S.A the frequency of type I oculocutaneous albinism is approximately 1 in 17,000 to 1 in 20,000. The frequency of type II oculocutaneous albinism is higher in the African American population, where it can be 1 in 10,000.

The prevalence in parts of Africa therefore, is far higher than the global average.[3]

2.2 Children in schools for the blind

Tumwesigye et al[5] described the inappropriate enrolment of children in schools for the visually impaired in East Africa. A total of 1062 children were examined. 361 children had a presenting visual acuity greater or equal to 1.00. Retinal disease accounted for 95 of these cases. Albinism accounted for 73 out of 95 of them. In Kenya 76 out of 265 children in these schools had VA $\geq$ 1.00(Appendix I). The number of non-blind children enrolled in special schools and annexes for the blind in these countries is high and has apparently increased since it was reported to be 22% in 1995 in schools in the same areas.

Njuguna et al[9] studied causes of severe visual impairment and blindness in children in schools for the blind in Kenya, Uganda, Malawi and Tanzania in 2007. They also evaluated changes in the previous 14 years. A total of 1062 pupils less than 16 years old were examined, of whom 701 (66%) were blind or severely visually impaired (better eye vision <1.00). Pupils came from 3,15, 12, and 14 different schools in Kenya, Malawi, Tanzania and Uganda respectively. Refractive error, accounted for 4.3% (32 cases) in 2007 and this included 17 children with albinism. All these children improved to 1.00 or better with refraction. This study provides a better idea of the need for refractive services among SVI/BL children in the schools because this information is useful for those planning services.

Gilbert et al[6] examined 491 pupils in schools for the blind in Kenya, Malawi and Uganda in 1995. The findings for children in the schools in Kenya: At presentation, 11.0% had no visual impairment, 22.7% had visual impairment (< 0.48-1.00); 17.2% were severely visually impaired (SVI) (<1.00-1.30) and 49.1% and had vision in the range of less than 1.30 - no light perception (BL). Overall, 62.9% were blind and 14.1% severely visually impaired, but 1 in 3 and 1 in 4 pupils in Kenya and Uganda, respectively, had a visual acuity in the better seeing eye of 1.00 or better.

2.3 General features

Patients with albinism usually present in early infancy.[1] Witkop C.J described the chief clinical characteristic as hypopigmentation of the skin, hair and eyes (oculocutaneous albinism) or just the eyes(ocular albinism)[10]

2.4 Ocular symptoms

Photophobia

Parents often report that their children with albinism squint or close their eyes when exposed to bright light, beginning in infancy. Abadi et al found that increased light scattering in eyes of

individuals with albinism is reported, and that represents reduced filtering of light by the deficient ocular melanin pigment. Lack of uveal and retinal pigment results in high levels of retinal irradiance. Light scatter by the iris and multiple internal reflections produce both discomfort and disability glare. The combination of these two features is termed photophobia.[11,12]

Reduced vision

Abadi et al found that in general, visual acuity in albinism tends to fall within the range of 1.00 to 0.60.[12] Resolution is reduced in albinism due to several reasons. Not only is the retinal image degraded by optical blur (as a result of high refractive errors) and intra-ocular light scatter, it is also constantly in motion over a retina without a fully differentiated fovea. Wildsoet at al[13] studied albinism and its implications for refractive development and found all their pupils showed reduced acuity; the mean visual acuity of the group was 0.90 compared to the standard for normal visual acuity of 0.00 logMAR units. Summers CG[14] examined a group of 29 persons with albinism. However, only those patients who were noted to have the appearance of melanin pigment present within the macula on ophthalmoscopic examination were studied. She found that twenty of the 29 patients had visual acuity of 0.48 or better, and only 7 patients had vision better than 0.30.Her results from a selected group of patients with albinism and clinically detectable melanin pigment in the macula, suggest that this sign is associated with better visual acuity.

Most of the text books of ophthalmology only mention visual acuity being less than 1.00 in Albinism.[15]

2.5 Ocular features

Refractive errors

Published refractive profiles for albino populations are generally abnormal, with high refractive errors, including high with-the-rule astigmatism, being frequently encountered. [13,16,17,18,19] However, there are discrepancies between studies in terms of the overall bias in refractive errors, with both myopia[15,16] and hyperopia[13,18,19] being reported.

Wildsoet et al[13] found all their subjects had reduced visual acuity (mean: 0.90, logMAR) and overall, there was a bias toward hyperopia in their refractive errors (mean: +1.07 D). However the refractive errors of the group covered a broad range (SD: 4.67 D) and included both high myopia and high hyperopia. On average, albinos were found to have with-the-rule astigmatism.[15, 20] Sampath et al[20] studied distribution of refractive errors in albinos and persons with congenital nystagmus. None of the persons with albinism were emmetropic and they found horizontal nystagmus in all their subjects.

Nystagmus

Nystagmus is a universal feature of all forms of albinism. Abadi et al, [21] Collewijn et al[22] and St John et al[23] described the bilateral, involuntary, ocular oscillations as conjugate, occurring predominantly in the horizontal plane, although torsional and occasionally vertical eye movements have also been recorded.

Wildsoet et al[13] studied twenty-five albino subjects ranging in age from 3 to 51 years. All exhibited horizontal pendular nystagmus. Summers et al[14] found nystagmus in 26 out of 29 subjects. The lower prevalence may be due to the fact that only those subjectss who were noted to have the appearance of melanin pigment present within the macula on ophthalmoscopic examination were included in that study.

Strabismus

Abadi et al[12] and Summers CG[14] report that a high incidence of strabismus is reported amongst the albino population with both horizontal and vertical deviations being noted. Presence of strabismus can be traced to the misrouting of nerve pathways from the retina to the brain's visual cortex.

Stereoacuity

Lee et al[24] studied stereopsis and clinical correlates in 45 patients with albinism. They excluded pupils with strabismus of more than 10 prism dioptres and found 19 with stereoacuity measured with the Titmus vectograph. The individuals who had stereoacuity had significantly better visual acuity, more pigment in their iris, reduced or absent nystagmus, had melanin pigment identified in their maculae and a rudimentary foveal reflex when compared with those without measurable stereoacuity. Summers et al[14] found 6 out of 29 pupils had stereoacuity. Again, it is important to note that the higher prevalence of stereoacuity found may be due to the fact that only those patients who were noted to have the appearance of melanin pigment present within the macula on ophthalmoscopic examination were included in that study.

2.5.1 Anatomical features

2.5.1.1 Anterior segment

Transillumination of iris

In albinism, because of deficient melanin pigment in the iris stroma and posterior iris epithelium, light reflected from the retina is not filtered, and individuals with albinism show pink, diaphanous-appearing irides. Summers et al[14] found only 2 out of 29 pupils had full iris pigment (no iris transillumination).

2.5.1.2 Posterior segment

Foveal hypoplasia

Abadi RV and Dickinson CM[25] found the albino central retina to have unique features. Absence of a foveal pit, and hence no foveal reflex; Reduction in the usual foveal hyperpigmentation; Lack of the macular pigment xanthophyll and there may be retinal vessels which cross the fovea. Naumann et al[26] and O'Donnell et al[27] found histological evidence for this, reporting that the ganglion cell layer seems to be present throughout the retina and that no rod-free area is identifiable. Spedick et al[17] observed small optic discs in patients with albinism.

2.5.2 Visual pathway abnormalities

Guillery et al[28] and Hoffmann et al[29] found that instead of remaining on the ipsilateral side, afferent fibres from the first 20 degrees of both temporal retinae decussate with fibres from the nasal retinae and project to the contralateral lateral geniculate nucleus (LGN). As a result of this anomalous innervation, there is a fragmentation of the normal retinotopic organization of the LGN, which is reflected in a disruption of its laminar structure. The demonstration of optic track misrouting by visual evoked potential studies is currently the single definitive diagnostic test to confirm a diagnosis of albinism[24,30]. The major consequence of this disorganization of the visual pathway is that all albinos lack the neuroanatomical substrate for normal binocular vision.[12]

2.6 Other syndromes

Hermansky-Pudlak syndrome

Described in 1959 by Hermansky F and Pudlak P[31], this syndrome is a rare lysosomal storage disease of the reticulo-endothelial system. It is associated with haemorrhagic diathesis due to platelet dysfunction and the systemic accumulation of a ceroid-like substance. Kinnear PE and Tuddenham GD[32] studied 4 patients with the syndrome. The patients report a lifelong history of easy bruising and prolonged bleeding after minor injuries or surgical procedures. The incidence of this disease has been found to be higher among people of Puerto Rican origin.

Chediak-Higashi syndrome

Described in 1952 by Chediak M [33] and Higashi O [34] in 1954 this disease is rare and characterized by leucocytic abnormalities leading to recurrent infections. Leukocytes contain giant cytoplasmic granules, suggesting a membrane defect. They also exhibit prolonged bleeding times and easy bruisability. Individuals with CHS who survive the recurrent infections typically develop cranial and peripheral neuropathies and serious lymphoreticular infiltration.

