*CASE REPORT*

# An Unusual Case of Refsum Disease

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**ABSTRACT**

Refsum disease is a rare autosomal recessive disorder characterized by the accumulation of phytanic acid in plasma and tissues due to deficiency of the enzyme phytanoyl-CoA hydroxylase that break down phytanic acid. This case report presents a 55-year-old male patient with Refsum disease exhibiting classical symptoms such as retinitis pigmentosa, peripheral neuropathy, hearing loss, and skeletal abnormalities, however he had some atypical features of very small pupil (microcoria). Management primarily involves dietary restrictions, particularly the avoidance of foods rich in phytanic acid. Additionally, symptomatic treatment aimed at managing the neurological and sensory impairments was already employed. This case highlights the importance of early diagnosis and multidisciplinary care in mitigating the progression of Refsum disease and improving the patient's quality of life.

**Key words:** Refsum disease, small pupil, retinitis pigmentosa

## INTRODUCTION

Refsum disease is an autosomal recessive neurocutaneous syndrome that is characterized by the accumulation of phytanic acid in plasma and tissues. Most of the patients with refsum disease have skin, hearing, vision, balance and smell problems along with retinitis pigmentosa. This patient has some different ocular findings than the routine patients of refsum disease. These unusual findings include unilaterally constricted pupil and micro cornea.

## CASE REPORT

A male patient of age 55 years patient presented to me with complaints of decrease of vision in both eyes. Vision was markedly decreased at night since childhood but now his day vision also started to deteriorate. His vision in right eye (RE) was counting finger (CF), not improving with refraction and left eye (LE) was 6/60 with +2DS.

Intra ocular pressure (IOP) measured with Goldman applanation tonometer was 14mm of Hg RE and 16mm of Hg Left eye. Right Eye had unusually small pupil and microcoria.The pupil was not getting dilated with 1% atropine drops instillation. There was dense cataract in the RE and fundus details were not visible. Left eye also had small pupil but patient was pseudophakic with posterior capsular opacification and fundus showed typical triad of Retinitis Pigmentosa i.e Bony spicules arteriolar attenuation and pale waxy optic disc. On further history and examination patient had ataxia, gait disturbance, neurosensory deafness with right ear completely deaf and left ear working with ear device. Patient had ichthiosis, dry scaly skin, memory loss and short and partially lost fingers of both hands.

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COI: The author has disclosed no conflict of interest.

**Figure 1**

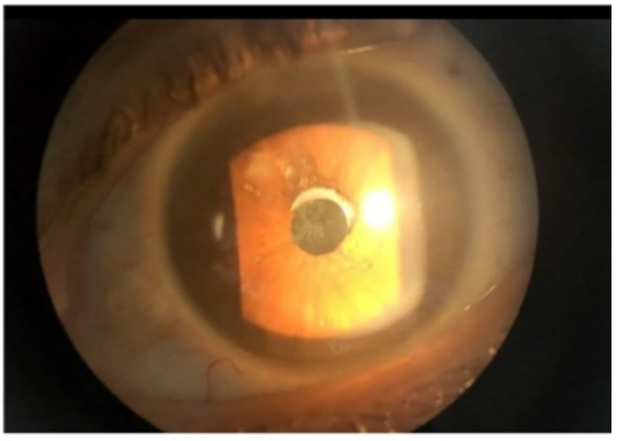

**Figure 2**

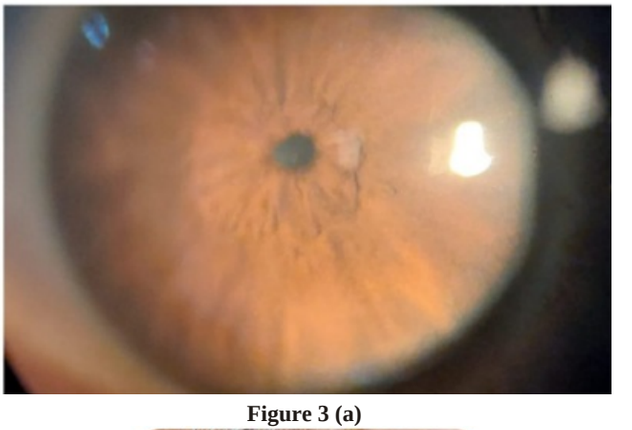

**Figure 3 (a)**

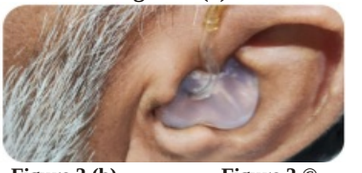

**Figure 3 (b)** **Figure 3 ©**

## DISCUSSION

Whenever we receive patients with deafness and decrease of vision, our topmost differentials diagnosis are Usher Syndrome and Refsum Disease. Mostly we get to differentiate these two on the basis of above mentioned characteristics of refsum disease. But here we diagnosed a rarer refsum disease case with other ocular features like microcoria and cataract as well. RP is present in all cases but microcoria and cataract are not as marked as we saw in this patient. The other interesting aspect was normal sense of smell. Usually we get anosmia in cases of refsum disease. So I advised the patient phacoemulsification with Foldable Intra Ocular Lens (FIOL) implant for right eye with pupil expansion devices and advised Nd YAG capsulotomy in the Left Eye.

## Conclusion

This patient had classical features for Refsun disease with unusual micro cornea and cataract but sense of smell was intact.

## REFERENCE

Jayaram H, Downes SM. Midlife diagnosis of Refsum Disease in siblings with Retinitis Pigmentosa–the footprint is the clue: a case report. J Med case rep 2008 Dec; 2:80,1-4.doi: 10.1186/1752-1947-2-80.