SAS Journal of Medicine
Abbreviated Key Title: SAS J Med
ISSN 2454-5112
Journal homepage: https://saspublishers.com

OPEN ACCESS

Cardiology

Cardiac Involvement in Usher Syndrome: A Case Report

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DOI: https://doi.org/10.36347/sasjm.2024.v10i08.021 | **Received:** 15.07.2024 | **Accepted:** 23.08.2024 | **Published:** 29.08.2024

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Abstract **Case Report**

Background*:** Usher syndrome is a genetic disorder characterized by the concomitant occurrence of hearing and visual impairments. While cardiac complications in this condition are uncommon, they have been documented in the literature. ***Case Summary: In this case study, we describe a 29-year-old patient with pre-existing deafness and vision problems who subsequently developed dilated cardiomyopathy leading to acute heart failure. Transthoracic echocardiography showed biventricular dilatation and severe left ventricular dysfunction. At the 6-month follow-up, the cardiomyopathy appeared to have stabilized through a multimodal treatment approach, including the use of beta-blockers, ACE inhibitors, mineralocorticoid receptor antagonists, and SGLT2 inhibitors. ***Discussion*:** Usher syndrome is the most common cause of combined hearing and vision impairment. Some patients also experience balance issues and lack of vestibular function. The diagnosis is primarily based on the presence of congenital hearing loss and retinal pigmentation, which typically manifests in childhood or early adulthood. It is unfortunate that there are few cases in the literature of cardiac involvement in this syndrome. This makes the use of echocardiography vital for diagnosing and assessing the ejection fraction, managing heart failure episodes and enhancing treatment.

Keywords: Case report, Heart failure, Usher syndrome, ciliopathy.

INTRODUCTION

Usher syndrome is an autosomal recessive genetic disorder characterized by a combination of sensorineural hearing loss, retinitis pigmentosa, and occasionally vestibular abnormalities. This syndromic ciliopathy has been associated with a rare case of myocardial disease, which was attributed to concurrent mutations in the MYO7A and CALR genes [1].

CASE DESCRIPTION

We report the case of a 29-year-old male patient with pre-existing congenital deafness and a family history of congenital deafness and blindness in his 42-year-old sister. The patient presented to the emergency department with stage III dyspnea and lower limb edema that had been progressing for one month prior to admission.

Clinical examination revealed the patient was hemodynamically and respiratorily stable, but appeared pale and exhibited signs of global cardiac decompensation, including bilateral basal crepitation, lower limb edema extending to the thighs, an elevated jugular venous pressure, hepatojugular reflux, and ascites.

The ECG showed a regular sinus tachycardia at 120 bpm with narrow QRS-LVH (Fig 1).