3.0 RATIONALE

There is a large number of non-blind albino children enrolled into special schools for the blind.[5,6,9]. Currently, policies or guidelines regarding admission of children to schools or annexes for the blind are limited or non-existent[5]. Children can be referred for admission by health workers, teachers, parents or others. Children who attend such schools may have trouble integrating into society after leaving school. Furthermore, schools for the blind are often ill equipped to provide for children other than those requiring Braille, which roughly means vision of 1.80 or less. Once the magnitude of treatable or correctable ocular morbidity in this population is addressed, measures can be taken to improve the situation.

This information will facilitate further education of health workers. Albinism, being poorly understood, even by health workers, exposes albino children to stigmatization arising from superstitious beliefs within their community.[3] Once educated, the health worker can reassure parents that the reason their child appears different from the siblings is scientific rather than witchcraft or superstition.

There is currently no data on ocular findings in albinism in Kenya. Previous studies on pupils attending special schools for the blind classify albinism simply under 'Retina.'[5] There is therefore a large gap in the specific symptoms and findings in persons with albinism. It is not possible to overestimate the importance of this information, especially in planning of health care programs.

Previous studies conducted on persons with albinism have been in Western countries, in white populations. This data may not be representative of our albino population here in Kenya.

4.0 OBJECTIVES

4.1 Major objective

The main objective is to describe pattern of ocular findings in persons with albinism in Kenya.

4.2 Specific objectives

1. To determine ocular symptoms in persons with albinism.
2. To determine causes of reduced vision in the study population.
3. To describe pattern of refractive errors in persons with albinism.
4. To determine the use of refractive correction/low vision aids in the study population.

5.0. METHODOLOGY

5.1 Study population

Persons with albinism attending schools for the blind and integrated education programmes in Eastern and Central Province.

These include pupils attending Thika School for the Blind, St.Lucy's Igoji and Kitui Integrated Programme.

5.1.1 Inclusion criteria

Albino persons attending schools for the blind and integrated education programmes in Eastern and Central Province, Kenya, in whom a complete ocular examination was performed.

5.1.2 Exclusion criteria

Pupils without informed consent.

5.2 Study design

A school-based cross-sectional descriptive study.

5.3 Sample size determination

The schools that cater for the pupils with albinism in Kenya:

1. Thika School for the blind- 40 pupils
2. St.Lucy's Igoji, Meru- 46 pupils
3. Kitui integrated program-22 pupils (boarding)
4. Likoni school for the blind-20 pupils
5. St.Oda-15 pupils
6. Kilimani integrated program-10 pupils
7. Muthaiga integrated program- 4 pupils
8. Our Lady of Mercy, Shaurimoyo- 4 pupils
9. St.Lucy's Kibos-1 pupil
10. St.Francis Kapenguria- not able to get information

The sample size was determined using the statistical formula for cross sectional population studies where

$n = Z^2 Pq/d^2$

Where:

- nf= desired sample size
- Z= is the standard normal deviate. At 95%CI it is set at 1.96 and
- P= is the proportion of the target population estimated to have a particular characteristic. In this study there is no available estimate 0.50 is used.
- q= 1-p
- d= is alpha, either 0.05

therefore : $n= 1.96^2 * 0.5*0.5/0.05^2$

n= 384

The schools are sampled purposively therefore based on the schools with more pupils, Meru, Thika and Kitui formed the sampling frame .

Since the population of persons with albinism is less than 10,000, records show 108 pupils in the sampling frame; therefore

$nf = n/1+ n/N$

nf = 384/(1+384/108)

desired n= 84 pupils

5.4 Study Definitions

Foveal hypoplasia : Absence of a foveal reflex; Reduction in the usual foveal hyperpigmentation; Lack of the macular pigment xanthophylls; Retinal vessels which cross the fovea.

Iris transillumination : Defects in iris pigment which may be scattered or complete absence of pigment, with visualization of the edge of the lens

Nystagmus : A repetitive involuntary back and forth oscillation of the eyes

Visual impairment : Visual acuity of worse than 0.48(6/18) and up to 1.30(3/60) (LogMAR)

(Appendix 1)

Significant hyperopia : more than +2 D[35-43]

Significant myopia : more than -0.5 D[35-43]

Significant astigmatism : more than ± 1.5 dioptre (D)

Hyperopic astigmatism : astigmatism in which all meridians are hyperopic but to different degrees, or one meridian is hyperopic while the one opposite to it is without refractive error

Mixed astigmatism : astigmatism in which one meridian is hyperopic and the other is myopic

Myopic astigmatism : astigmatism in which all meridians are myopic but to different degrees, or one meridian is myopic while the one opposite to it is without refractive error

Strabismus : Misalignment of the eye, which may be manifest(tropia) or latent (phoria)

5.5 Procedure

5.5.1 Administrative logistics

Approval of the Albinism Foundation of East Africa was obtained and presented to the School Head-teachers in Eastern and Central Province. The examination was done at a pre-arranged time to ensure minimal interference with the school schedules. Informed consent was given by the Head-teachers of the respective schools. A brief explanation of the procedure was given to the teachers and children on arrival at each school. A demonstration of key tests like taking of visual acuity was done. All children present on the day of examination with consent given were recruited into the study.

Registration of each pupil and findings were recorded in individual questionnaires.

5.5.3 Assistants

Besides the principal researcher, there was one assistant to help in collecting of the data. The study involved both quantitative and qualitative parameters so as to capture any morbidity in the pupils. Thus, to ensure standardization and reproducibility of the findings, one resident in the department of Ophthalmology of comparable length of training was recruited. To enhance the examination techniques already attained during their training, both examined patients in the paediatric eye clinic, run twice a week in the base unit (KNH) and findings were validated by the consultant in charge, who is one of the supervisors in this study.

5.5.4 History taking

Any known history and complaints that was related to an ocular problem was noted with the help of teachers.

5.5.5 Measurements

5.5.5.1 Visual acuity

This was the first assessment done in a well-lit area using near LogMAR charts with distance equivalent, held at a distance of 25 centimetres(cm). Visual acuity for each eye was taken separately after occluding the other eye. Each correctly read line was recorded as the visual acuity. This assessment will be repeated after best subjective correction.

5.5.5.2 Stereoacuity

The Titmus Fly Test was used to assess stereoacuity. When held at a distance of 40 cm. the fly has a disparity of 3,600 sec of arc. In the stereoscopic assessment the pupils were asked to indicate the subjective plane of the fly's wings manually by their own finger. The test card was turned 90° in order to rule out the use of monocular cues. The pupils that were able to see the wings of the fly at 0° and 180° were subjected to the stereoanimals and stereocircles test.

5.5.5.3 Orthoptic assessment

Any abnormal head posture was noted followed by Brueckner test using a direct ophthalmoscope (Heine K180 Ophthalmoscope) at one metre; corneal light reflexes (Hirschberg) using a torch and finally cover tests at far and near.

Extraocular motility was assessed and presence of nystagmus noted.

5.5.5.4 Amsler Grid Test

The Amsler Grid chart was used to describe any defects in macular function. The chart used consists of a grid of black lines on white paper. The chart was held at a distance of 25 cm and each pupil was asked several questions as follows: 'Can you see the central white dot? If you can see the central black dot keep looking directly at it. Do you see all four corners and all four sides of the chart? Are there any areas of the chart that are missing or distorted in any way and are any of the lines not straight or unequal in size? If not, describe what you see then draw what you see.'

5.5.5.5 Anterior segment examination

This was examined using a magnifying loupe (20 D Volk double aspheric) and torch or direct ophthalmoscope and a portable hand-held slit lamp biomicroscope.

Following is a description of the minimal characteristics identified per structure:

Lids: masses, scars

Conjunctiva: discharge, injection, masses

Cornea: clarity, opacity

Anterior chamber: depth, hyphema, hypopyon

Iris: presence, shape, transillumination defects

Pupil: assessment using a source of light in dim lighting to determine anisocoria,, reaction to light, abnormal pupillary reflex

Lens: presence, clarity, position, opacity

5.5.5.6 Posterior segment examination

All pupils had their pupils dilated with Tropicamide dilating drops for fundus examination.
Direct and indirect ophthalmoscopes were used to look for optic nerve, macula and retinal pathology. Particular attention was paid to the macular area, looking out for absence of a foveal reflex; reduction in the usual foveal hyperpigmentation; lack of the macular pigment and retinal vessels which cross the fovea.

5.5.5.7 Cycloplegic refraction

This was performed on all pupils. One drop of 1% cyclopentolate was instilled into each eye three times at 10 minute intervals. Objective refraction was done 45 minutes after instillation of the first drop using a Heine Beta 200 Skiascope.

