Original Research Paper

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Ophthalomology

# CASE SERIES OF STARGARDT'S DISEASE

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**ABSTRACT** **Background:** Stargardt's disease is a macular dystrophy and most common cause of decreased central vision in adults less than 50 years. It is caused due to mutations in ABCA4 gene, located on chromosome 1, which encode ATP-binding cassette (ABC) Protein transporter expressed by rod outer segment. Characterized by central macular atrophy and yellow-white flecks at the posterior pole, primarily at the level of the RPE. **Materials And Methods:** Series of 10 cases studied over a period of 6 months in tertiary health center **Results:** Total of 10 patients were included in this case series. out of 10 patients 6 are males and 4 are females. 7 patients are below 20 years and 3 patients are above 35 years of age. 7 patients have visual acuity of 6/60. 2 patients have 6/36. 1 patient have 6/18. All patients are showing colour vision disturbances. Fundus examination of all patients show beaten bronze appearance. **Conclusion:** Stargardt disease, which is highly heterogeneous both phenotypically and genetically, is one of the most common forms of inherited vision impairment in children and adults. Significant progress has been made in diagnosing the disease at an earlier stage, identifying clinical characteristics that allow for more accurate prognosis information, performing accurate and rapid molecular genetic testing, and understanding the underlying disease mechanisms.

**KEYWORDS :** Stargardt's disease, macular dystrophy, lipofuscin

## INTRODUCTION:

Stargardt's disease is the most common inherited macular dystrophy, and is associated with a variable phenotype and disease severity. Inherited retinal degenerative diseases are a clinically and genetically heterogenous group of disorders that constitute a major cause of vision loss in the world population.

They are typically characterized by the progressive loss of rod and cone photoreceptor cells often leading to severe blindness. Stargardt's disease is known to be one of the most common cause of inherited macular dystrophy in both adults and children. It has typical onset during the early part of second decade of life and presents initially with central vision loss, yellow flecks around macula and retinal mid-periphery, and progressive atrophy of retinal pigment epithelium. ABCA4 is an ATP-binding cassette transporter found in the rim of outer segments of rods and cones. Autosomal recessive stargardt's disease, the most common macular dystrophy, is caused by mutations in the encoding ABCA4, a photoreceptor ATP binding cassette (ABC) transporter. The clinical Diagnosis of Stargardt disease is "dark choroid" or silent choroid on fluoroscein angiography.[5,6]

### Aims And Objectives:

To assess the outcome of visual acuity in Stargardt's disease

## MATERIALS AND METHODS:

This is a prospective observational study done over a period of 1 year in patients with decreased vision since childhood. Detailed history followed by best corrected visual acuity using snellen's charts, Colour vision by Ishihara charts, Amsler grid, complete ophthalmic examination including anterior segment using slit lamp and posterior segment by using indirect ophthalmoscope with 20D lens and fundus photographs with CX-1 canon fundus camera with an imaging angle of 45 degrees along with auto fluorescene was done.

## RESULTS:

Out of 10 patients 60% are males, 40% are females. 70% are below 20 years of age, 30% are >35 years of age. All presented with chief complaint of decreased central vision. All patients showed colour vision abnormalities and born out of nonconsanguineous marriage. On ocular examination 7 patients have visual acuity of 6/60 in both eyes, 2 patients have 6/36 and 1 is having 6/18. Anterior segment is normal in all cases. On fundus examination by slit lamp biomicroscopy using 90D lens and indirect ophthalmoscopy along with fundus photographs shows media is clear with optic disc of normal size and shape with distinct margins,cup disc ratio of 0.3:1, AV ratio of 2:3 and an ill-defined circular lesion was seen at the macula with beaten bronze appearance in all cases. Pigmentary changes around the macula is found in 4 cases. On fundus autofluorescence a circular hypo autofluorescent lesion is found in macular region along with multiple hypo autofluorescent flecks in the posterior poles.