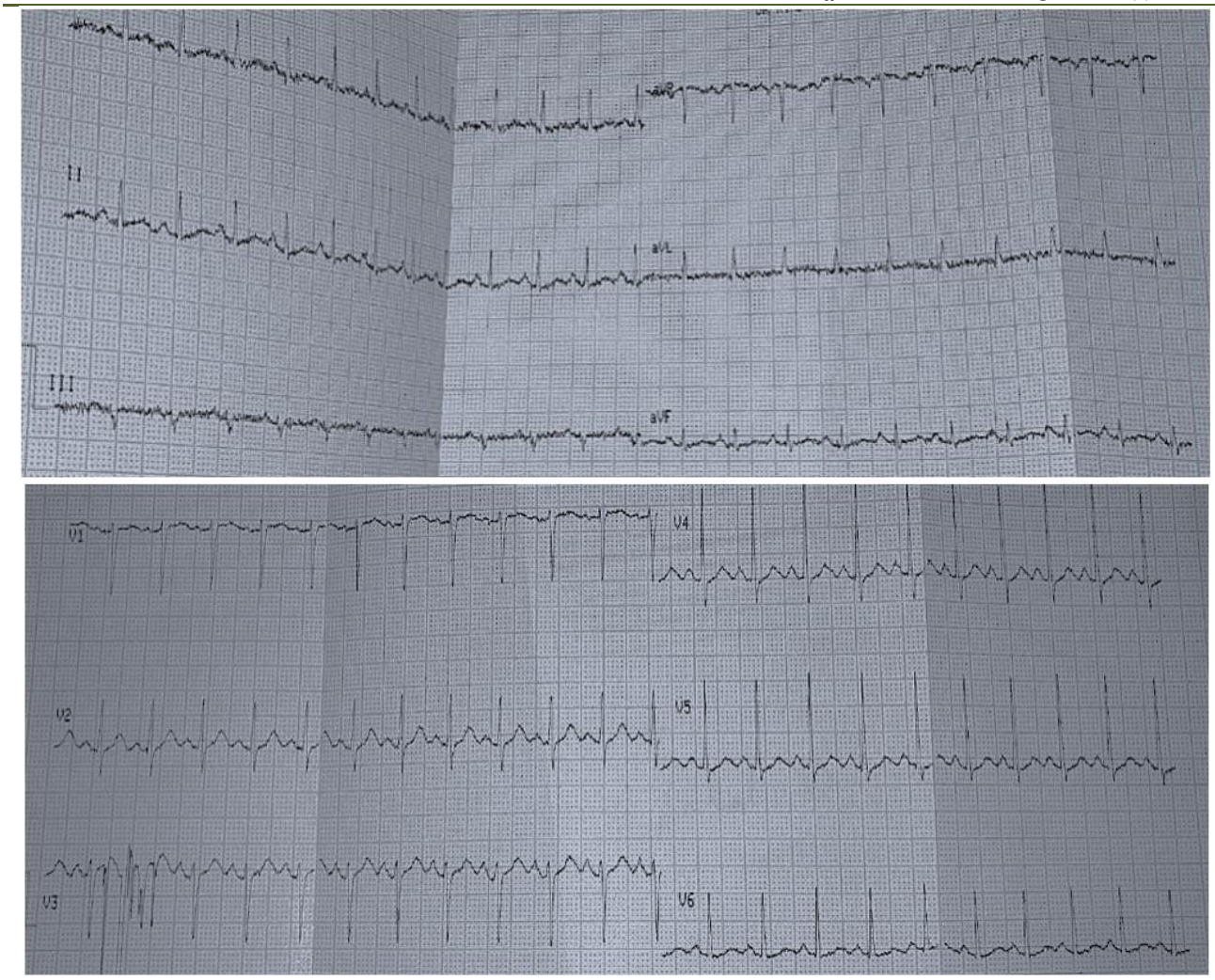

Fig 1: Narrow QRS sinus tachycardia with LVH

The transthoracic echocardiogram showed a dilated and hypokinetic left ventricle with severe dysfunction. Left ventricular ejection fraction (LVEF) was 20-25% and end-diastolic/end-systolic dimensions (EDD/ESD) were 73/62 mm.

Additionally, a restrictive profile with moderate mitral regurgitation was observed, along with dilated and echo-free atria, a dilated non-hypertrophied right ventricle with preserved function, a SPAP at 46 mmHg, and a small pericardial effusion. The global longitudinal strain (GLS) value was -7% (Fig 2).

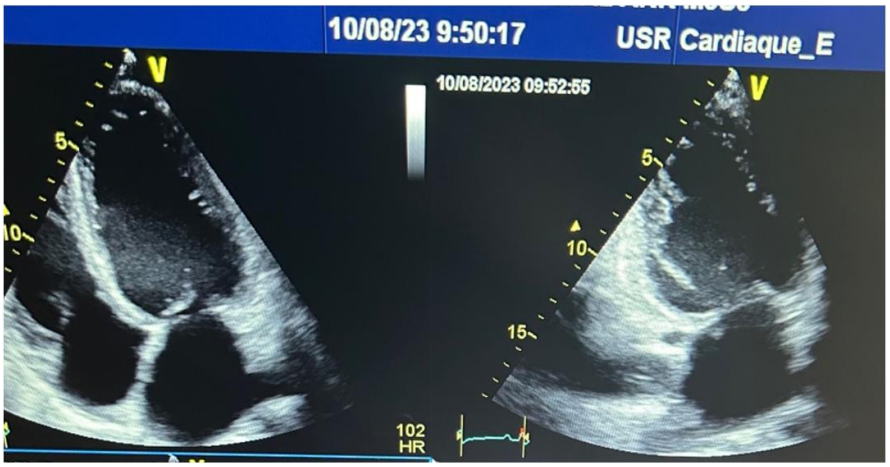

Figure 2a: TTE showing a dilated LV (A Four and Two chamber views)

