

# NEUROPSYCHIATRIC SYNDROME IN CHOROIDEREMIA- A RARE CASE REPORT

## ABSTRACT

Biomarkers of Alzheimer's disease (AD), and other neurodegenerative disorders are most exploring areas of clinical research in Geriatric Psychiatry. Presently neuroimaging, blood and CSF biomarkers are expensive and invasive to perform and are considered impractical for research purpose. The retina presents a readily accessible tissue for monitoring the brain and could potentially be used in combination with or as a substitute for current AD and other neurodegenerative disease biomarkers. Data on retinal involvement in AD and neurodegenerative diseases have been controversial and inconsistent. So, we are reporting a case of Choroideremia with neuropsychiatric presentations and complications.

**KEY WORDS** – Choroideremia, Neuropsychiatry, Retina, Vision

## INTRODUCTION

Choroideremia is a x-linked recessive, progressive, degenerative disorder of retina and choroid that mainly affects males. It is caused by loss of function mutations in the Choroideremia gene resulting in decreased Rab escort protein-1 (REP-1) expression which leads to degeneration of the retinal pigment epithelium, photoreceptors, and choroid. The first symptom of this condition is usually an impairment of night vision (night blindness), which can occur in early childhood. A progressive narrowing of the field of vision (tunnel vision) follows, as well as a decrease in the ability to see details (visual acuity).[1] These vision problems are due to an ongoing loss of cells (atrophy) in the specialized light-sensitive tissue that lines the back of the eye (retina) and a nearby network of blood vessels (the choroid). The vision impairment in Choroideremia worsens over time, but the progression varies among affected individuals. [2] Visual impairments due to Choroideremia are associated with emotional, functional, and economic burden. Prevalence is 1 in 50,000 to 100,000 and accounts 4% of all blindness.[3] There have been some reports of Choroideremia patients with neurological complications. **Bibliografia** But there is scarcity of literature on neuropsychiatric symptomatology and Choroideremia.[4]

## CASE PRESENTATION

A 65-year-old gentleman, right-handed, educated up to post graduation, non-hypertensive, non-diabetic and a known case of Choroideremia which was diagnosed with genetic and ophthalmic investigations, with complains of Increased irritability for 2 years, Dizziness and Frequent falls, for 1.5 years and increased for 1.5 months, tremulousness of right side of hands at rest for 1.5 years, Forgetfulness for 6 months increased for 1.5 months. On further enquiry patient reports that he is also having bi-occipital dull aching headache for 1.5 months which gets partially relieved on he turns his head upwards. So, most time of day he would keep his head upward position. He is dependent on family members for daily activities for and unable to handle his finance, laundry, medications for 1.5 months. History of frequent head injury due to fall. In premorbid personality he had obsessive compulsive personality traits. On general examination he was conscious, oriented. Neurological examination suggestive of cogwheel rigidity in right upper limb, power, reflexes in all limbs normal. Pill

rolling tremors in right hand at rest. For walking he needed assistance and had shuffling gait with stopped posture. **Hoehn- Yhar??** Bilateral pupils reacting to light but only perception of light was present. On Mental Status Examination- recent memory impaired, with Mini Mental Status Examination score 22/28, copying and writing he could not do due to vision impairment. **insuficiente**

#### Chart 1: INVESTIGATIONS:

Investigation	Report
Total Leukocyte count	9800
Haemoglobin	12.4 g/dl
Random Blood Sugar Levels	142 mg/dl <b>normal????</b>
Serum Bilirubin	0.7
SGOT	44
SGPT	42
Thyroid Stimulating Hormone	5.84
T4	5.29
T3	0.871
EEG	Awake EEG within normal limits
CT Brain	Subdural hygroma with diffuse cerebral atrophy
MRI Brain	Moderate size subdural hygroma in bilateral frontotemporal region extending to partial region producing diffuse compressing effects

#### TREATMENT

Considering irritability patient started on Tablet Quetiapine 25 mg at night and as per needed. In view of MRI Brain changes patient referred to neurosurgery services and he started on Tablet Syndopa 125mg four times daily.

#### OUTCOME AND FOLLOWUP

Patient followed 2 weeks and improvement was noticed in the irritability, tremors but he still needed support for walking.

## **DISCUSSION**

Choroideremia is a degenerative eye disease which affects night vision, usually in the first or second decade of life. Then peripheral field loss occurs over the next few decades. The disease presents with patchy areas of depigmentation which spreads from periphery to central part of choroid retinal layer with a characteristic salt and pepper mottling. [5,6] In the involved areas, there is diffuse atrophy of the chorio capillaries and Retinal pigmented epithelium. The central visual acuity is preserved until the patient's 50s, and retinal blood vessels are not affected. The macular area remains intact until late in the course of disease, with preserved visual acuity. There are case reports of Choroideremia with clinically undiagnosed psychiatric syndromes. Some case has presented with isolated symptoms of nyctalopia, progressive visual painlessness, painless and foreign body sensation. [7,11,12]

In this case report subject had unreported visual symptoms till the age of 30. He started symptoms of difficult vision in night at his 30's but he was diagnosed with the condition from ophthalmologist by genetic testing and with optical coherence tomography. Although neuropsychological symptoms are difficult to diagnose and under reported and this case report shows the rarest presentation of Choroideremia. [7-9] As the disease is X linked his son has got a risk of developing the condition so the geniting counselling was also conducted. The absolute diagnosis of Choroideremia is based on the identification of clinical features combined with additional imaging exams which is done in all the reported cases. [10-12]

## **PROGNOSIS**

As this a genetic condition with unsatisfactory prognosis symptoms can be controlled with periodic ophthalmologic examination especially when central vision is affected, and cystoid macular oedema is suspected. The neuropsychiatric sequel can be successfully controlled with early diagnosis and preventive measures.

## **CONCLUSION**

Choroideremia is a genetic retinal disease which is rare and causes blindness mainly in middle-aged men. We are yet to know how the biochemical defect that causes the retinal-specific disease and if it has any hidden effects on the rest of the body especially the brain. This case report throws light into the unusual neuropsychiatric symptoms in an established case of Choroideremia.

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