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Retintis Pigmentosa Sine Pigmento In Usher Syndrome

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Abstract :
Usher syndrome is a rare genetic disorder characterized by congenital neurosensory hearing loss and progressive retinitis pigmentosa (RP), leading to vision impairment. Classified into three types based on the severity of hearing loss and age of onset of retinal degeneration, Usher syndrome affects approximately 2–6 individuals per 100,000 worldwide. Diagnosis of the subtype retinitis pigmentosa sine pigmento (RPSP), which lacks typical pigmentary changes, is particularly challenging, often resulting in delayed recognition, especially when accompanied by congenital deafness.(1)
This report presents a rare case of RPSP in a 10-year-old male with profound congenital deafness, born to consanguineous parents. Despite normal vision in family members, the patient exhibited symptoms including reduced visual acuity, retinal vessel narrowing, and macular edema, confirmed by fluorescein angiography and spectral-domain optical coherence tomography (SD OCT). Electroretinography revealed characteristic abnormalities, affirming the diagnosis of RPSP.
The case underscores the importance of clinical suspicion and comprehensive evaluation in individuals with congenital deafness and visual impairment. Treatment options for Usher syndrome are limited to supportive care, including cochlear implants and low-vision aids. Ongoing research aims to elucidate genetic mechanisms, refine diagnostic approaches, and explore potential therapies.
Keywords: *Usher syndrome, retinitis pigmentosa, retinal disease, macula edema, genetic disorder*

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I. Introduction

Usher syndrome is a severe and very rare genetic disorder that involves congenital neurosensory hearing loss and progressive retinitis pigmentosa (RP), leading to impaired vision. We classify Usher syndrome into three main categories based on the severity of hearing loss, the presence of vestibular dysfunction, and the age at which retinal degeneration starts. This syndrome is inherited in an autosomal recessive manner and has a prevalence of roughly 2–6 cases per 100,000 individuals worldwide. (1)

The retinitis pigmentosa in Usher syndrome is characterized by the gradual deterioration of photoreceptor cells in the retina, leading to peripheral vision loss. (2) Nevertheless, the diagnosis of retinitis pigmentosa sine pigmento (RPSP) is challenging since it lacks the typical peripheral bone-spicule-like pigmentary alterations observed during fundus examination. Therefore, the diagnosis is often delayed, especially when there is congenital hearing loss. (1)

The purpose of this report is to present a rare case of retinitis pigmentosa sine pigmento in a patient diagnosed with Usher syndrome.

This case underlines the unusual manifestation of RPSP in the context of Usher syndrome, highlighting the significance of clinical suspicion and thorough diagnostic assessment in individuals with congenital deafness and visual impairment.

II. Case Report

A 10-year-old male patient, born to consanguineous parents with profound congenital deafness, was examined because of the visual impairment of both eyes for 6 months.
No other illnesses were reported.

His parents were normal-sighted. All his close relatives, including his sisters and brothers, reported normal vision.

The eye examination showed reduced best visual acuity on the right eye, measuring 4/10, a normal anterior segment, and in fundoscopy, diffuse retinal blood vessel narrowing, no retinal pigmentation, and macular edema. While in the left eye, the best visual acuity was 3/10, normal anterior segment, and in fundoscopy, diffuse

retinal blood vessel narrowing, no retinal pigmentation, and macular edema Color vision stereopsis, ocular motility, and biomicroscopy were normal.

Fluorescein angiography Showed salt-and-pepper retinopathy and narrowing of the arterioles SD OCT showed Bilateral cystoid macula edema The suspected diagnosis of pigmentary retinopathy without pigment was confirmed by electroretinography, which showed undetectable scotopic and abnormal photopic responses. The patient was treated with **acetazolamide** for 1 month without any improvement in visual acuity.
All the relatives have been examined without any signs of usher syndrom.
A cochlear implantation is planned.

III. Discussion :

Usher syndrome is an autosomal recessive hereditary disease. It affects hearing, vision, and, in some cases, vestibular function.

Usher syndrome can be classified into three forms depending on the severity of hearing loss, the presence of balance problems, and the age at which symptoms appear. (3)

Retinitis Pigmentosa Sine Pigmento (RPSP): RPSP is an uncommon variant of retinitis pigmentosa seen in Usher syndrome that doesn't have peripheral pigmentary changes. This variant is challenging as it may present with atypical findings such as diffuse retinal vessel narrowing, macular edema, or cystoid macular degeneration instead of the typical pigment deposits.

