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A Rare Case of Joubert Syndrome with Pigmentary Retinal Degeneration

Mohammad Ali Imtiaz[1*], Amar Salman Dalol[1], Omar Al Qahtani[1], Devendra Kumar[2], Zamzam Al Baker[1], Rana Sabah Ismail[1]

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ABSTRACT

Joubert syndrome is a rare genetic X-linked recessive disease that is inherited and characterized by hypotonia, ataxia, developmental delay, intellectual disability, a specific midbrain malformation known as the "Molar Tooth sign,". The case report presents a 20-year-old male with Joubert syndrome, characterized by cerebellar hypoplasia, hypotonia, developmental delays, and multiple organ involvement. Diagnostic assessments included neuroimaging, funduscopic examination, and laboratory tests to evaluate the multisystemic effects. The patient underwent multimodal treatment, including hemodialysis for renal failure, regular ophthalmologic evaluations, and genetic counseling to assess the hereditary nature of the condition and provide family planning advice. Retinal degeneration is associated with mutations in AH 11 and CEP290. Joubert syndrome is an uncommon pleiotropic condition that might manifest significantly in the eyes. This report necessitates the need for prompt diagnosis and management and to increase awareness about its complications.

INTRODUCTION

Joubert syndrome is an abnormal brain development condition that may influence several areas of the body (Spahiu *et al.,* 2022). It is identified by the underdevelopment or absence of the cerebellar vermis, brain stem malformation, thicker and longer cerebellar peduncles, and a deeper interpeduncular fossa (Merlini & Poncet, 2013). When combined, these cause the distinctive "Molar tooth Sign" on an MRI. While there are many different signs and symptoms, hypotonia, irregular breathing patterns (often hyperpnea, newborn apnea), atypical eye movements (oculomotor apraxia), difficulties initiating rapid horizontal eye motions, unique facial characteristics, and intellectual incapacity are among the most prevalent symptoms (Merritt, 2003). Besides the neurological symptoms, Joubert Syndrome can also involve other organs, potentially affecting the kidney, liver, and eyes, leading to a broader spectrum of clinical manifestations (Sattar & Gleeson, 2011). The condition is typically diagnosed through clinical evaluation, neuroimaging, and genetic testing (Devi *et al.,* 2020).

Due to its rarity, comprehensive epidemiological data on Joubert Syndrome (JS) is limited, but its prevalence is approximately between 1 in 80,000 to 1 in 100,000 live births (Fang *et al.,* 2023). The condition's non-specific and highly variable clinical presentation often leads to delayed diagnosis despite the manifestation of clinical features during the neonatal period. Maria *et al.* reported that the average age at diagnosis is approximately 33 months (Alam *et al.,* 2021). Most affected children reach adulthood and generally have a favorable prognosis. However, the incidence of JS and associated genetic mutations occur in both consanguineous and nonconsanguineous marriages (Mandura & Arishi, 2022). Early detection is crucial as it allows for timely intervention, potentially improving outcomes.

Pigmentary retinal degeneration (PRD) encompasses a group of inherited disorders affecting the light-sensitive cells in the retina (Carullo *et al.,* 2020). Key features include night blindness, tunnel vision, and eventual loss of peripheral vision (Moos *et al.,* 2022). These characteristic cells, called rods and cones, degenerate over time, causing symptoms like decreased night vision, peripheral vision loss, and, eventually, central vision impairment (Moos *et al.,* 2022). Fundoscopic examination often reveals retinal abnormalities such as pigmentation changes, thinning of the retina, and attenuation of blood vessels (Padungkiatsagul *et al.,* 2020). Importantly, some subtypes of Joubert syndrome are known to be associated with PRD, highlighting the potential for extra-neurological involvement in this condition (Gana *et al.,* 2022).

This case study reports a rare presentation of Joubert syndrome with co-existing pigmentary retinal degeneration. The aim of presenting this case was to Contribute to the understanding of the clinical spectrum of JS by illustrating a case with combined neurological and ophthalmologic features. Furthermore, this report emphasized the importance of comprehensive clinical evaluation in JS patients, including ophthalmologic examinations, especially when specific subtypes are suspected to involve other organ systems. It also potentially guides future research on the underlying mechanisms linking JS and PRD.