5.5.6 Referrals

Three pupils with conditions requiring management (allergic conjunctivitis, post operative uveitis) were given appropriate treatment and referrals.

5.5.7 Data collection and analysis

Structured questionnaires were used to collect data and stored in confidential files. All data was entered into a computer database designed in Microsoft Access. The data was exported to the Statistical Package for Social Scientist software (SPSS) version 12.5 for data analysis.

Data was cleaned and validated before actual analysis and analysis was done using appropriate statistical tests with the aid of a biostatistician.

P-value of <0.05 was considered statistically significant.
Presentation of data in the form of tables, graphs and pie charts where appropriate.

6.0 ETHICAL CONSIDERATIONS

1. Informed consent by pupil.
2. Confidentiality of each pupil's records was observed throughout the study.
3. No invasive examination was carried out. Use of medication for the purpose of examination was explained to the parents/ teachers.
4. Approval by Kenyatta National Hospital Ethical Committee.
5. Facilitation by Albinism Foundation of East Africa.
6. Approval by the appropriate School Principals.
7. Pupils with other ocular findings or diseases requiring management were treated and referred to the nearest eye unit as appropriate.
8. Discussion of findings, making of recommendations and submission of results to the University Of Nairobi Department Of Ophthalmology.

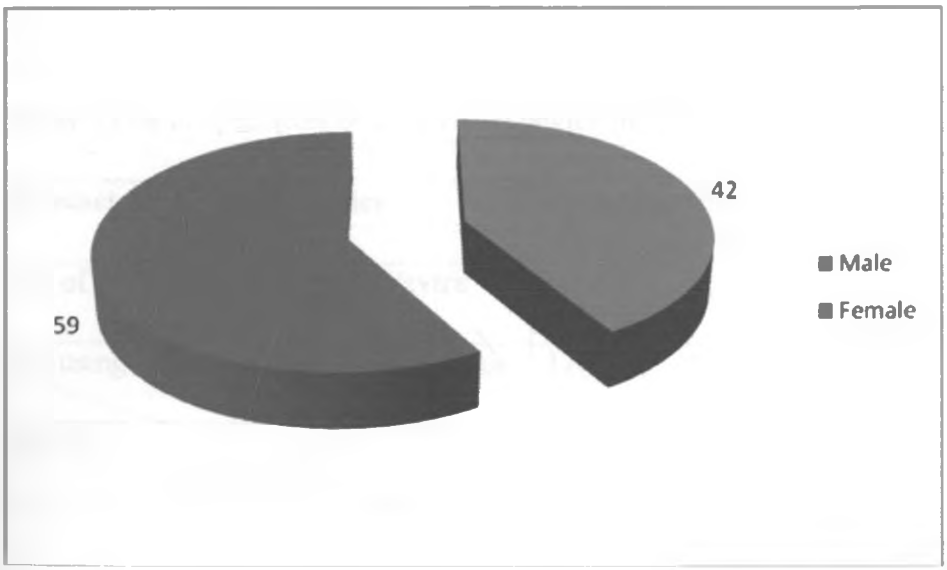
7.0 RESULTS

One hundred and one pupils participated in the study. One pupil declined to give consent.

Table 1. Distribution by school (n=101)

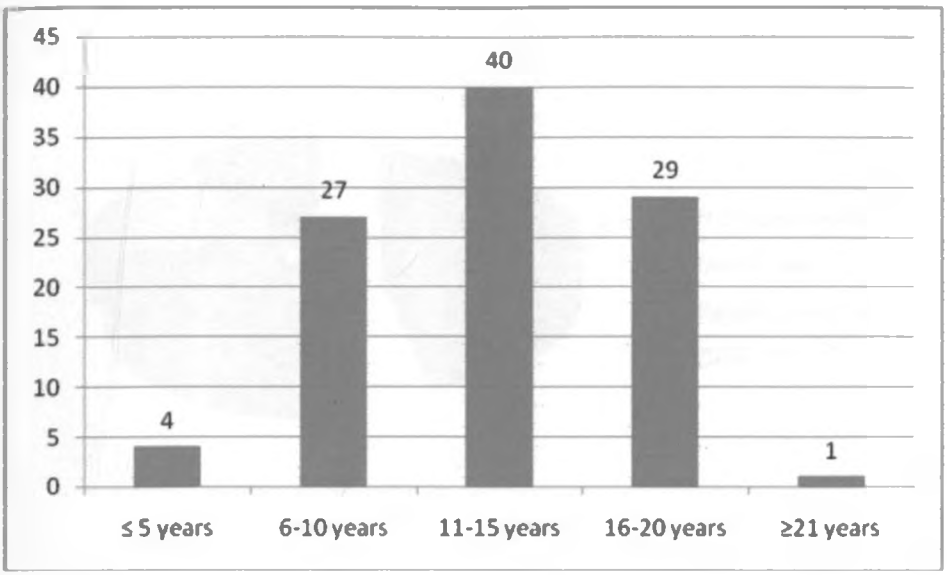
School	Frequency(%)
St Lucy Igoji	45(44.6)
Thika school	33(32.6)
Kitui Integrated Programme	23(22.8)
TOTAL	101(100)

Figure 1. Distribution by Sex (n=101)



The Female to Male ratio was 1.4:1

Figure 2. Distribution by Age



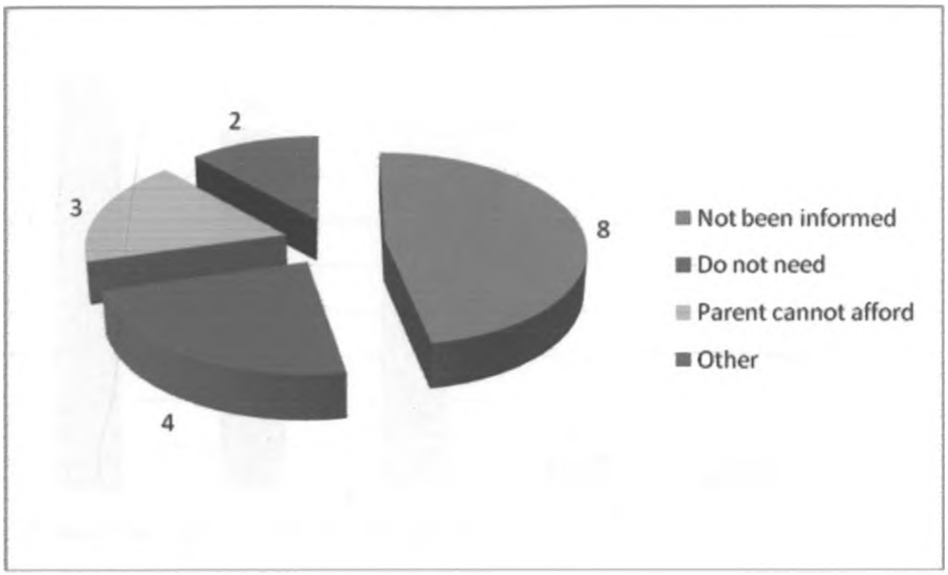
The mean age was 12.59 years ± 4.16 (95%CI 11.77-13.41). The youngest was 4 years and the oldest was 21 years old.

Table 2. Use of spectacles or Low vision device (n=101)

Spectacles/Low vision device	**Frequency(%)**
Use of spectacles/ Low vision device	84(83)
Not using spectacles/ Low vision device	17(17)
TOTAL	101(100)

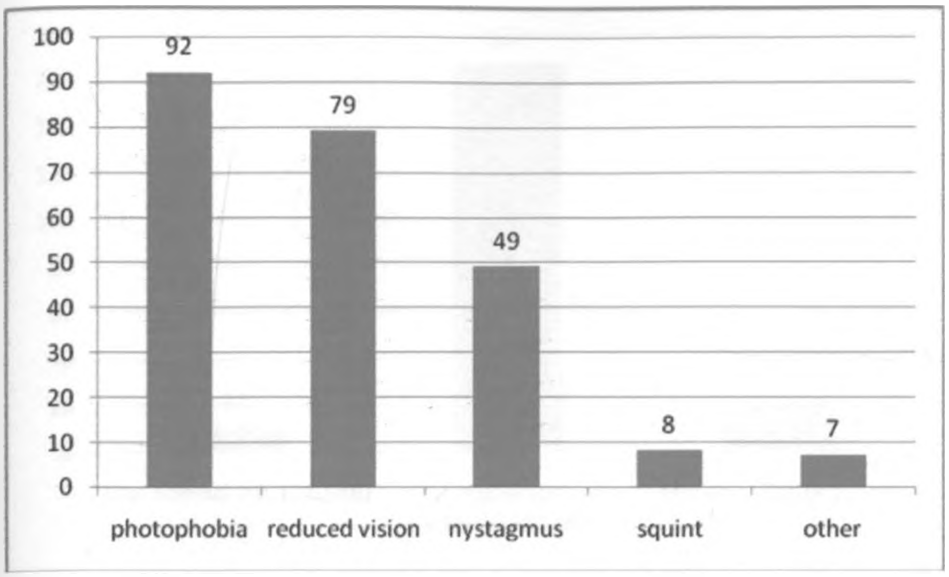
Most of the pupils had photochromic spectacles, prescribed by an ophthalmologist. Two of them had telescopes mounted on spectacles(low vision devices), the rest had photochromic spectacles.