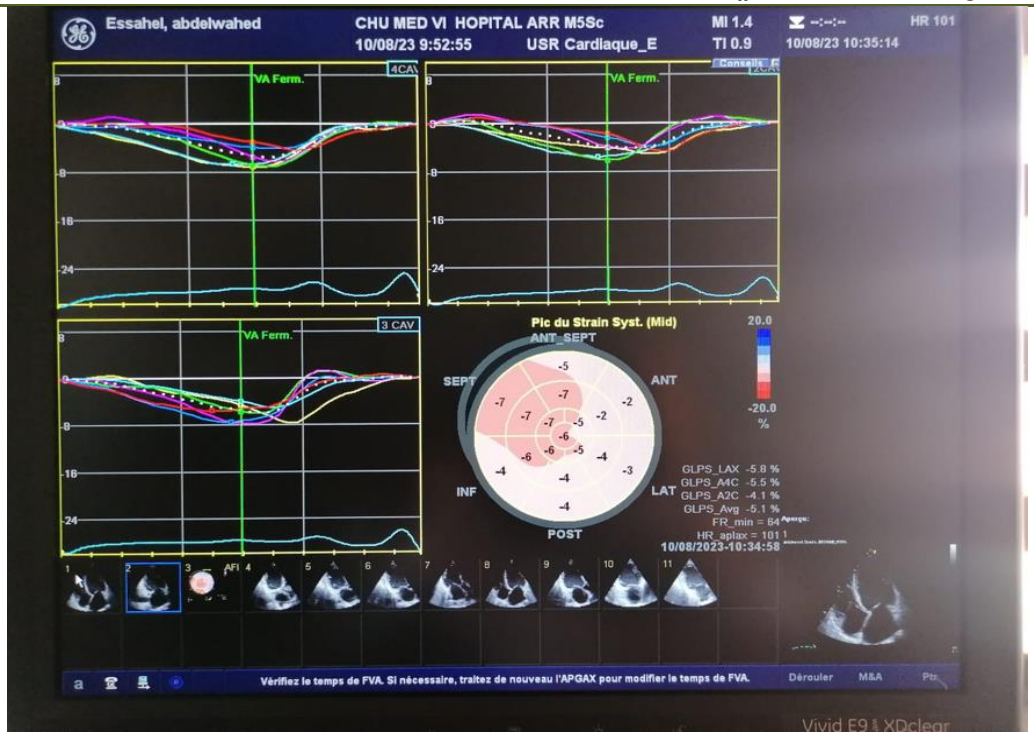

Figure 2b: TTE showing an altered GLS

Biological findings: microcytic hypochromic anemia at 7.5 g/dl; hepatic cytolysis with SGPT/SGOT at 20 times normal, low prothrombin level at 27% and GFR at 57 ml/min/1.73 m2.

As part of the etiological work-up for CMD: HIV/HBV/HCV serological test /syphilitic serology were negative, with negative anti-nuclear and anti-native DNA antibodies; thyroid, phospho-calcium, Lactate/pyruvate ratio laboratory tests were normal.

Due to the patient's congenital deafness and family history, Usher syndrome was suspected. An audiogram was performed, which revealed bilateral sensorineural deafness. Additionally, a fundoscopic exam showed bilateral retinitis pigmentosa, including filamentous vitreous with diffuse arterial narrowing and peripheral osteoblasts (Figures 3 and 4).