[1] Department of Ophthalmology, Al Wakra Hospital, Hamad Medical Corporation, Doha Qatar
[2] Department of Radiology, Al Wakra Hospital, Hamad Medical Corporation, Doha Qatar
[*] Corresponding author's e-mail: maimtiaz123@outlook.com

Case Presentation

Patient Demography

This case report presents a 20-year-old male patient with no documented familial history of ocular anomalies. Parental consanguinity was absent, and the patient's nine siblings (five males and four females) were reported to be healthy.

Editorial Policies and Ethical Considerations

The study received approval from the appropriate ethics committee. Informed consent was obtained from all the participants in the research.

Clinical History

The patient's medical history revealed the presence of Joubert syndrome (JS) since early childhood, characterized by cerebellar hypoplasia, hypotonia, and developmental delay. Congenital abnormalities were also evident at birth, including hepatic fibrosis and cardiac septal defects. Renal involvement manifested later in life, with cystic renal dysplasia progressing to end-stage renal disease requiring hemodialysis initiation in 2008. Cyanotic congenital heart disease and a bicuspid aortic valve were diagnosed when he was an infant. While no initial family history of ocular abnormalities was reported, progressive vision problems were noted. Funduscopic examination revealed bilateral peripheral retinal pigmentary degeneration and optic disc pallor, suggestive of retinal dysfunction. Additionally, atrophic changes at the macula, a hallmark of retinitis pigmentosa, were observed. This presentation highlighted the multisystemic nature of the patient's condition.

Diagnostic Assessment

Radiographical Investigation

Neuroimaging with head CT revealed hypoplasia of the cerebellar vermis with a cyst in continuity with the fourth ventricle, as shown in Figure 1. Additionally, the CT scan demonstrated the characteristic "bat wing" and "molar tooth" signs, indicative of Joubert syndrome. These findings were accompanied by prominent and elongated superior cerebellar peduncles and a thickened skull bone. A chest X-ray showed bilateral hilar congestion, left basal infiltrates, and blunting of the right costophrenic angle suggestive of effusion as shown in Figure 1. Furthermore, an enlarged heart was visualized on both the chest X-ray and CT scan. The CT scan also revealed bilateral basal atelectatic patches and fibrotic strands, enlargement of various organs, including the heart and left liver lobe (with atrophy of the right lobe), mild splenomegaly, and bilateral atrophic kidneys with multiple cysts. Changes consistent with renal osteodystrophy were noted as well. These findings from multiple imaging modalities paint a picture of a complex multisystemic condition affecting the brain, lungs, heart, liver, kidneys, and skeletal system.

