IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 22, Issue 1 Ser.2 (January. 2023), PP 47-49
www.iosrjournals.org

A Case Report On Stargardts Disease

Dr. N. Asha jyothi[1], Dr. V. Raja sekhar[2], Dr. L. Bharadwaj[3], Dr.V. Rajeswari[4]
Post graduate, Assistant Professor, Post graduate, Post graduate, Department of Ophthalmology, Guntur medical college. Guntur, Andhra Pradesh, India.

***Abstract**:*
We report a case of Stargardt's disease in eleven-year-old male presented with decreased vision in both eyes since childhood. On fundus examination circular lesion is seen at macula with beaten bronze appearance suggesting Stargardt's disease.
***Keywords**: Stargardt's disease, macular dystrophy*

Date of Submission: 27-12-2022 Date of Acceptance: 07-01-2023

I. Introduction:

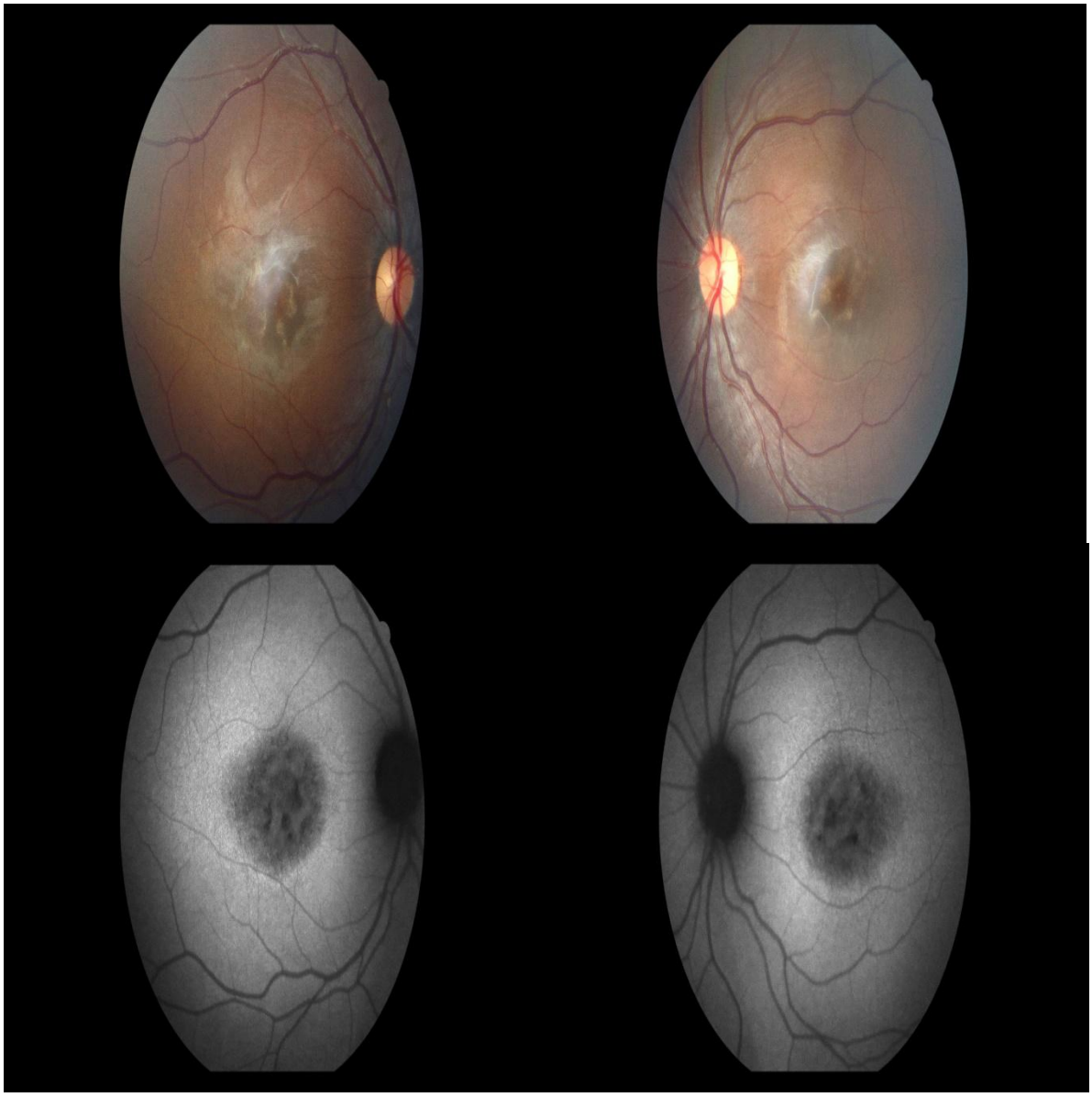
The Stargardt's disease is a frequent macular dystrophy and the most common cause of decreased central vision in adults below 50 years. It is caused by mutations in the ABCA4 gene, located on chromosome 1, which encode the ATP-binding cassette (ABC) protein transporter expressed by rod outer segments.it follows autosomal recessive inheritance pattern. There is no gender predilection and is heterogenous in both phenotypically and genetically with prevalence rate of 1 in 10000 people.it is characterised by deposition of yellowish discrete round or pisciform flecks around posterior pole at level of retinal pigment epithelium. Retinal degeneration in Stargardt's disease is believed to be caused by the toxic effects of lipofuscin deposition in the retinal pigment lumen (RPE) layer. The classical sign of phenotype of Stargardt disease is foveal atrophy surrounded by flecks with various shapes, yellowish, spherical, or pisciforms and is commonly referred to as the fundus flavimaculatus. The clinical Diagnosis of Stargardt disease is "dark choroid" or silent choroid on fluorescein angiography.[1-6]

