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Stargardt's Disease : A Case Report

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ABSTRACT

Introduction. The Stargardt disease is a frequent macular dystrophy and the most common cause of decreased central visus in adults below 50 years. This condition is caused by the presence of mutations in the ABCA4 gene, located in the chromosome chain 1 short sleeve, which encode the ATP-binding cassette (ABC) protein transporter expressed by the outer trunk cell segment.

Case Presentation. A man, 59 years old, civil servants, was living in the city, came to the eye Polyclinic RSMH on February 4, 2016. From anamnesis, main complaint of the right eye was view blurred since 2 years ago. The history of the illness since ± 2 years ago, the sufferer complained the right eye view blurred slowly, the longer the more blurred. Often glare (+), the sufferer also complains to see black spots, such as enclosed curtains (-).

Conclusion. A case of Stargardt disease has been reported in a 58-year-old male. Patients come with complaints of blurry vision slowly in both eyes. On posterior segment examination, a picture of atrophy in the macula that typically leads to Stargardt disease. Additional examination of OCT also showed a stretch of RPE depletion. Supportive therapy such as correction with low vision aids or the provision of sun protective glasses can help in the daily lives of patients.

Keywords : stargardt's disease, case report

Introduction

The Stargardt disease, also known as the Fundus Flavimakulatus, is a frequent macular dystrophy and the most common cause of decreased central visus in adults below 50 years.

The vast majority of these diseases are in an autosomal recap. 1 This condition is caused by the presence of mutations in the ABCA4 gene, located in the chromosome chain 1 short sleeve, which encode the ATP-binding cassette (ABC) protein transporter expressed by the outer trunk cell segment. Retinal degeneration in Stargardt disease is believed to be caused by the toxic effects of lipofusin accumulating in the retinal pigment lumen (RPE) layer.[1-5]

Stargardt disease can be about individuals in all genders, races, and ages, with incidence number 1:10,000 inhabitants.[1,2,5] sharp Vision, overview of Funduscoffee, and the severity of the disease varies. Typically, the fundus flecks and Atropi in the macular and midperipheral retina are observed, with varying degrees of function loss including only macular dysfunction or accompanied by conical cell dysfunction and stem in the peripheral. [5]

The classical sign of phenotype of Stargardt disease is the emergence of symptoms of Atropi fovea surrounded by flecks with various shapes, yellowish, spherical, or pisciforms that appear in the age of teenagers. This condition is commonly referred to as the *fundus flavimakulatus.* The clinical Diagnosis of Stargardt disease is ensured by the description of "dark choroid" with a fluorine examination of angiography.[1-6]

Until now 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.

Case Report

A man, 59 years old, civil servants, was living in the city, came to the eye Polyclinic RSMH on February 4, 2016, with a medical record number of 936028. From anamnesis, main complaint of the right eye was view blurred since 2 years ago.

The history of the illness since ± 2 years ago, the sufferer complained the right eye view blurred slowly, the longer the more blurred. Often glare (+), the sufferer also complains to see black spots, such as enclosed curtains (-). But the sufferer never went to medicine. Since ± 1 year ago, the sufferer complained of a blurred right eye view followed by a blurred left eye view. Since ± 1 month ago, sufferers complained the view of both eyes increasingly blurred and increasingly disrupted the activity, so the sufferer decided to take treatment to RSMH Palembang

History of the former DM history (-), History of Hypertension (-), history of wearing glasses (-), History of previous trauma (-), a history of blurred eyes since childhood denied, family history with the same complaint denied.