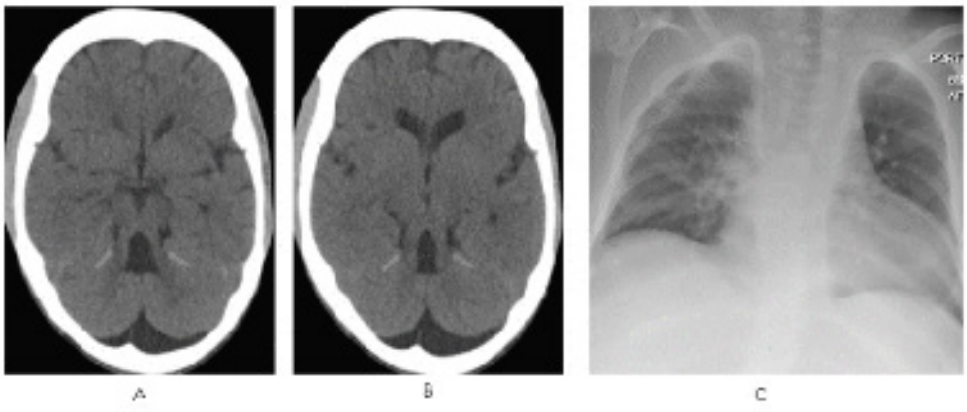


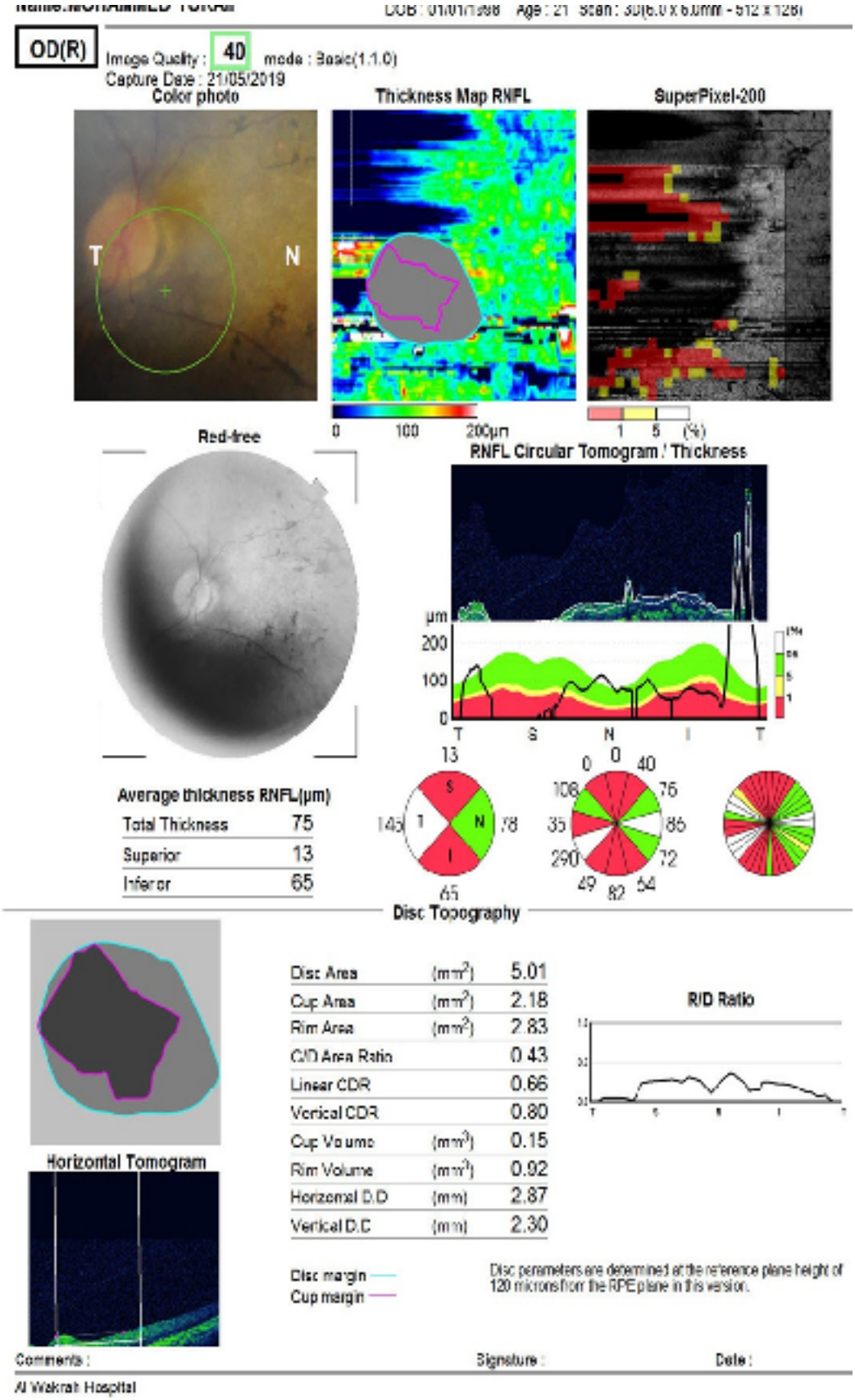
Figure 1: A, B, C: Axial CT non-contrast images (A, B) show hypoplastic vermis and elongated superior cerebellar peduncles (blue arrows) giving classical appearance of 'Molar Tooth sign'. Chest radiograph (C) shows diffuse cardiomegaly with bilateral perihilar vascular congestion with right central line.

