# Visual Rehabilitation of Patients with Stargardt's Disease

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## ABSTRACT

**Objective:** To assess the role of low vision devices in visual rehabilitation of patients with Stargardt's disease. **Study Design**: Descriptive study.

**Place and Duration of Study**: The Khyber Institute of Ophthalmic Medical Sciences, Hayatabad Medical Complex, Peshawar, Pakistan, from June 2003 to June 2005.

**Patients and Methods**: Patients diagnosed as having Stargardt's disease and confirmed by two senior ophthalmologists clinically and after fluorescein fundus angiography and Electro-Retinogram (ERG) when needed, were included in the study. Patients with fundus pathologies other than Stargardt's disease were excluded. Each subject underwent an ophthalmic examination. Visual acuity was tested with a logarithm of the minimum angle of resolution chart; Feinbloom chart and Snellen type. Low vision assessment was performed on all individuals. SPSS version 10 was used for analysis of the data regarding the severity of visual impairment, visual acuity and devices used for aiding vision. Chi-square test was used for comparison of proportions.

**Results**: Of this cohort of 64 patients, 72% were aged between 7 to 15 years and 28% were 16 to 32 years. Using WHO low vision criteria, the percentage of visually impaired, severe visually impaired and blind (at the time of presentation) were 56.3%; 31.3% and 9.4% respectively. Among those patients, 3.1% had distance visual acuity of 6/18 or better in the better eye and 53% had normal near visual acuity of 1M (0.8 print size). Telescopes were prescribed to 53% patients for enhancement of distance visual acuity to meet their needs.

**Conclusion**: Stargardt's patients respond well to magnification. Simple bifocal glasses may be used in the early stages. Visual rehabilitation can help Stargardt's patients to learn independence in their activities of daily living.

Key words: Stargardt's disease. Blindness. Low vision. Visual aid. Rehabilitation. Magnification. Bifocal lens.

## **INTRODUCTION**

According to the Pakistan National Blindness and Visual Impairment Survey, the prevalence of blindness (visual acuity less than 3/60 in the better eye on presentation) was 3.4% while the prevalence of severe visual impairment and blindness (presenting visual acuity <6/60 in the better eye) was 4.9%.<sup>1</sup> The prevalence of blindness after correction was 2.7% and the prevalence of severe visual impairment and blindness after correction was 3.3%.<sup>1</sup> The estimated all-age prevalence of blindness is 1.8%.<sup>2</sup> The main cause of functional low vision in NWFP and Balochistan is retinal disease.<sup>3</sup>

The nomenclature and definition used for different level of visual loss are confusing. The WHO describes a person with impairment of visual functioning even after treatment and/or standard refractive correction and having visual acuity less than 6/18 to light perception, or

a visual field of less than 10° from the point of fixation, but who uses, or is potentially able to use, vision for the planning and/or execution of a task.<sup>4</sup>

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Visual rehabilitation is a multifaceted process involving the assessment of visual capabilities and the evaluation of functional performance (e.g., reading, writing, and mobility) within the context of lifestyle (e.g., employment. family activities), attitudes, and psychological well-being. Rehabilitation goals are defined in terms of what matters most in a person's life, and attempts are made to solve functional problems through adaptive options (e.g., vision enhancement and substitution devices, environmental modifications) and coping strategies. Stargardt's disease is a form of macular dystrophy that begins early in life.

The condition was first described in 1909 and remains one of the most common forms of juvenile macular degeneration. In a previous study Stargardt's disease contributed to 13.94% of the causes of low vision in age under 16 years.<sup>5</sup> Stargardt's disease may occur in one of every 20,000 children over the age of 6 and is usually diagnosed before the age of 20. Boys and girls are equally affected by this condition. It is characterized by progressive loss in central vision and bilateral atrophicappearing macular changes surrounded by yellowishwhite fundus lesions at the level of the Retinal Pigment Epithelium (RPE) and degeneration of photoreceptors. Difficulty occurs in recognising faces, reading, writing and other close work, such that the affected can see objects "out of the corner of his/her eye" only (peripheral or 'side' vision). However, this vision is useful for moving about with premises.

Children may be misdiagnosed for a psychological vision loss since macula appears normal initially. In time, characteristic changes occur in the retina that help facilitate diagnosis. During angiography, the damage to the retina blocks the flow of light from the choroid causing a "dark choroid" (blood vessel layer behind the retinal due to deposition of lipofuscin that causes degeneration of rods and cones). This test alone is not considered to be completely diagnostic of the disease. Several electrophysiologic tests, such as an Electro-Retinogram (ERG), Electro-oculogram (EOG), and darkadaptation testing can be used in the diagnosis and follow-up of people with Stargardt's macular dystrophy. However, these tests are not always used to make the diagnosis, particularly when the yellow-white fundus flecks are apparent on examination.