Figure 3. Reason for not using spectacles (n=17)



Common reasons given for not using spectacles were that they did not know whether or not they needed spectacles or a low vision device and had never had a prescription for them; They felt they did not need them; Their parents could not afford the cost of the spectacles or they felt their vision was worse with spectacles.

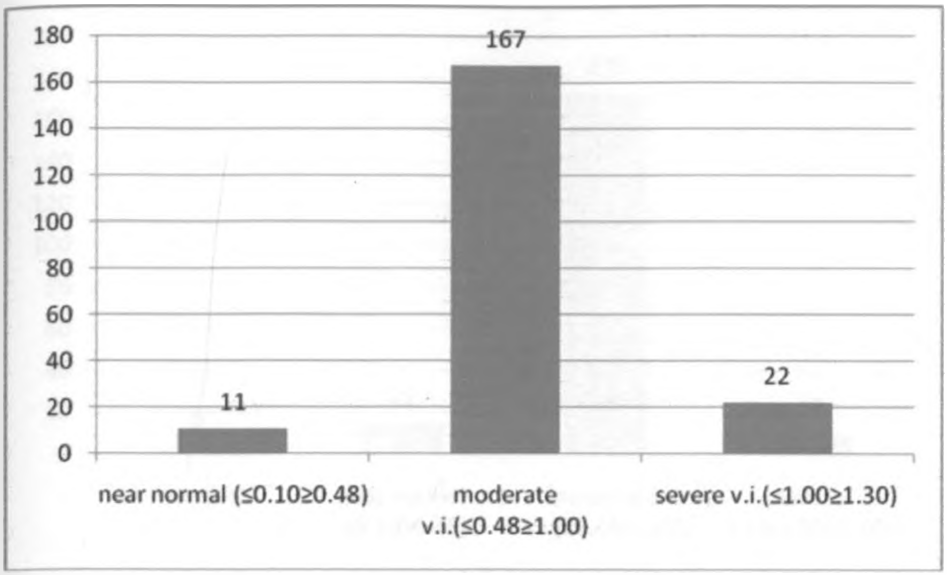
Figure 4. Ocular Symptoms (n=101)



The commonest ocular symptoms were photophobia and reduced vision.

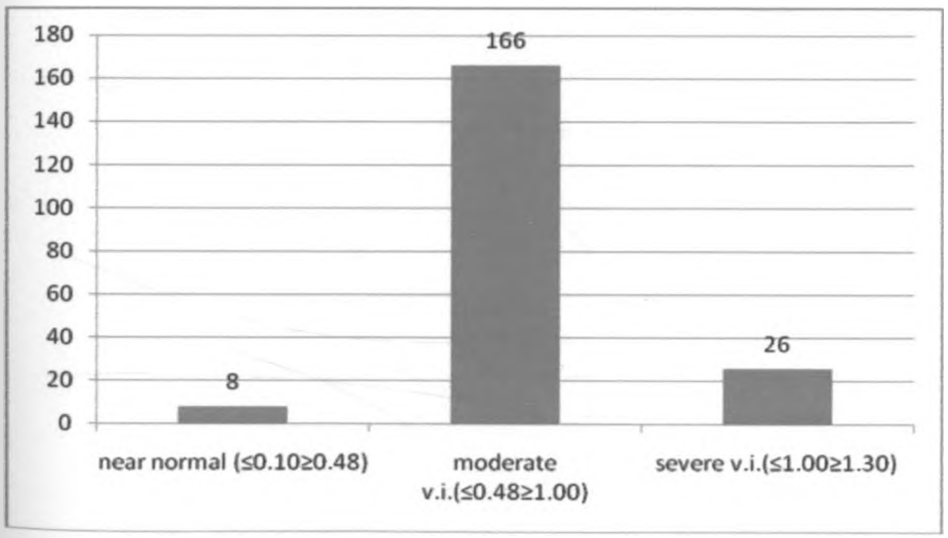
Seven pupils had other symptoms including pain, redness, discharge and itchy eyes.

Figure 5. Visual acuity before correction (n=200eyes)



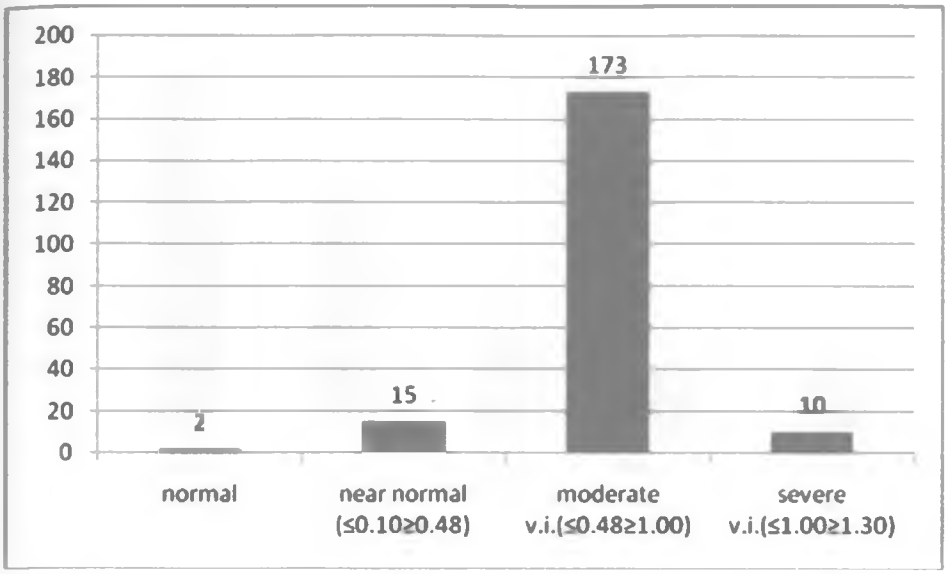
Most of the pupils had moderate visual impairment. Only 22 eyes (11%) had severe visual impairment. One pupil could not respond to subjective testing of visual acuity.

Figure 6. Visual acuity using own correction (n=200 eyes)



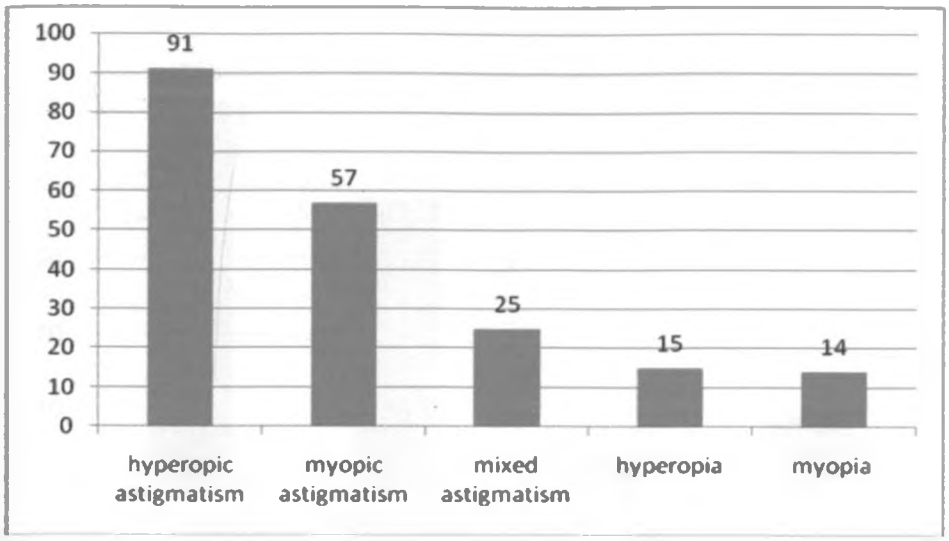
There was some worsening of visual acuity with spectacle correction in some of the eyes. This suggests either a change in the refractive error with time or these pupils had the wrong refractive correction. Information on the current correction was not available.