Funduscopic Examination

Ocular examination revealed bilateral anterior and posterior blepharitis with a follicular conjunctival reaction, suggesting inflammation of the eyelid margins and conjunctiva. However, the corneas, anterior chambers, pupils, and lenses appeared normal. Funduscopic evaluation demonstrated bilateral peripheral retinal pigmentary degeneration, characterized by attenuated retinal vessels as shown in Figure 2, optic disc pallor, and atrophic changes at the macula, findings consistent with retinitis pigmentosa. Further assessment with Optical Coherence Tomography (OCT) revealed a reduction in the average nerve fiber thickness in both eyes, potentially indicating optic nerve damage. Ocular motility examination showed no abnormalities, and nystagmus (involuntary eye movements) was absent, as shown in Figures 3, 4 and 5. Unfortunately, the patient's lack of cooperation prevented the assessment of visual acuity.

Laboratory Investigation

The laboratory investigations revealed abnormal values for several parameters. Blood urea (12.6) and creatinine (294) were significantly elevated, indicative of impaired kidney function. Calcium levels were low (1.83), potentially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Rim Area (mm²) 2.83
C/D Area Ratio 0.43
Linear C/DR 0.66
Vertical C/DR 0.80
Cup Volume (mm³) 0.15
Rim Volume (mm³) 0.92
Horizontal D.D (mm) 2.87
Vertical D.D (mm) 2.30
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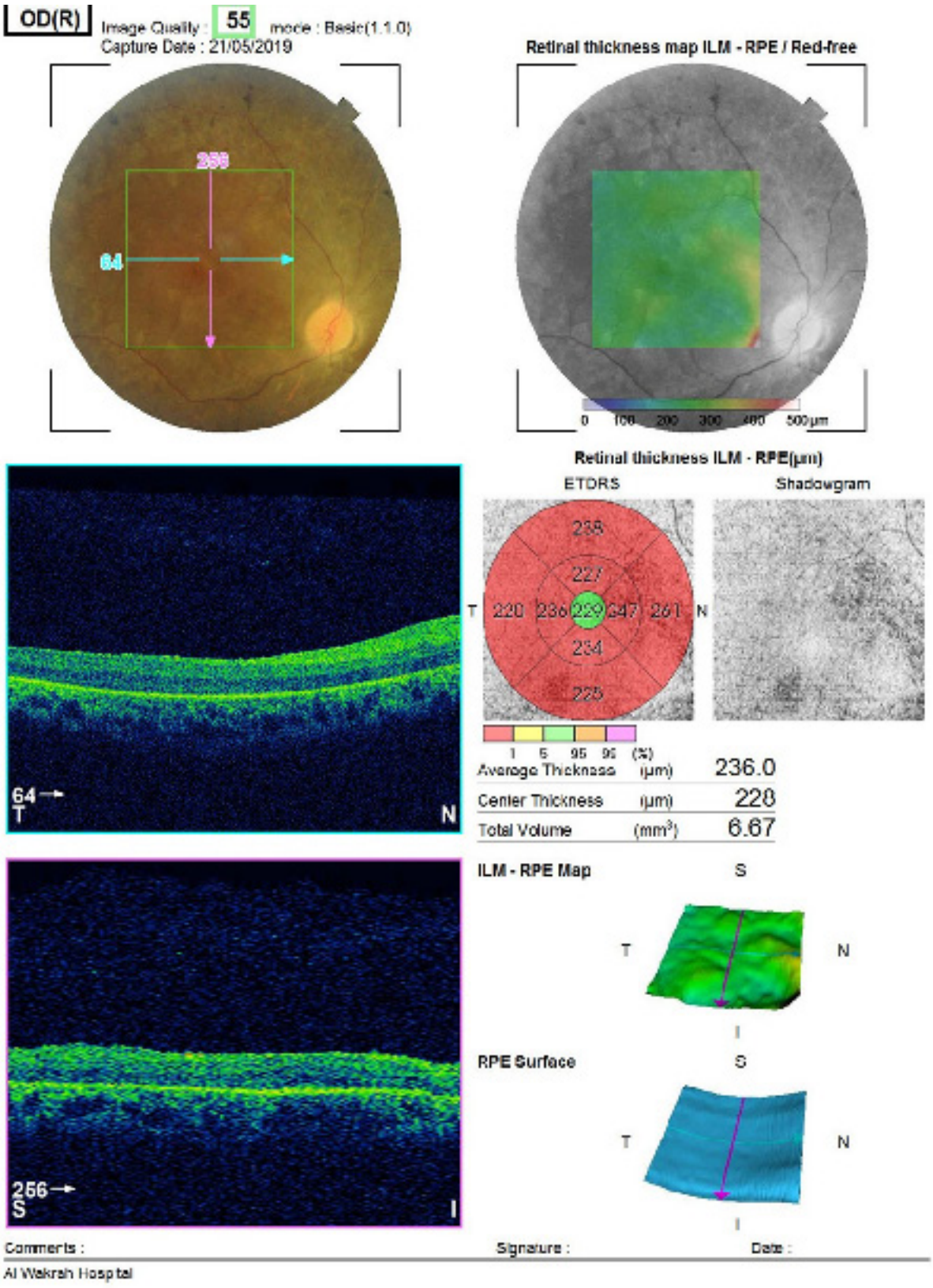
Figure 2: ???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Retinal thickness map ILM - RPE / Red-free
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Retinal thickness ILM - RPE(μm)
ETDRS
Shadowgram
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227
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225
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Average Thickness (μm) 236.0