Patients with Stargardt's disease usually presents by teenage.<sup>6-9</sup> The changes within the retina, as well as in the area of central vision, tend to be extensive and worsen over time. When a Stargardt's patient is exposed to bright sunlight, the retina may become bleached by the light, the sharpness of vision may decrease and blind spots may become denser. These are temporary conditions, but can be prevented or lessened by use of sun filters and hats. The good in Stargardt's is that it never causes total vision loss. Peripheral vision is left intact.

The low vision examination is an in-depth evaluation of the person's functional use of the remaining vision. A comprehensive, interdisciplinary approach to vision rehabilitation can help the vast majority of partiallysighted people learn to use their remaining visual capacity to its fullest and to be as independent as possible. This includes those people who have corrected visual acuity in the better eye of 6/18 or less, and/or a field of vision of 20 degrees or less.

The purpose of this study was to determine the role of Low Vision Devices (LVDs) including optical and nonoptical aids to maximize use of the residual vision in patients with Stargardt's disease.

### PATIENTS AND METHODS

This descriptive study was carried out in the low vision clinic at Khyber Institute of Ophthalmic Medical Sciences (KIOMS), Hayatabad Medical Complex, Peshawar, Pakistan. Patients having Stargardt's disease were included in the study. Record of 64 individuals with Stargardt's disease who attended the low vision clinic from June 2003 to June 2005 was analyzed.

Each subject underwent ophthalmic examination that included personal medical history; family history, functional and occupational assessment, determination of best-corrected Visual Acuity (VA); slit-lamp examination of the anterior segment and vitreous; determination of intraocular pressure; and fundus examination. Fluorescein angiography and electroretinogram (ERG) was performed when needed. Diagnosis was reconfirmed by a senior ophthalmologist.

Patient's presenting distance visual acuity was measured with a logarithm of the minimum angle of resolution chart at 4m and, if necessary, at 3, 2m on each eye separately while the patient wore current spectacles (if any). Visual acuity was recorded as the total number of characters read correctly. The logarithm of the minimum angle of resolution chart measures a range of Snellen equivalent visual acuity from at best 6/3 at 4m to at worst 1/60 at 1m.<sup>10</sup> Feinbloom chart for the partially sighted and Illiterate E were used for patient who could not read English depending on the level of cooperation. If visual acuity could not be measured with these charts, a sequential approach was used with the following tests: counting fingers, hand movement, and light perception. For near visual acuity "Near Reading Card for the partially sighted" by William Feinbloom and Lea Cards for near visual acuity were used.

The print size used in books for students are not standardized. For the purpose of this study, near acuity was banded in three groups; 1M (newspaper size) or better which would allow access to most printed materials, <1M to 3.2M (display materials) which would allow only limited access to ink print; and < 3.2M.<sup>5</sup>

Retinoscopy was performed on all patients, which was followed by subjective refraction using standard techniques. The best corrected distance and near acuity, the refractive error and eye to chart distance were recorded. Optical aids varying in type from simple magnifiers to telescopes for seeing at distance; microscopes mounted in glasses for reading; and even closed circuit televisions that can magnify print upto 100 times, enabling the person to read again, were used during the low vision assessment.

All the variables were evaluated on SPSS version 10 and analyzed in terms of frequencies and chi-square statistics for comparing proportions.

## RESULTS

Fifty (78%) were males and 14 (22%) were females. Sixty four patients were classified into two arbitrary age groups, 7 to15 years and 16 to 32 years. Mean age was 14 years. 3.1% (2 patients) were having visual acuity 6/18 or better in the better eye. Thirty six (56.3%) patients were visually impaired (having visual acuity < 6/18 to 6/60 in the better eye). 31.3% (20 patients) had severe visual impairment (with visual acuity < 6/60 to 3/60 in the better eye) and 9.4% (6 patients) were blind (having VA < 3/60 in the better eye) according to the WHO categories of visual loss (Table I). Among these patients 60% (38) had positive family history for Stargardt's disease and 40% (26) had no family history for Stargardt's disease. Thirty six (56%) had positive

**Table I**: Distribution of presenting distance VA (visual acuity), VA diasses and VA with LVDs (low vision devices) (n = 64).