Figure 7. Visual acuity after best subjective correction (n=200eyes)



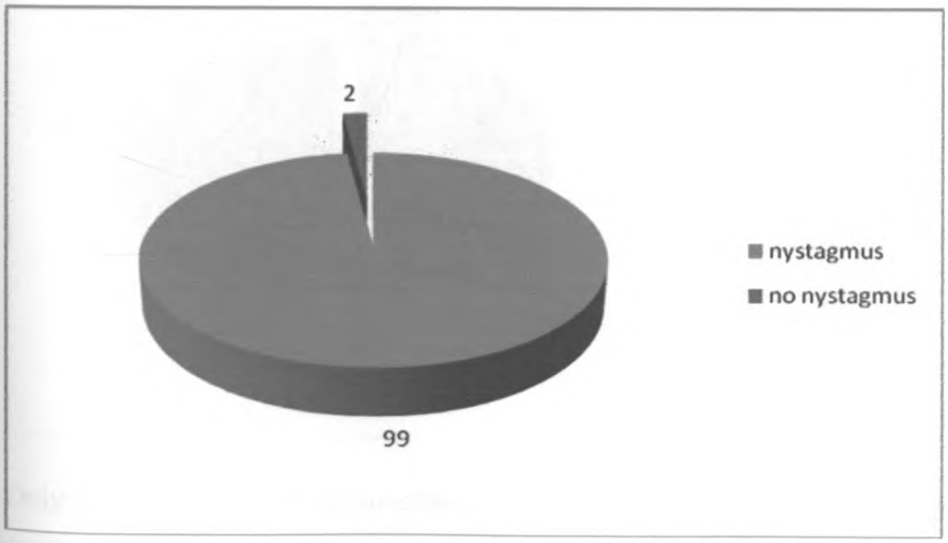
There was some improvement of visual acuity with spectacle correction. However, the visual acuity of most of the eyes still constituted moderate visual impairment.

Figure 8. Pattern of refractive errors (n=202 eyes)



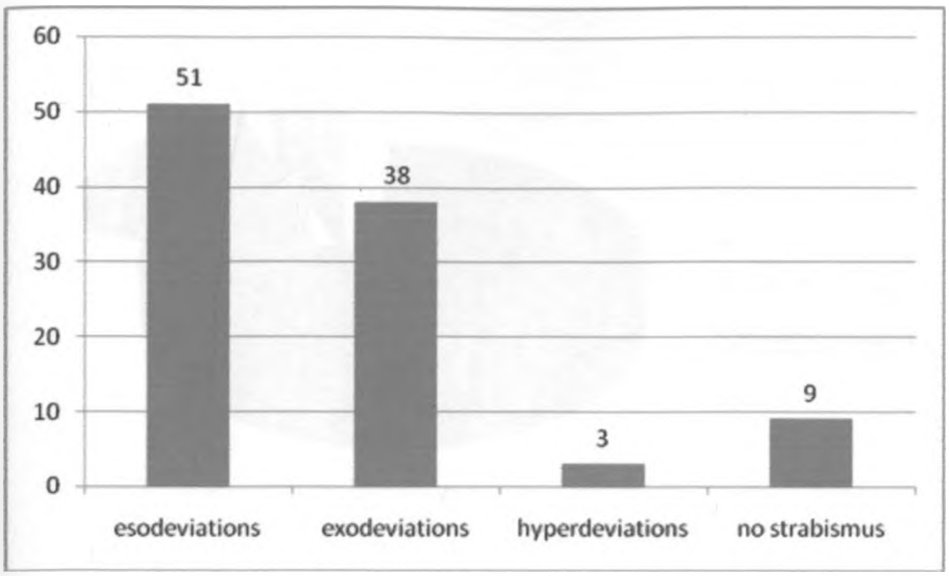
The equivalent spherical refractive error data for the pupils yielded a mean of +0.31D representing low hyperopia. However, the standard deviation describing this data is large, 4.58 D, reflecting the large scatter in the data. Overall, the refractive errors ranged from -16.00D to +10.00 D, with 16 eyes having a spherical equivalent of ≥-6.00D and 12 eyes having a spherical equivalent of ≥+5.00D.

Figure 9. Prevalence of nystagmus (n=101)



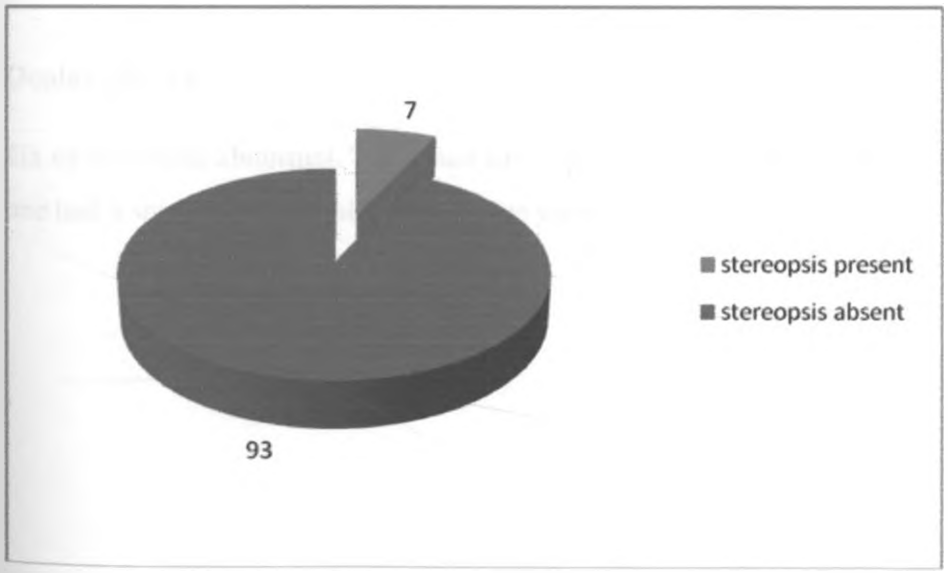
The prevalence of nystagmus was 98%.

Figure 10. Prevalence and pattern of strabismus (n=101)



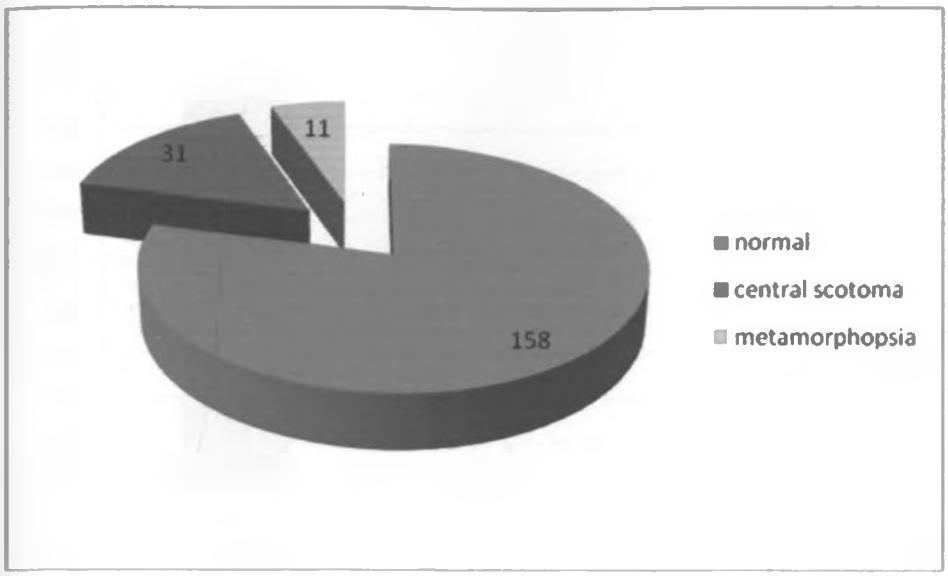
The prevalence of strabismus was 91%, consisting of horizontal deviations. Esodeviations occurred more frequently than any other deviations.

Figure 11. Presence of stereoacuity (n=100)



Only seven of the pupils demonstrated gross stereopsis (Titmus Fly stereotest). One pupil could not respond to subjective tests such as the Titmus Fly Stereotest.

Figure 12. Amsler grid assessment (n=200 eyes)

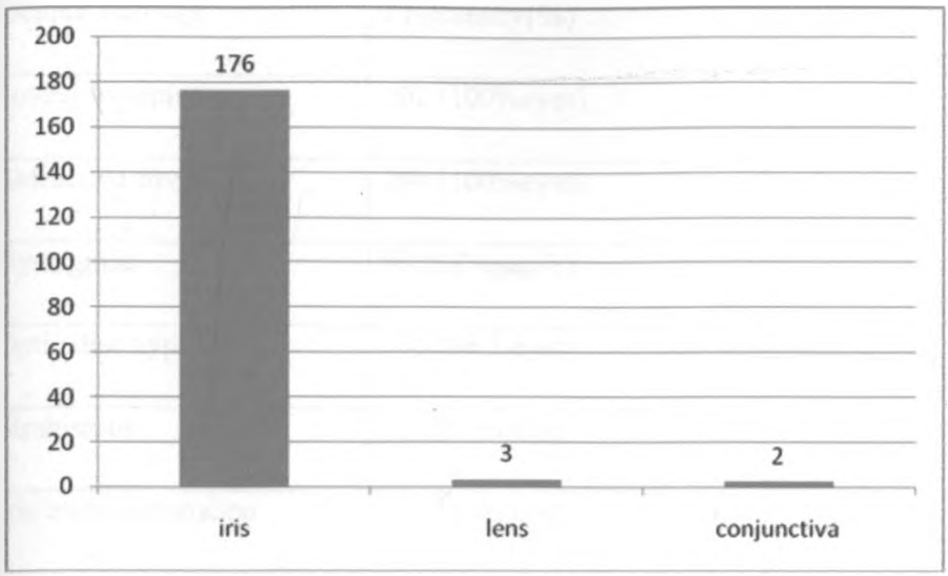


Most of the eyes (79%) had a normal Amsler grid assessment, with no scotomata and no metamorphopsia. One pupil could not respond to subjective tests like the Amsler grid assessment.