Center Thickness (μm) 228
Total Volume (mm³) 6.67
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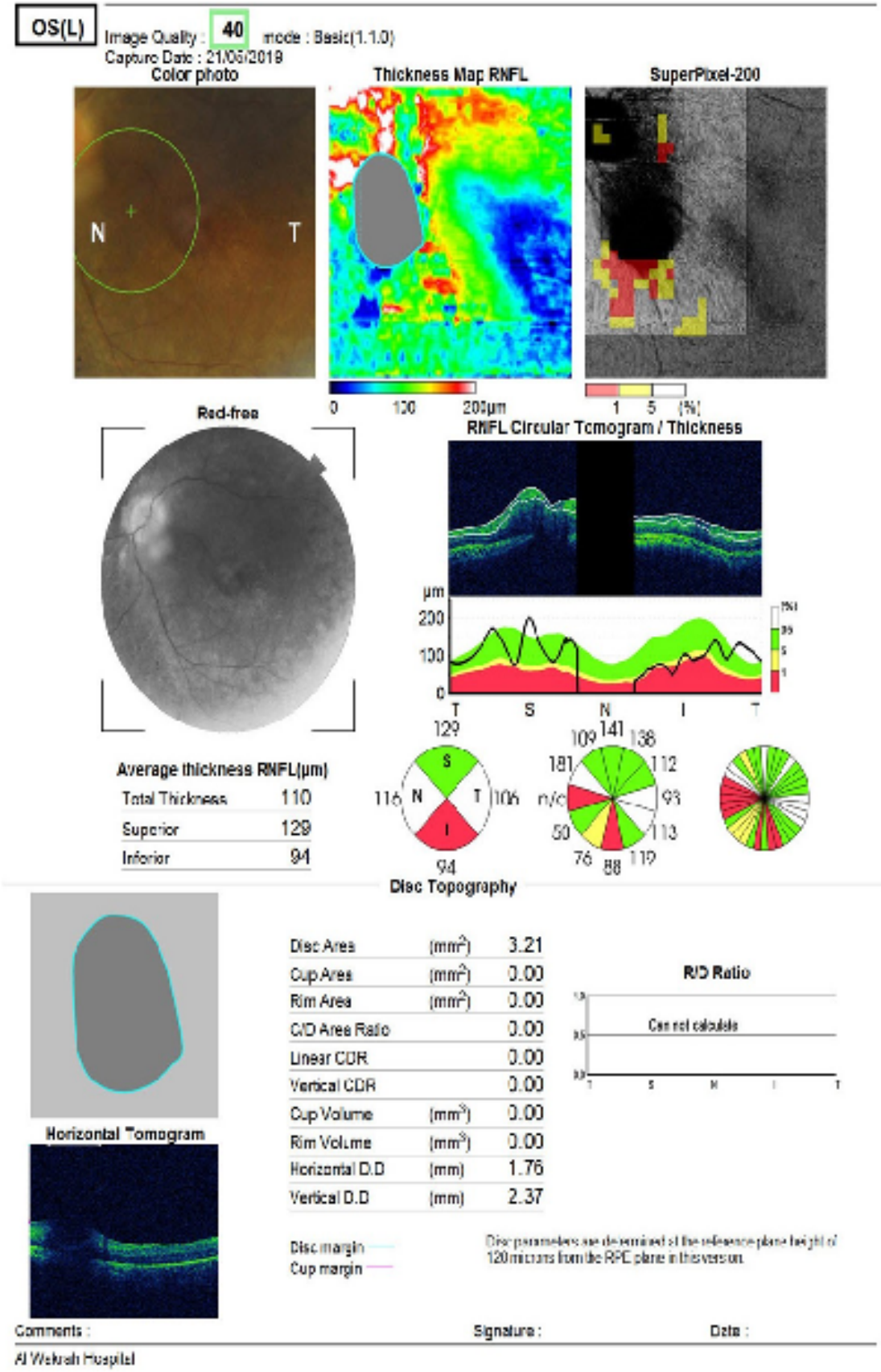
Figure 3: ???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Average thickness RNFL(µm)
Total Thickness 110
Superior 129
Inferior 94
129
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116
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94
109
141
138
181
112
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93
50
113
76
88
119
Disc Topography
Horizontal Tomogram
Disc Area (mm²) 3.21
Cup Area (mm²) 0.00
Rim Area (mm²) 0.00
C/D Area Ratio 0.00
Linear CDR 0.00
Vertical CDR 0.00
Cup Volume (mm³) 0.00
Rim Volume (mm³) 0.00
Horizontal D.D (mm) 1.76
Vertical D.D (mm) 2.37
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Disc margin
Cup margin
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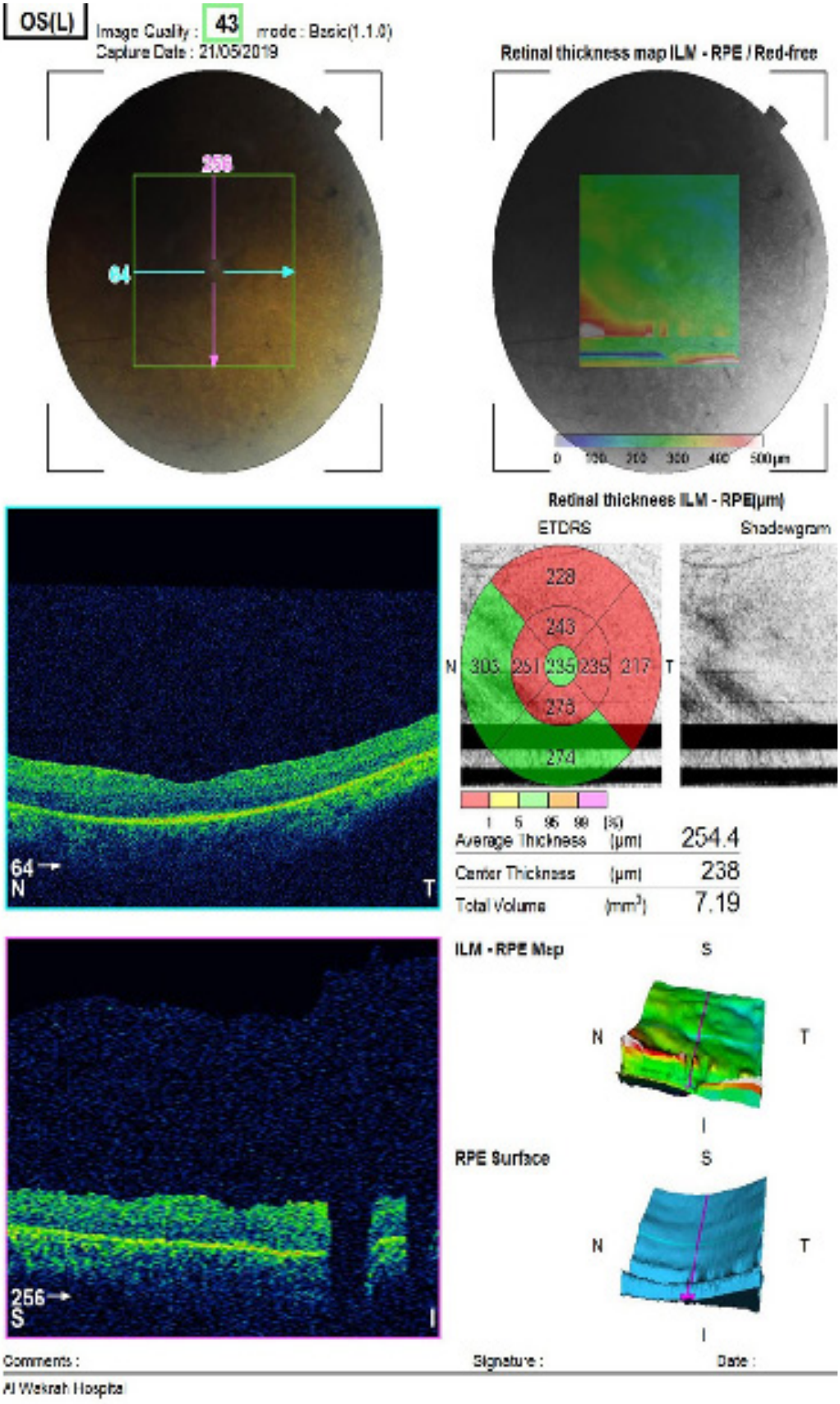
Al Wehrah Hospital