with glasses			(11 - 07).
VA	Presenting VA	VA with glasses	VA with LVDs
	n (%)	n (%)	n (%)
6/18 or better	2 (3.1)	2 (3.1)	60 (93.8)
< 6/18-to-6/60	36 (56.3)	50 (78.1)	0 (0)
<6/60-to-3/60	20 (31.3)	8 (12.5)	0 (0)
< 3/60	6 (9.4)	4 (6.3)	4(6.2)
Total	64 (100)	64 (100)	64 (100)

consanguinity and 44% negative. Forty eyes had no refractive error, 28 eyes were hypermetropic and 60 eyes were myopic. With refraction, the number of patients having visual acuity 6/18 or better in the better eye was the same while the number of visual impaired increased to 78.15% (50 patients) by decreasing the number of severe visually impaired and blind to 12.5% (8 patients) and 6.3% (4 patients) respectively. With low vision devices, in 93.8% (60 patients), distance visual acuity was enhanced to 6/18 or better in the better eye while 6.2% (4 patients) remained in the blind category (p=0.006).

In males (64%) of the population with Stargardt's disease had visual impairment while severe visual impairment and blindness was greater in females contributing to 57.1% and 14.3% respectively (p=0.06). Blindness increased with age from 8% at age 7 to 15 years to 11.1% at age greater than 15 to 32 years (Table II, p=0.006). With LVDs, 96% males and 86% females achieved visual acuity of 6/18 or better (p=<0.05). Fourteen percent females and 4% males remained in the blind category (Table III). Vision enhancement was better in age group 7 to 15 years (95.7%) compared to age group from 16 to 32 years (89%, p=0.006). Vision could not be enhanced in 4.3% of the age group 7 to 15 years and 11% of the age group from 16 to 32 years (Table III).

 Table II: Distribution of visual impairment by gender and age at the time of presentation (n=64).

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Gender		Age		ge		
VA	Male	Female	p-value	7-5 years	15-32 years	p-value
	n (%)	n (%)		n (%)	n (%)	
6/18 or better	2 (4)	0 (0)	0.06	2 (4.3)	0 (0)	0.006
< 6/18-6/60	32 (64)	4 (28.6)	0.06	28 (60.9)	8 (44.4)	0.006
< 6/60-3/60	12 (24)	8 (57.1)	0.06	12 (26.1)	8 (44.4)	0.006
< 3/60	4 (8)	2 (14.3)	0.06	4 (8.7)	2 (11.1)	0.006
Total	50 (100)	14 (100)		46 (100)	18 (100)	

Table III: Distribution of visual impairment by gender and age with LV.

	Gen	der		Age		
VA	Male	Female	p-value	7-5 years	15-32 years	p-value
	n (%)	n (%)		n (%)	n (%)	
6/18 or better	48 (96)	12 (86)	0.05	44 (95.7)	16 (89)	0.006
< 6/18-6/60	0 (0)	0 (0)	0.05	0 (0)	0 (0)	0.006
< 6/60-3/60	0 (0)	0 (0)	0.05	0 (0)	0 (0)	0.006
< 3/60	2 (4)	2 (14)	0.05	2 (4.3)	2 (11)	0.006
Total	50 (100)	14 (100)		46 (100)	18 (100)	

The results from near visual acuity measurements are shown in Table IV. Thirty four patients with low vision were able to discern 1M (newspaper print) or better in the better eye without near addition. Twenty had near visual acuity < 1M to 3.2M and 10 had < 3.2M (p=<0.05). With low vision aids, 93.8% (60 patients) achieved 1M (0.8 print size) or better, and 6.2% (4 patients) had < 3.2M (p=<0.05). Spectacles were prescribed to 30 patients and telescopes were prescribed to 15 patients. Spectacles and telescope both were prescribed to 17 patients and 16 patients were feeling better with filters. Spectacles were the most common devices prescribed for patients with Stargardt's disease.