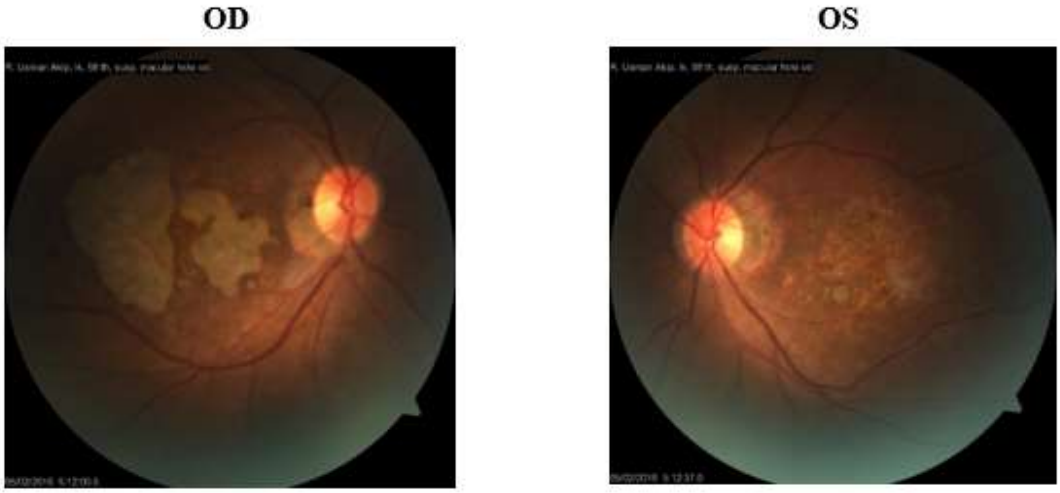
Physical examination of the generalist status of Sens: Compost mentis, blood pressure: 120/80 mmhg, Nadi: 78 x/mg, Temp: Afebris, head: Eye (see Oftalmologicus status), neck: No KGB enlargement, Cor: no abnormalities, Pulmo: vesicular N, wheezing (-), retraction (-), abdominal: flat, Pain Press (-) H/L uncandied, extremities: no abnormalities.

Table Ophthalmology Status

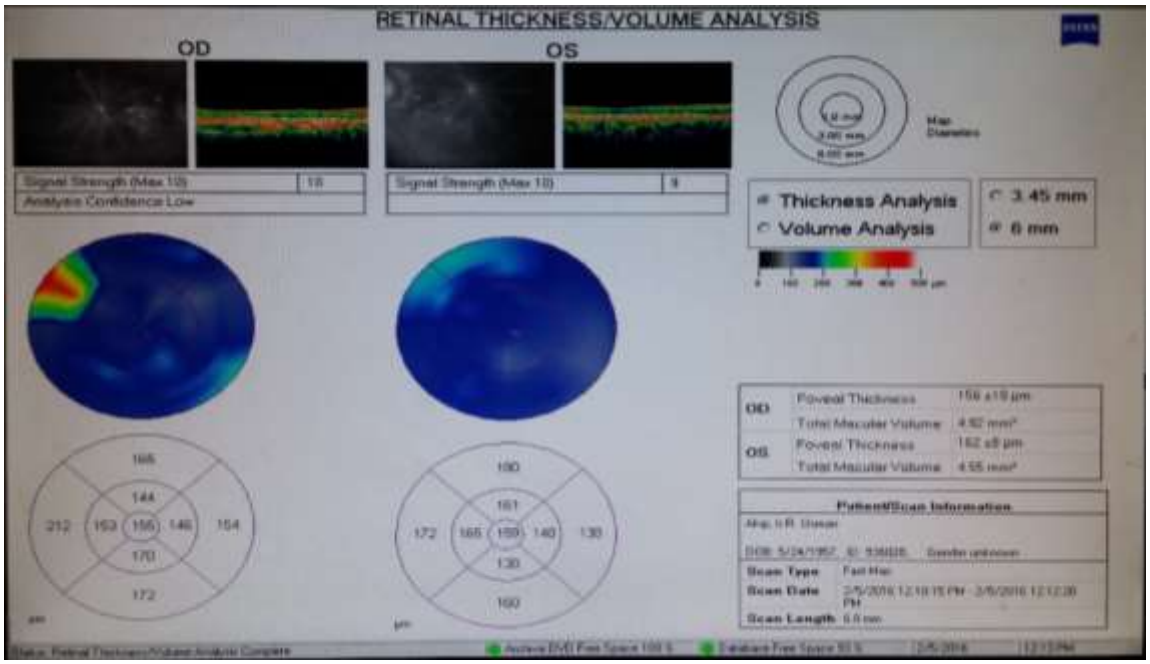
	Right eye	Left Eye
Vision	1/60	6/60 pH 6/15
TIO	18,5 mmHg	18,5 mmHg
KBM	Orthophoria	
GBM	Good in all directions	Good in all directions
Palpebra	Quite	Quite
Conjunctiva	Quite	Quite
Cornea	Transparent	Transparent
BMD	Medium, VH grade 4	Medium, VH grade 4
Iris	Good image	Good image
Pupil	B, C, RC (+), Ø 3 mm	Round, C, RC (+), Ø 3 mm
Lens	Transparent	Transparent
Posterior segment	RFOD (+)	RFOS (+)
Papil	Round, firm boundary, normal red color, c/d 0.3, a/v 2:3, the papillary atrophy (+)	Round, firm boundary, normal red color, c/d 0.3, a/v 2:3, the papillary atrophy (+)
Macula	RF (-), macular atrophy (+)	RF (+) ↓, exudate (+), macular atrophy (+)
Retina	Good blood vessel contour	Good blood vessel contour