Figure 4: ???


OS(L)
Image Quality : 43
mode : Basic(1.1.0)
Capture Date : 21/05/2019
Retinal thickness map ILM - RPE / Red-free
Retinal thickness ILM - RPE(μm)
ETDRS
Shadowgram
228
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Average Thickness (μm) 254.4
Center Thickness (μm) 238
Total Volume (mm³) 7.19
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Figure 5: ???

contributing to bone complications. Bilirubin (526) was markedly elevated, suggesting problems with bile metabolism or liver function. While total protein (54) was within the normal range, AST (95) and ALT (50) enzymes, markers of liver damage, were slightly elevated. Glucose (5.6) was normal, but CRP (79.3) and procalcitonin (6.22) levels were elevated, suggesting ongoing inflammation or infection. These findings further highlight the complexity of the patient's medical condition.

Treatment

Due to the patient's complex medical picture, a multimodal management approach was adopted. End-stage renal disease necessitated the initiation of hemodialysis therapy in 2008, which continues to be the main treatment. Congenital heart disease and hepatic fibrosis require ongoing monitoring and potential medication adjustments to optimize their function. Supportive measures for hypotonia and developmental delays involve physical therapy and the use of symptom-specific medications.

A comprehensive approach was implemented for vision loss. Regular ophthalmologic evaluations are crucial for monitoring the progression of retinal degeneration and tailoring interventions accordingly. Management of the anterior and posterior blepharitis will involve topical medications or other therapeutic modalities. Depending on the severity of the visual impairment, low-vision aids could be employed to maximize the remaining visual function. Genetic counseling was offered to discuss the potential hereditary basis of the patient's condition and its implications for future offspring, facilitating informed family planning decisions.