Ocular adnexa

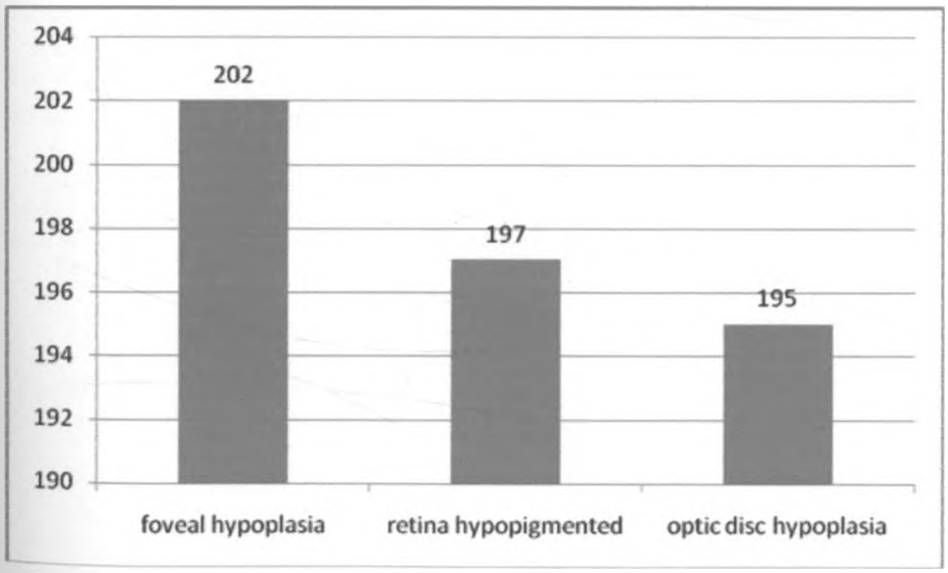
Six eyelids were abnormal. Three had small, pale, raised warty lesions. Two had ectropion and one had a small scar. Overall, the adnexae were normal (97%).

Figure 13.Anterior segment findings (n=202 eyes)



The commonest anterior segment finding was iris transillumination defects. Two eyes had posterior synechiae and one pupil had completely blue irides.

Figure 14: Posterior segment findings (n=202 eyes)



Posterior segment findings were hypopigmentation of the retina (97.5%), foveal hypoplasia (100%) and optic nerve hypoplasia(97%).

Table 3. Frequency of common ocular findings

Ocular findings	**Frequency(%)**
Foveal hypoplasia	202 (100%eyes)
Refractive error	200 (100%eyes)
Nystagmus	99 (98%pupils)
Optic disc hypoplasia	195 (96.5 eyes)
Strabismus	92 (91 pupils)
Iris transillumination	176 (87% eyes)

A total of one hundred and eighty three eyes had reduced vision **(Figure 5)**. This consisted of seventy eight pupils who had reduced vision in both eyes and nine pupils who had unilateral reduced vision.

Table 4. Relationship between the findings and reduced vision

Ocular finding	**Visual impairment (≤0.48 logMAR) in the better eye**
Foveal hypoplasia	100%
Refractive error	94.2%
Nystagmus	98.9%
Optic disc hypoplasia	95.4%
Strabismus	89.7%
Iris transillumination defects	85.2%

All the eyes with reduced vision had foveal hypoplasia, 94.2% had significant refractive errors, 95.4% had optic disc hypoplasia, 89.7% had strabismus and 85.2% had iris transillumination defects. This information suggests a strong relationship between all these ocular findings and reduced vision in persons with albinism. However, these findings were also present in the pupils without visual impairment. Therefore, it was difficult to single out one main cause of reduced vision in these pupils.

8.0 DISCUSSION

This study was done among pupils with albinism attending three schools for the blind in Eastern and Central province. A total of 101 subjects were examined, this is a much larger group than in previous studies.[13, 14, 17, 21,22,23,24]

Visual acuity was measured using Bailey-Lovie logMAR acuity charts, which are appropriate for this age group. This also allows for more accurate analysis of the information obtained. Tumwesigye et al studied the inappropriate enrolment of children in schools for the visually impaired in East Africa.[5] Of all the children they examined, 361 children (34%) had visual acuity equal to or better than 6/60. The children with albinism all had visual impairment (visual acuity <6/18>6/60). In this study, only 5% of the eyes had acuity of <6/60>3/60 (<1.00>1.30 LogMAR) after correction of refractive errors.(**Figure 5**) All but two of the subjects were able to read print, with or without the use of spectacles. Available records showed two pupils used low vision devices. This supports previous data that the number of non-blind children enrolled in special schools and annexes for the blind in East Africa is high.[5, 6, 9] Few records of history of enrolment were available and complete. The reasons for this inappropriate enrolment cannot therefore be fully understood from this study.

A majority of the subjects (83%) use photochromic spectacles, which were prescribed to them by an ophthalmologist. **(Table 2)** Spectacles were prescribed by an ophthalmologist; in our setting it is unlikely for the ophthalmologist to prescribe low vision devices. Records available showed that only two pupils used low vision devices. In the last two years or so there has been increasing advocacy for persons with albinism by the Albinism Foundation of East Africa, with emphasis placed on education of the public, provision of sunscreen lotion, hats and photochromic spectacles to persons with albinism. This data suggests that these efforts are bearing fruit. However if the spectacles get lost or broken, replacement may be difficult due to their relatively high cost.

Photophobia and reduced vision were the commonest complaint from the subjects. **(Figure 4)** Nystagmus was also a common complaint at 48.5%. Most of the subjects did not complain of

squint, only 8% being aware of deviation of their eyes. Seven subjects had other complaints including pain, redness, discharge and itchy eyes.

Photophobia occurs due to the lack of uveal and retinal pigment which results in high levels of retinal irradiance. As mentioned previously, photochromic spectacles were found to be available to most of the subjects we examined. Reduction of the photophobia by reducing the amount of incident light entering the eye is a very important factor in the management of albinism. As well as increasing visual comfort, the alleviation of photophobia in albinism is beneficial as it can result in a decrease in the nystagmus intensity and hence an improvement in visual performance.[12]

A large proportion of the eyes had moderate visual impairment both before (83.5%) and after (86.5%) spectacle correction. **(Figures 5,7 Appendix 1)** Only 5% of the eyes showed severe visual impairment (>1.00<1.30 logMAR; >6/60<3/60) and none showed profound visual impairment or blindness. Past reports on the visual acuity profiles in albinism show reduced visual acuity.[12. 13. 14]

Previously documented causes for the reduced vision in persons with albinism are high refractive errors, intraocular light scatter and due to nystagmus and foveal hypoplasia, the retinal image is constantly in motion over a retina without a fully differentiated fovea.[10]

None of the subjects were emmetropic, and refractive errors included both high hypermetropia and high myopia. The subjects were generally also highly astigmatic. The definition for significant astigmatism was more than ± 1.5 dioptre D; significant hyperopia was more than +2 D; significant myopia was more than -0.5 D. (see study definitions) The equivalent spherical refractive error data for the albino subjects yielded a mean of +0.31 D, representing low hyperopia. However, the SD describing these data is large, 4.58 D, reflecting the large scatter in the data. Overall, the refractive errors ranged from -16.00D to +10.00 D, with 16 eyes having a spherical equivalent of ≥-6.00D and 12 eyes having a spherical equivalent of ≥+5.00D.

After cycloplegic retinoscopy we found that the commonest refractive error was hypermetropic astigmatism(45.5%) followed by myopic astigmatism(27.5%).**(Figure8)** There have been discrepancies between studies on the overall bias in refractive errors, with both myopia[16,17] and hyperopia being reported.[13,18,19] In these previous studies, the sample population studied has

been smaller. Also, in one of them, different subtypes of albinism were represented.[13] They found no significant differences in the refractive profiles between the ocular and oculocutaneous subtypes. The results of the refractive profiles were similar to those in this study. In this study, according to the characteristic physical appearance and clinical examination, the subjects had oculocutaneous albinism. This suggests that refractive error is independent of the subtype of albinism. However, more information about the specific subtypes of albinism in the subjects is not available from this study.