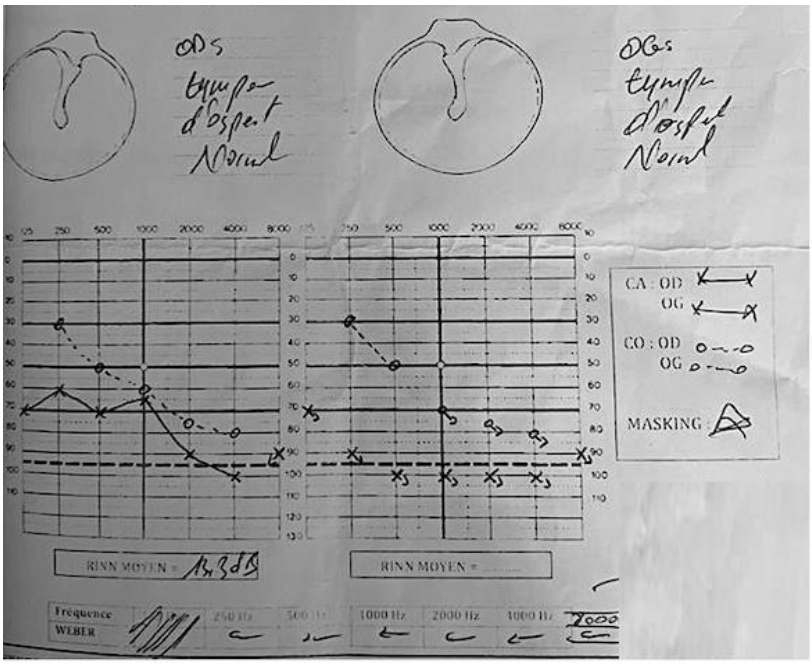

Figure 3: Audiogram revealing bilateral sensorineural deafness

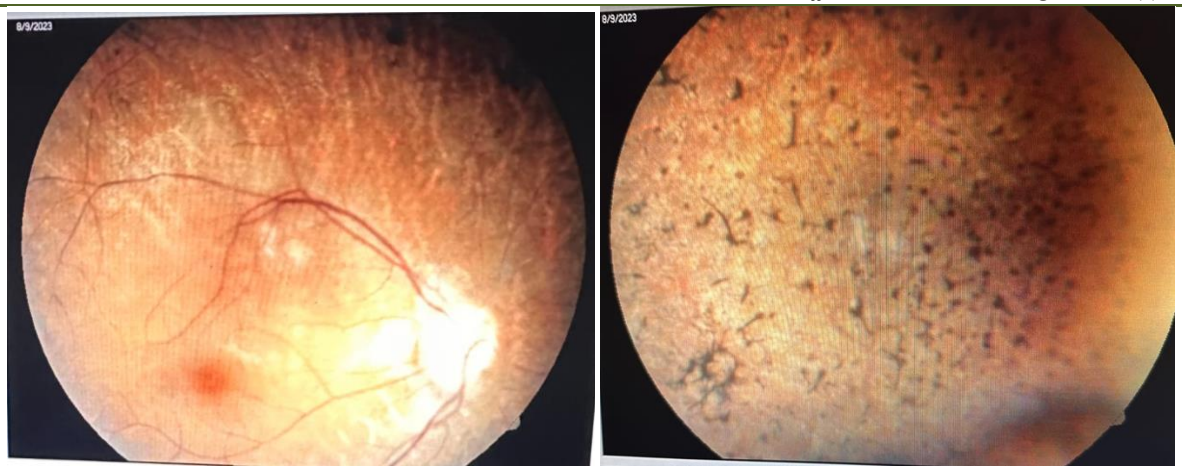

Figure 4: Fundoscopic exam showing bilateral retinitis pigmentosa, filamentous vitreous with diffuse arterial narrowing and peripheral osteoblasts

Unfortunately, genetic sequencing was not possible due to the absence of a sequencing panel in our hospital.

The patient was treated with diuretics, intravenous iron supplementation, and transfusion. After decongestion, they were put on heart failure quadri-therapy (ACEi, Betabloquer, MRA, and SGLT2i) sequentially, with therapeutic optimization spread over a 3-week period until the maximum tolerated dose was reached. LV dimension and function remained stable, and dyspnea significantly regressed. No acute cardiac decompensation episodes were registered at the 6-month follow-up.

DISCUSSION

Usher syndrome is a rare genetic disorder characterized by hearing and vision loss [5]. It is the most common cause of combined hearing and vision impairment. Some individuals with Usher syndrome also experience balance issues and lack of vestibular function. The diagnosis of Usher syndrome is primarily based on the presence of congenital hearing loss and retinal pigmentation, which typically manifests in childhood or early adulthood. Currently, no subtype of Usher syndrome involves late-onset hearing or vision loss [3].

Usher syndrome is traditionally classified into three main subtypes (USH1, USH2, and USH3) based on the severity and timing of hearing loss, the presence or absence of vestibular dysfunction, and the age at which retinitis pigmentosa begins [5].

Cardiac involvement in Usher syndrome is rare, with only one reported case in the literature. This case involved a unique form of dilated cardiomyopathy resulting from concurrent mutations in the MYO7A and CALR genes [2], which led to cardiomyocyte disconnection and mitochondrial dysfunction. The affected genes are believed to impact the sites where cardiomyocytes connect and the mitochondria, resulting in left ventricular dilation and dysfunction, ventricular arrhythmias, and mitral valve prolapse with annular disjunction due to hypotonia and dyssynergy of the papillary muscles. This differs from both idiopathic dilated cardiomyopathy and left ventricular non-compaction cardiomyopathy [2].

CONCLUSION

Usher syndrome is a genetic disorder that presents with the concomitant occurrence of hearing and visual impairments. While cardiac complications in this condition are rare, they have been documented in the literature. Therefore, early screening of this disease can lead to a more favorable outcome and the prevention of future complications.

Consent: The authors confirm that written consent for submission and publication of this case report including images and associated text has been obtained from the patient in line with the COPE guidelines.

Conflict of interest: None declared.

Funding: None declared.

Learning Points

- Cardiac involvement in Usher syndrome is rare
- Echocardiography is vital for assessing the ejection fraction, managing heart failure episodes and enhancing treatment.
- Early screening of this disease can lead to a more favorable outcome and the prevention of future complications.

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