ERG and EOG becomes subnormal in late stages. Disease presents as variably due to environmental factors and gentical modifiers[7,9,10]. Until now there has not found medical therapy for Stargardt disease. However, low vision therapy is usually beneficial for patients, and protection against exposure to bright sunlight.[1] The purpose of this case report was to report the case of Stargardt disease.

II. Case Report:

A 11 years old child was brought to OPD by parents with a chief complaint of defective vision in both eyes since 1 year associated with difficulty in identifying colours and persons. There is no similar complaint in the family and siblings were normal. There is no history of trauma, usage of drugs or any addictions. General and systemic examination is within normal limits. On ocular examination visual acuity on Snellen's chart was 6/60 in right eye and 6/36 in left eye. Colour vision was defective and not able to identify Ishihara plates. On fundus examination there is clear media and red glow is present. Optic disc of both eyes appears normal with distinct margins, vessels arising from centre of the disc with normal 2:3 AV ratio. A circular lesion which is ill-defined seen at macula with absent foveal reflex. On fundus autofluorescence there is hyper autofluoroscent areas involving macula. A diagnosis of Stargardt's disease was made. Fluorescein angiography is not done because of unavailability. Electrophysiological tests are not done. Patients were counselled about prognosis of this condition in complete details and advised to use low vision aids.

Fundus pictures of both eyes.



Fundus autofluorescent pictures of both eyes

III. Discussion:

Stargardt's disease is genetic condition caused by mutations in the ABCA4 gene, located on chromosome 1, which encode the ATP-binding cassette (ABC) protein transporter expressed by rod outer segments. This protein is needed for vision and its deficiency eventually leads to collection of lipofuscin in the retina. [11,12] Mutations in ABCA4 can also cause autosomal recessive cone rod dystrophy.[13]

Some ABCR-variant alleles also enhance susceptibility to age-related macular degeneration but further studies are required in this regard.[14]

Glazer and Dryja have proposed three step pathophysiology of Stargardt's disease which states

(1) defective Rim protein encoded by ABCA4 gene cause build-up of protonated N-retinyledine-PE in the outer segments of rods

(2) A2E a by-product of N-retinyledinePE collects in retinal pigment epithelium cells and cause toxicity

(3) photoreceptors ultimately atrophy owing to loss of retinal pigment epithelium support function.[15]

In most cases, the parents of people with Stargardt disease each have one damaged ABCA4 gene. A child that inherits a damaged gene from each parent will be affected. This is autosomal recessive inheritance. However, the risk of a person with Stargardt disease having an affected child is very low.

There is another condition called Stargardt-like disease. It is due to mutations in the ELOVL4 gene. In this case, even one damaged gene is enough to cause the condition. This is called autosomal dominant inheritance.

Stargardt like disease can be present in multiple generations of a family. Genetic testing and counselling can distinguish between these conditions.