Nystagmus was present in most of the subjects(98%).**(Figure 9)** According to previous studies, nystagmus has been found to be a universal feature of all forms of albinism.[11,12,21] Often these persons develop an anomalous head posture to damp the nystagmus, referred to as the null point, where visual acuity is usually improved.[14] To manage the nystagmus, prisms may be prescribed (with the bases in the opposite direction to the preferred gaze direction) which will allow maintenance of the eyes in this eccentric gaze direction whilst keeping the head in the primary position, thus improving cosmesis and comfort. Also, several surgical procedures have been performed such as horizontal extraocular muscle surgery[11, 12, 14] in an attempt to transfer the null point to a position closer to primary gaze. Improvement of vision after surgery is often only modest. None of the subjects in this study had undergone any of these procedures.

We found that 92% of the subjects had strabismus. **(Figure 10)** This is comparable with past studies[12, 14] Presence of strabismus can be traced to the misrouting of nerve pathways from the retina to the brain's visual cortex.

Stereopsis measured using the Titmus Fly vectograph revealed that most of the subjects (93%) did not have stereoacuity.**(Figure 11)** As mentioned previously, a hallmark of albinism is excessive decussation of retinostriate projections at the optic chiasm. This misprojection leads to abnormalities in the retinal correspondence and may account for the usual absence of stereovision[14,24]However, it has also been found that persons with albinism who do not demonstrate stereoacuity on clinical tests may have stereoscopic perception that commonly used clinical tests do not detect. Moreover, some of them even use this poor stereopsis in judging the

size of stereoscopically presented images.[35] Therefore, this may lead to an over-estimation in the absence of stereopsis seen in this population.

The Amsler grid is an important tool in the examination of central vision. It has been widely used to test for central or paracentral scotomata and metamorphopsias. The results were mostly normal, with 15.5% found to have central scotomata and 5.5% had metamorphopsias.**(Figure12)** However, the test has been found to have several shortcomings, especially with a negative result. It is impossible to know how good an observer a patient is, defects may hide between the grid lines and quantification may be difficult.[36,37]

The commonest anterior segment finding was iris transillumination defects, at 87% of the subjects.**(Figure 12)**

Posterior segment findings were hypopigmentation of the retina(97.5%), foveal hypoplasia(100%) and optic nerve hypoplasia(97%).**(Figure 14)** These findings are due to the absence of melanin pigment in the retinal pigment epithelium and choroid. Dilated fundoscopy of the subjects typically showed loss of the annular and foveal reflexes in the macula. Choroidal vessels were easily visible in the retinal periphery and were often being seen in the macula. The retinal vessels were disordered and coursed directly through the expected area of the fovea. We also observed small optic discs and in most of the subjects, the normal bright orange- pink appearance was replaced by the appearance of a small orange area surrounded by a hyperpigmented area. In some of the subjects, the hyperpigmented area replaced the disc entirely, with the central retinal vessels emerging from it. The reduction in visual acuity in persons with albinism is most likely due to the abnormalities in the retina.

Interestingly, despite the invariable appearance of foveal hypoplasia in the subjects, visual acuity varied from 0.10 to 1.30 logMAR(6/7.5 to 3/60). This may represent varying grades of macular differentiation.

A strong relationship was found between some ocular findings and reduced vision in the subjects **(Table 4)**. Foveal hypoplasia was found in all the eyes with reduced vision. However, not all the eyes with foveal hypoplasia had reduced vision. Other ocular findings which were frequent in

eyes with reduced vision were nystagmus, optic disc hypoplasia, refractive error, strabismus and the presence of iris trans-illumination defects.

9.0 STUDY LIMITATIONS

Incomplete records at details of enrolment for some of the subjects. This made it difficult to find out details of enrolment for many subjects.

There is no available data on the prevalence or incidence of albinism in Kenya, making the sample size estimation difficult.

Two subjects declined to give consent and were therefore excluded from the study.

10.0 CONCLUSION

1. The most common ocular symptoms in persons with albinism are photophobia (91%) and reduced vision (78%). Common ocular findings were iris transillumination defects, nystagmus, strabismus, refractive errors, absence of stereoacuity, foveal hypoplasia, retinal hypopigmentation and optic nerve hypoplasia.
2. Most of the pupils with reduced vision had foveal hypoplasia, nystagmus, optic disc hypoplasia, refractive error and strabismus. However, these findings were also present in the pupils without visual impairment. Therefore, it was difficult to single out one main cause of reduced vision in these pupils.
3. All the subjects were attending schools for the blind or an integrated program. After subjective correction of refractive errors, most of the subjects examined had moderate (86.5%) and to a lesser extent severe visual impairment (5%), with none of them having severe visual impairment. Only two of these subjects required the use of low vision devices to read print. This supports previous studies that suggest an inappropriate enrolment of children into schools for the blind.
4. There was a large spectrum of refractive errors, ranging from high hypermetropia to high myopia. The most common refractive errors were hypermetropic astigmatism (45.5%) and myopic astigmatism (27.5%)
5. Most of the subjects had photochromic spectacle correction (83%).

11.0 RECOMMENDATIONS

1. Persons with albinism should have an eye examination before admission to schools for the blind. Clear admission policies must be developed, enforced and made known to doctors, teachers and parents of children with visual impairment.

2. Conduct surveys to determine the prevalence of albinism in the country.

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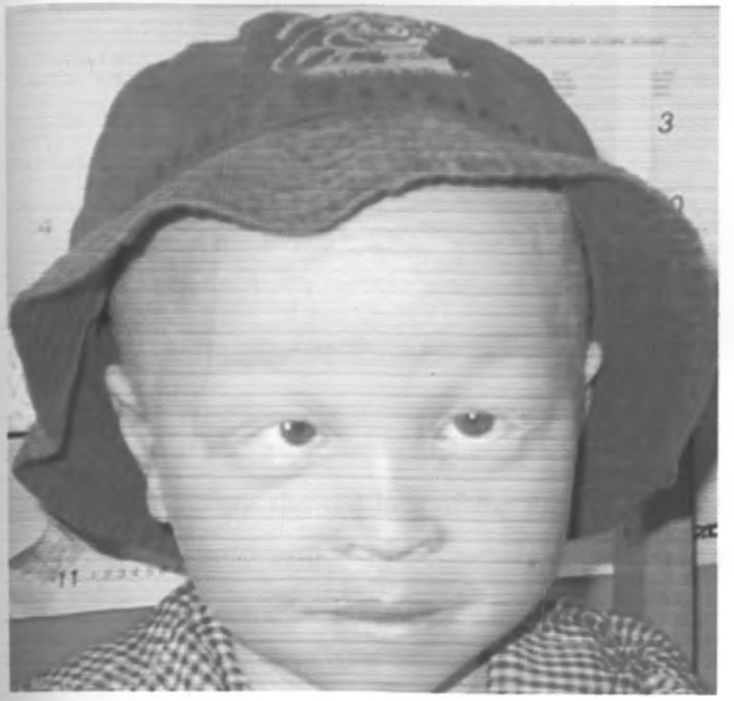
13.0 CASE PRESENTATION



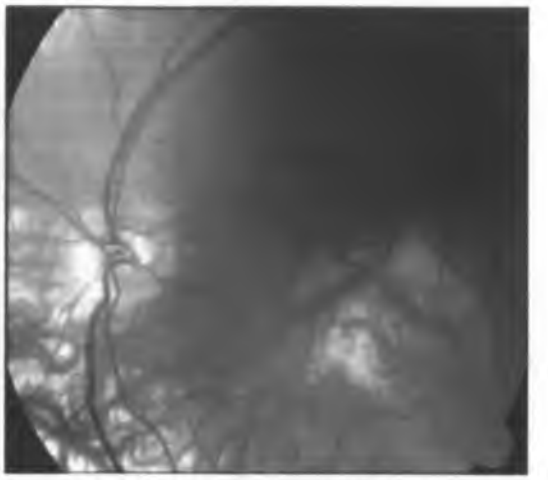
Picture 1

The above pupil is a five year old male child. He was using a pair of photochromic spectacles which he received approximately one year prior to the study period. His ocular symptoms were photophobia and reduced vision. His visual acuity before correction was 1.0 in the right eye and 0.6 in the right eye. After best subjective correction the visual acuity improved to 0.9 in the right eye and 0.4 in the left eye. On orthoptic assessment he had horizontal nystagmus, his right eye had an exotropia of 30° (60Δ) and he had no gross stereopsis (Titmus Fly stereotest). Both his right and left eyes had a normal Amsler grid assessment.

Anterior segment examination showed iris transillumination defects.