Genetic Counselling

Joubert syndrome (JS) and related disorders typically exhibit autosomal recessive inheritance, although rare cases follow an X-linked recessive pattern. According to this genetic pattern, in order to develop the disorder, an individual must inherit two mutant copies of a gene, one from each parent. Parents who carry a single mutated copy are typically unaffected but have a 25% chance of passing it on to their offspring in each pregnancy. Prenatal diagnosis offers the possibility of early detection for JS. Chorionic villus sampling (CVS) at 11 weeks' gestation can be used to analyze fetal chromosomal and genetic material. However, a more recent and potentially more informative approach involves fetal magnetic resonance imaging (MRI) before 24 weeks' gestation. This non-invasive imaging technique can delineate posterior fossa malformations, a hallmark feature of JS, facilitating earlier diagnosis and allowing families to make informed decisions regarding pregnancy management.

Discussion

Joubert syndrome (JS) is an uncommon genetic condition specified by a distinctive combination of neurological features, including cerebellar ataxia, hypotonia, intellectual disability, and characteristic brain malformations (Spahiu *et al.,* 2022). While the primary manifestations of JS are neurological, a growing body of research suggests the potential involvement of other organ systems, including the eyes (Bachmann-Gagescu *et al.,* 2020). In 1968, Joubert *et al.* made the initial diagnosis of Joubert Syndrome (JS) in four siblings who had agenesis of the cerebellar vermis, ataxia, abnormal eye movements, episodic hyperpnea, developmental delay, intellectual disability, and a characteristic mid-hindbrain malformation identified as the "molar tooth sign." The syndrome is also associated with renal cysts and hepatic fibrosis (Spahiu *et al.,* 2022). The most common ocular findings in JS include ocular motor apraxia (80%), strabismus (74%), nystagmus (72%), ptosis (43%), chorioretinal coloboma (30%), optic nerve atrophy (22%), and pigmentary retinal degeneration (38%) (Devi *et al.,* 2020).

This case report presents a unique instance of a 20-year-old male patient with JS who also exhibited pigmentary retinal degeneration (PRD). PRD is a group of inherited disorders characterized by progressive vision loss due to the degeneration of light-sensitive retinal cells (Fenner *et al.,* 2022).

In this case, the researchers observed the presence of PRD in JS patients, highlighting this case as a distinctive presentation. Known JS subtypes associated with retinal involvement often include mutations in specific genes such as AHI1, CEP290, and TMEM67, which can contribute to both cerebellar and retinal abnormalities. The potential mechanisms linking JS and PRD involve disruptions in ciliary function, as both the cerebellum and retina rely on primary cilia for proper development and function (Amorini *et al.,* 2023). Mutations affecting ciliary proteins can thus manifest in both neurological and retinal phenotypes.

Managing such a complex case poses several challenges. The patient requires multidisciplinary care addressing neurological, renal, hepatic, and ocular issues. Regular ophthalmologic evaluations are essential to monitor the progression of retinal degeneration and manage vision loss (Menghini *et al.,* 2020). At the same time, ongoing hemodialysis and monitoring of cardiac and hepatic conditions are critical. The variability and severity of symptoms across different organ systems necessitate a coordinated approach to provide comprehensive care and improve the quality of life of the patient. Early diagnosis and intervention are important in JS to address developmental delays and organ-specific complications effectively, emphasizing the importance of a thorough and integrated diagnostic workup, including neuroimaging, genetic testing, and detailed ocular examinations (Spahiu *et al.,* 2022).

CONCLUSION

This case report emphasized the importance of early detection and management of Joubert syndrome (JS) to minimize the potential for serious complications. The co-occurrence of pigmentary retinal degeneration (PRD)

in this patient underscores the multisystemic nature of JS and the need for comprehensive evaluations that include ophthalmologic assessments. The spectrum of JS presentations can vary widely, ranging from mild ocular motility issues that may improve with age to severe retinal degeneration leading to blindness. This case report serves to raise awareness of the importance of early detection and prompt intervention in JS patients. Multidisciplinary management strategies should be implemented to address not only the neurological manifestations but also any potential complications involving the eyes, kidneys, and liver. While the prognosis for JS can be guarded and depends heavily on the severity of co-occurring complications like those observed in this case (ocular, renal, and hepatic), early diagnosis and comprehensive management can potentially optimize the quality of life for affected individuals.

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