The diagnosis of RPSP includes clinical evaluation with visual field testing, electroretinography (ERG), and occasionally genetic testing.
Currently, there is no cure for Usher syndrome. Management focuses on early diagnosis and supportive care.

Treatment options include hearing aids, cochlear implants for severe hearing loss, and low-vision aids to optimize visual function and quality of life.

Current research on Usher syndrome aims to understand the underlying genetic mutations responsible for each subtype, develop gene therapies to prevent or slow disease progression, and improve diagnostic strategies for earlier intervention. (4)

IV. Conclusion

RPSP represents a rare and challenging variant of retinitis pigmentosa within the spectrum of Usher syndrome. Its presentation is atypical, and a full clinical evaluation, specialized diagnostic testing, and genetic analysis are necessary for its diagnosis. Continued research efforts are crucial for advancing our knowledge of RPSP, improving diagnostic strategies, and developing targeted and efficient therapies.

Figures :

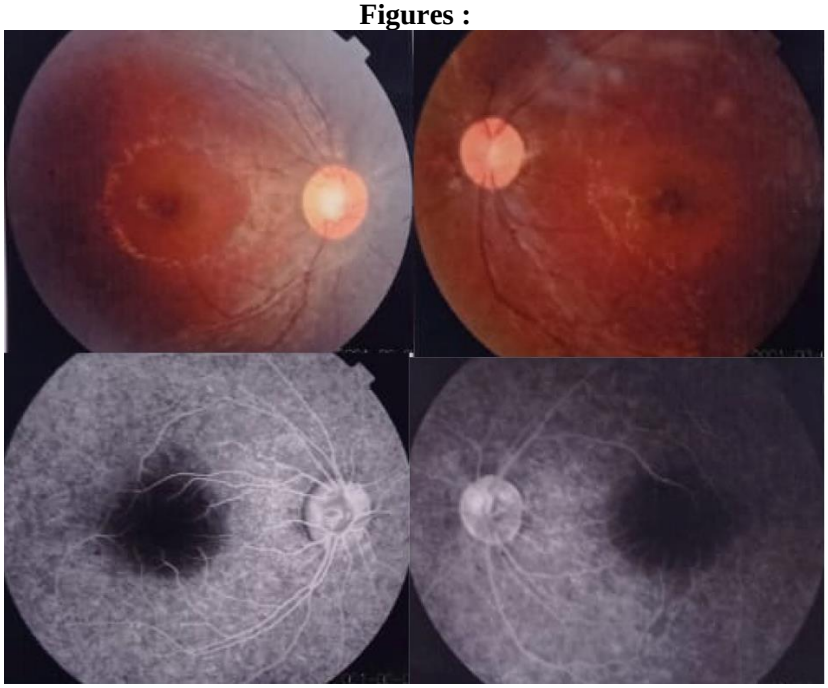

Figure 1: Fluorescein angiography showing salt-and-pepper retinopathy and narrowing of the arterioles

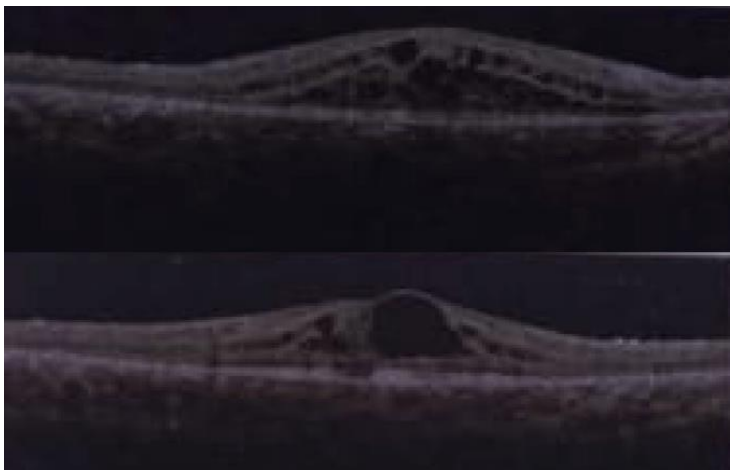

Figure 2: SD OCT showing Bilateral cystoid macula edema