**Table IV**: Near visual acuity at presentation and with low vision aids (n =64).

(11 -	-0+).		
VA	Presenting near AV	Near VA with LVDs	p-value
	n (%)	n (%)	
1M or Better	34 (53.1)	60 (93.8)	0.05
<1M-3.2M	20 (31.3)	0 (0)	0.05
<3.2M	10 (15.6)	4 (6.2)	0.05
Total	64 (100)	64 (100)	

#### DISCUSSION

People with Stargardt's disease usually have some loss of central vision by the age of twenty years. Sometimes, however, vision loss may not be noticed until the affected person reaches early thirties or forties. The National Blindness and Visual Impairment Survey is the largest and most comprehensive population-based eye survey to be conducted in Pakistan. The prevalence of blindness found in adults and in all age groups was 2.7% and 0.8% respectively.<sup>1</sup>

No study has been conducted in the region on rehabilitation of patients with Stargardt's disease. In this study more than half (60.9%) of patients with Stargardt's disease had vision impairment (6/60<VA<6/18 in the better eye) in age group 7 to 15 years and 26.1% had severe vision impairment while in age group 16 to 32 years, the number of visual impairment as well as severe visual impairment was 44.4%. The number of profound vision impairment/blind was 11.1% in older group as compared to 8.7% in the younger. An earlier study had shown that Stargardt's disease contribute to 13.94% of the causes of low vision in age less than 16 years.<sup>5</sup> In the developed countries, the primary cause of blindness is age-related macular degeneration. Retinal diseases are more common in Balochistan and NWFP<sup>3</sup> and Stargardt's disease is the second leading cause of low vision after nystagmus in children under 16 years of age in NWFP.5

The condition is usually transmitted as an autosomal recessive trait. Recent studies have shown that Stargardt's disease is caused by sequence variations in the gene encoding ABCA4, a retina-specific protein expressed in rod and cone photoreceptors.<sup>11,12</sup> The

ABCA4 gene product is an active transporter that in rods facilitates the movement of all-trans retinal, the retinoid product of rhodopsin bleaching, from the lumen of the rod outer segment disc to the rod cytosol.<sup>13-17</sup> This translocation and the subsequent enzymatic reduction of all-trans retinal terminate its ability to combine with opsin, and thus promote the shut-off of excitation in the phototransduction cascade.<sup>18,19</sup> In this study, 60% have positive family history for Stargardt's disease. This greater number is probably due to inter family marriages as 56% had positive consanguinity.

This study showed that in females, 57.1% had severe visual impairment and 14.3% were blind while in males, 24% had severe visual impairment and 8% were blind. Hence, the percentage of severe visually impaired and blind was greater in females compared to males. This finding shows that females have significantly higher odds of having severe visual impairment and blindness which may be reflective of their relatively disadvantaged social status. It is interesting to note a trend towards higher prevalence of moderate visual impairment in females caused by refractive error, especially hyperopia, and cataract.<sup>20</sup>

In this study it was found that more than half of this population with Stargardt's disease had normal near visual acuity. Spectacles including prismatic glasses meet the need of the majority of patients with Stargardt's disease for reading. Spectacles are the most commonly used form of refractive correction since they are the most inexpensive and the simplest treatment for refractive errors in the developing countries.<sup>21</sup> Most visually impaired people have useful sight but there is underprovision of visual aids, inadequate training in their uses, and poor understanding of simple methods of visual function for daily living.

In this study, 31.3% had Visual Acuity (VA) < 6/60 to 3/60 and 9.4% had VA less than 3/60 in the better eye. Therefore, about 40% were blind by visual acuity criteria. Regarding blindness, there were 6 patients who had VA <3/60. Four of those 6 patients were 7 to 15 years of age and 2 patients were between 16 to 32 years of age.

Low vision services improve quality of life and mental state.<sup>22-24</sup> Clinical trials providing evidence of the effectiveness of specific interventions for individuals with functional low vision are lacking.<sup>25</sup> Further research is recommended to compare different types of low vision devices according to the underlying cause and need of the individual. Provision of low vision services was a neglected area in the past but recent governmental policies have prioritized the provision of low vision services and their integration into national eye care programme in the second 5-year plan vision 2020. A recent survey conducted throughout India showed that only 48 (6.8%) of 701 eyecare institutions had a dedicated low vision services.<sup>26</sup> Such services are

similarly lacking in Pakistan as well.

Low vision care and rehabilitation can help Stargardt's patients lead normal lives. Following diagnosis, every Stargardt's disease patient should have a low vision examination by an eyecare worker skilled in low vision rehabilitation. Keeping in view the wide variety in types and the extent of vision impairments, it is necessary to perform a low vision examination to analyze where the best residual vision is and what is the specific type of optical aid that will best help the person meet their visual needs.

This study suggests that overall, patients with Stargardt's macular dystrophy can contribute and function well in contemporary society; however, there is a need for improving diagnosis, referring patients to appropriate services and increase awareness of the specific needs for this group of patients.

#### CONCLUSION

Stargardt's patients respond well to magnification. Simple bifocal aids may be used in the early stages. Visual rehabilitation can help Stargardt's patients to be indendepent about activities of daily chores.

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