The work Diagnosis is Stargardt of ODS disease. Stewardship is Informed consent, pro Photo fundus ODS, pro OCT astylopsis ODS, Pro FFA checkup, Pro ERG examination, pro Consul Refraction subdivision.

Fundus ODS photo image



Pictures of OCT Makula ODS



The Prognosis is Quo ad vitam: Bonam, Quo ad functionam: Dubia ad Malam.

Discussion

A man 59 year old man came to a polyclinic of RSMH eyes with a complaint of both eyes blurring since 2 years ago. The right eye view is felt first blurred followed by blurring on the left eye. The sufferer complains like seeing black dots covering his views. Sufferers also complain often glare especially when the eyes are exposed to bright sunlight.

At the ophthalmology examination obtained sharp vision on the right eye 1/60 while on the left eye 6/60 ph 6/15. The other anterior segment is within normal limits. In the posterior

segment test, there is no Reflec fovea on the right eye accompanied by an image of Atropi on the macula. While on the left eye there is a decrease in the Reflec fovea and the description of exudate on astylopsis and there is a astylopsis Atropi network. The depiction of Atropi on this astylopsis causes a reduction of visus in both patients ' eyes slowly. The existence of this defect also caused the complaint of black spots on the eyes. A central scotoma may occur in patients. It is advisable to do a field inspection to assess how broadly the defect occurs in both eyes. The OCT additional check is done to assess the thickness of the astylopsis in both eyes. Acquired image thinning coating indicating tissue atropi

From the history, ophthalmology examination and additional examination, the diagnosis of Stargardt disease is made. Standard disease is a genetic disorder inherited by autosomal rersesif with an incidence of 1: 10,000 inhabitants. The hallmark of Stargrdt disease is a sharp slowing down of vision in both eyes. From the examination of the posterior segment, it is found that there is atrophy of the macula surrounded by yellowish exudate spots. This disorder is caused by a genetic mutation in ABCA4, resulting in protein dysfunction. This disorder causes the accumulation of lipofuscin in RPE so that it will cause an acceleration of cell death.[3] A typical feature in Stargardt disease is 'beaten bronze' in the macula due to atrophy of RPE tissue.

Other tests that are useful in establishing the diagnosis of this case are Fundus Fluorescens Angiography (FFA) and electroretinogram (ERG) examination. FFA examination is carried out to assess atrophy areas. However, this examination was not carried out due to the unavailability of fluorescent substances in RSMH. ERG examination also can not be done because of the unavailability of tools in RSMH.

Management of patients with Stargardt disease to date has still not been found. There is no specific medical therapy that can be given because the cause of the disorder is genetic mutations. Until now, trials are still carried out for stem cell therapy, but there are no satisfactory results. As a tool, it can be consulted to the refraction section to provide low vision aids so that patients will be more helped in their daily work. Provision of eye protection against sunlight can also be given to reduce glare complaints in patients.

The prognosis of quo ad vitam bonam is because there are no abnormalities in the systemic organs that can threaten the life of the patient. While the prognosis of quo ad functionam dubia ad night is because the lesions are in the macula and there is no specific therapy that can treat this disease.

Conclussion

A case of Stargardt disease has been reported in a 58-year-old male. Patients come with complaints of blurry vision slowly in both eyes. On posterior segment examination, a picture of atrophy in the macula that typically leads to Stargardt disease. Additional examination of OCT also showed a stretch of RPE depletion.

Until now, there is no specific therapy for Stargardt disease. Supportive therapy such as correction with low vision aids or the provision of sun protective glasses can help in the daily lives of patients. Prognosis for visual function in dubia ad night because until now there is no specific therapy.

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