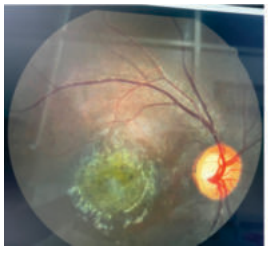

**Figure 1**

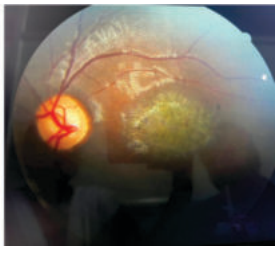

**Figure 2**

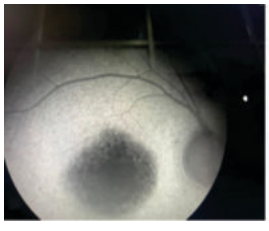

**Figure 3**

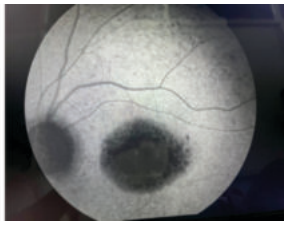

**Figure 4**

**Figure 1,2 :** Fundus images of right and left eye
**Figure 3,4 :** Fundus autofluorescence images

## DISCUSSION:

Stargardt disease (STGD) is the most common childhood recessively inherited macular dystrophy. The condition has a genetic basis due to mutations in the ABCA4 gene, on chromosome 1, that encodes a retinal transported protein; it results from the accumulation of visual cycle kinetics-derived byproducts in the retinal pigmented epithelium (RPE) with secondary photoreceptor dysfunction and death.[7] The transmembrane transporter protein, which is expressed by rod outer segments, is not produced by the ABCA4 (ABCR) gene as a result of these mutations. Since this protein is essential for vision, a lack of it eventually causes lipofuscin to accumulate in the retina.[2,3] Another crucial feature is jet-black dark choroid or silent choroid in the fluorescein angiogram. Flecks are yellowish-white round or pisciform (fish-tail) shaped lesions of varying sizes located within the RPE. They tend to develop centrifugally from the macular region and may even

progress beyond the vascular arcades to the mid-periphery but usually spare the far periphery. Peripapillary sparing (absence of flecks around the optic disc) is a diagnostic feature in STGD1 that is more prominently visible on fundus autofluorescence imaging. Around 30% of patients with childhood-onset disease may not show flecks, which may appear as the child grows. This is a critical cause of misdiagnosis. Noble and Carr classified STGD1 (Stargardt disease and fundus flavimaculatus) based on fundus appearance into 4 groups:

- **Group 1:** macular degeneration without flecks
- **Group 2:** perifoveal flecks with macular degeneration (Stargardt disease)
- **Group 3:** diffuse flecks with macular degeneration (fundus flavimaculatus with macular degeneration)
- **Group 4:** diffuse flecks, absent macular degeneration (pure fundus flavimaculatus)

Fundus autofluorescence of patients with STGD1 initially demonstrates reduced central autofluorescence in the macula, which represents RPE atrophy, surrounded by an increased signal or a bull's eye maculopathy appearance.[1] A study of patients with late-onset STGD1 (after the fourth decade of life) found that late-onset disease had clinical overlap with juvenile-onset disease except for the factors of later age at onset, slower disease progression, and greater visual acuity.[4] The consanguinity present in relatively isolated populations may be responsible for the founder effect that is commonly found in patients of this disease.[8] In FFA the hallmark finding is the "dark choroid," which appears as a highlighted retinal blood vessel against a hypofluorescent choroid. Flecks appear hypofluorescent due to the blocked transmission.Electrophysiological studies in Stargardt patients reveal that they typically maintain normal or subnormal full-field electroretinographic rods and cones responses.

Electrooculogram is affected variably and is more involved in eyes with flecks (69%) than eyes without flecks (42.5%)[9]. OCT allows early detection of lipofuscin accumulation in the RPE and photoreceptor layer disorganization.

## CONCLUSION:

Stargardt disease remains an incurable condition. As of right now, the FDA does not recommend any treatment for STGD1 patients in order to stop or reverse vision loss. Current therapeutic options include photoprotection and low-vision aids. Genetic testing also helps in genetic counselling, prognostication, and preconception planning. Patients should be given mental health counselling and informed that the disease progresses and that their deteriorating vision may eventually make it difficult for them to carry out daily tasks[(10)].

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