No treatments are currently approved to prevent or slow the vision loss associated with Stargardt disease. Low vision aids are prescribed as no other treatment is currently available.[16] However, it is important to have regular eye exams even if vision is constant in order to avoid serious but treatable complications such as macular edema.

Many researches have been carried out to propose treatments for Stargardt disease. Two types of treatment have already reached the stage of clinical trials. Gene therapy refers to treatments that aim to place healthy genes into retinal cells, replacing the gene mutation preventing further vision loss. A second set of trials is exploring the transplant of new RPE cells derived from stem cells. This might prevent or slow further vision loss.

IV. Conclusion:

Stargardt disease is one of the most common causes of inherited childhood and adulthood visual impairment. Stargardt disease is highly heterogeneous both phenotypically and genetically with significant advances have been made in our ability to identify the disease at the earliest stages, characterise clinical features that allow better-informed advice on prognosis, perform accurate rapid molecular genetic testing, and in our understanding of underlying disease mechanisms.

There is no conflict of interest.

Nil finance.

References:

[1]. American Academy of Ophthalmology 2014-2015. Vitreous and Retina. San fransisco. American Academy of Ophthalmology. 2014.
[2]. Deutman, AF, Hoyng, CB, et al. Macular Dystrophies. In: Retina 4th ed. New York; Elsevier.
[3]. Telander, DG, Small, KW. Macular Dystrophies. In: Ophthalmology 4th ed. 2014. New York; Elsevier.
[4]. Kanski, Jack. Clinical Ophthalmology A Systematic Approach. Seventh edition. 2011. Elsvier.
[5]. McBain, VA, Townend, J, Lois, N. Progression of Retinal Epithelial Atrophy in Stargardt Disease. Am J Ophthalmol 2012;154:146–154.
[6]. Teussink, MM, Lee, MD, et al. The Effect of Light Deprivation in Patients with Stargardt Disease. Am J Ophthalmol 2015;159(5):964–972.
[7]. Lambertus, S, Van Huet, RAC, et al. Early-Onset Stargardt Disease Phenotypic and Genotypic Characteristics. Ophthalmology 2015;122:335-344.
[8]. Kim, LS, Fishman, GA. Comparison of Visual Acuity Loss in Patients with Different Stages of Stargardt's Disease. Ophthalmology 2006;113: 1748–1751.
[9]. Westeneg-van Haaften, SC, Boon, CJF, et al. Clinical and Genetic Characteristics of Late-onset Stargardt's Disease. Ophthalmology 2012;119:1199–1210.
[10]. Testa, F, Melilo, P, et al. Macular Function and Morphologic Features in Juvenile Stargardt Disease—Longitudinal Study. Ophthalmology 2014;121:2399-2405.
[11]. Cremers FP, Van de Pol DJ, Van Driel M, et al. Autosomal recessive retinitis pigmentosa and conerod dystrophy caused by splice site mutations in the Stargardt's disease gene ABCR. Hum Mol Genet, 1998 Mar; 7(3): 355-62.
[12]. Maugeri A, Klevering BJ, Rohrschneider K, et al. Mutations in the ABCA4 (ABCR) gene are the major cause of autosomal recessive cone-rod dystrophy. Am J Hum Genet, 2000; 67: 960–966.
[13]. Rudolph G, Kalpadakis P, Haritoglou C, Rivera A, Weber BH. Mutations in the ABCA4 gene in a family with Stargardt's disease and retinitis pigmentosa (STGD1/RP19) Klin Monatsbl Augenheilkd, 2002; 219: 590–596.
[14]. Bernstein PS Leppert M Singh N Genotypephenotype analysis of ABCR variants in macular degeneration probands and siblings. Invest Ophthalmol Vis Sci., 2002; 43: 466–473.
[15]. Glazer LC, Dryja TP. Understanding the etiology of Stargardt's disease Ophthalmol Clin North Am, 2002 Mar;15(1): 93-100.
[16]. Fishman GA, Farber M, Patel BS, et al. Visual acuity loss in patients with Stargardt's macular dystrophy.Ophthalmology, 1987 Jul; 94(7): 809-14.

Dr. N. Asha jyothi, et. al. "A Case Report On Stargardts Disease." *IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)*, 22(1), 2023, pp. 47-49.