Posterior segment findings were hypopigmentation of the retina, optic disc hypoplasia and foveal hypoplasia.

Picture 2



Picture 3

Dilated fundoscopy showed loss of the annular and foveal reflexes in the macula. Choroidal vessels were easily visible in the retinal periphery. The retinal vessels were disordered and coursed directly through the expected area of the fovea. He also had small optic discs and the normal bright orange- pink appearance of the optic disc was absent. The central retinal vessels were seen to emerge from a poorly circumscribed area (**Picture 2**). The optic disc appearance and details of the blood vessels were better evaluated using red-free illumination (**Picture 3**).

14.0 APPENDICES

14.1 Appendix I: WHO Classification of vision and LogMAR equivalent

VISUAL ACUITY	LogMAR EQUIVALENT	CLASS
≥6/7.5	≥0.10	0 - Normal
≤6/7.5- 6/18	≥0.10-0.48	0 - Near normal vision
≤6/18-6/60	≤0.48-1.00	1 - Moderate visual impairment
≤6/60-3/60	≤1.00-1.30	2 - Severe visual impairment
BLINDNESS		
3/60- 1/60	1.30-1.80	3 - Profound visual impairment
1/60- PL	1.80-PL	4 - Near total visual impairment
NLP		5 - No light perception

14.2 Appendix II: Consent Form

I, Dr A.K. MOKAYA of the Department of Ophthalmology, University of Nairobi, am conducting a study to establish the eye problems in persons with albinism. The information obtained from the study will help to improve the care given to persons with albinism.

I will conduct a complete eye examination on the participant. Information from the participant, guardian or school principal will be entered in a questionnaire. I will enquire about any eye complaints and whether the participant uses spectacles or a low vision device. Measurement of vision will be performed. Tests of central vision will also be done (Amsler-Grid testing).

The participant will then have their eyes examined. Dilating drops (Tropicamide / Mydriacyl) and cycloplegic drops (Cyclopentolate) will be used to examine the back of the eye (posterior segment examination). It may cause mild irritation, transient blurring of vision and glare lasting between six and eight hours. You are advised to take extra care of yourself / the child and guide him / her until he /she regains usual vision.

Participation in the study is voluntary. If you wish to withdraw at any point you may do so. Treatment and care of yourself or your patient will not be altered in any way.

If you wish to take part in this study, please acknowledge;

I .... of (school) ..... hereby freely consent on behalf of *myself / the participant _____ to participate in the above study. I acknowledge that the procedure and the side effects of dilating and cycloplegic drops have been explained to me thoroughly by Dr_____. I further state that the procedure that is to be done has been explained to me in a language I understand well and I have agreed for the examination to be done on *myself / the participant. I understand that the participation is voluntary.

* Patient's / Parent's / guardian's signature.....Date.....

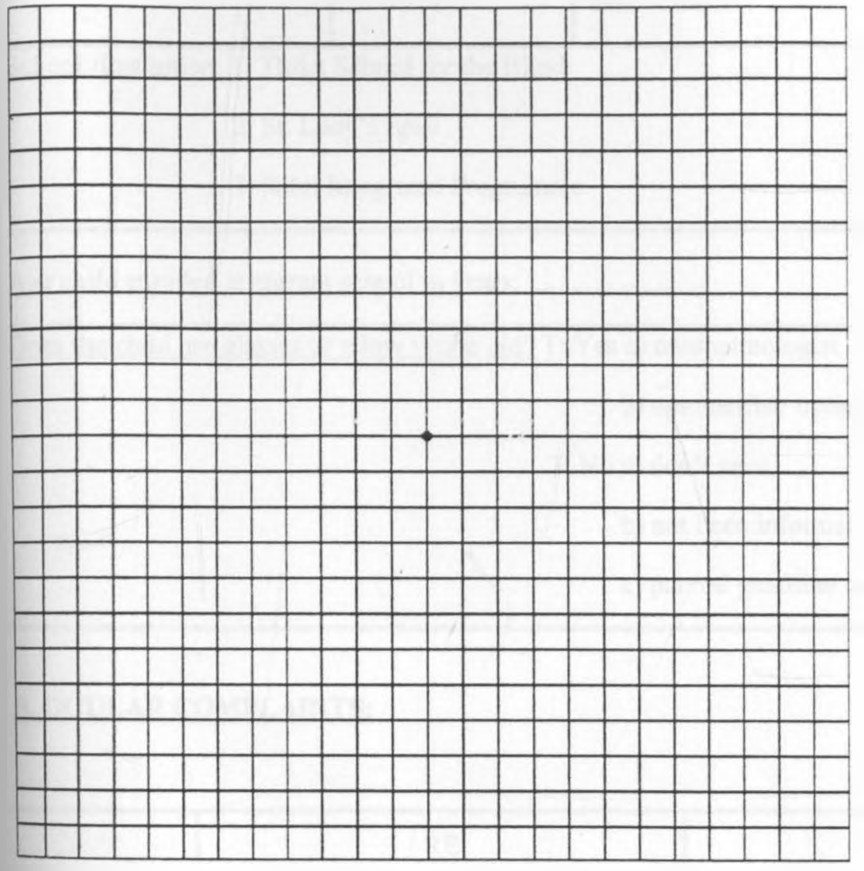
Doctor's signature..... Date.....

(Delete where not applicable)*

Contact person:

1. Researcher's contact 0722455576
2. Dr. D. Ilako, Dept. of Ophthalmology, University of Nairobi.

14.3 Appendix III:Amsler grid chart



14.4 Appendix IV:Questionnaire for pupil

A. DEMOGRAPHIC DATA

Identification no:	Sex: 1: M 2: F	Age in years: date of birth:

School /Institution: 1: Thika School for the Blind
2: St. Lucy's Igoji
3: Kitui Integrated Programme

Age child enrolled at current school in years: .....

Does the child use glasses or a low vision aid? 1: Yes a) ophthalmologist
b) optometrist/ optician
2: No a) don't need
b) not been informed
c) parent/ guardian can't afford

B. OCULAR COMPLAINTS:

	RE		LE	
	YES	NO	YES	NO
Photophobia				
Reduced vision				
Nystagmus				
Squint				
Other				

C. OCULAR FINDINGS		
	RE	LE
VA (sc)		
VA (cc / PH)		
Refraction:		

	YES	NO	YES	NO
Nystagmus:				
Strabismus:				

Stereoacuity, Titmus Fly test 1: Present

2: Absent

Amsler Grid test:	RE		LE	
	YES	NO	YES	NO
Central scotoma				
Metamorphopsia				

Anterior segment examination		
	RE	LE
Lids:	1: Normal 2: Abnormal- a) mass b) scar c) unidentified	1: Normal 2: Abnormal- a) mass b) scar c) unidentified
Conjunctiva	1: Normal 2: Abnormal- a) discharge b) injection c) mass	1: Normal 2: Abnormal- a) discharge b) injection c) mass
Cornea:	1: Normal 2: Abnormal a) opacity	1: Normal 2: Abnormal a) opacity
Anterior chamber	1: Normal 2: Abnormal a) Hyphema b) Hypopyon	1: Normal 2: Abnormal a) Hyphema b) Hypopyon
Iris:	1: Normal 2: Abnormal a) transillumination defect b) posterior synechiae c) aniridia	1: Normal 2: Abnormal a) transillumination defect b) posterior synechiae c) aniridia

Pupil:	1: Normal 2: Abnormal a) RAPD b) fixed. NRTL	1: Normal 2: Abnormal a) RAPD b) fixed. NRTL
Lens:	1: Normal 2: Abnormal a) cataract b) pseudophakic c) aphakic	1: Normal 2: Abnormal a) cataract b) pseudophakic c) aphakic

Posterior segment examination		
	RE	LE
Vitreous:	1: Normal 2: Abnormal a) vitritis b) hemorrhage	1: Normal 2: Abnormal a) vitritis b) hemorrhage
Retina:	1: Normal 2: Abnormal a) hypopigmented b) other	1: Normal 2: Abnormal a) hypopigmented b) other .....
Fovea:	1: Normal 2: Hypoplasia	1: Normal 2: Hypoplasia
Optic disc:	1: Normal 2: Abnormal a) atrophic b) other	1: Normal 2: Abnormal a) atrophic b) other

14.5 Appendix V: Questionnaire for teacher

A. DEMOGRAPHIC DATA

Identification no:	Sex: 1: M 2: F	Position: 1: class teacher 2: administrator 3: head teacher

School /Institution: 1: Thika School for the Blind
2: St. Lucy's Igoji
3: Kitui Integrated Programme

Duration of work at current school in years: .....

B. INFORMATION REGARDING CHILD

Age child enrolled at current school in years: .....

C. OCULAR COMPLAINTS:

	RE	LE
	YES	NO
Photophobia		
Reduced vision		
Nystagmus		
Squint		
Other		