Scurvy-induced severe pulmonary hypertension in a child with autism spectrum disorder: A rare and reversible cause

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SUMMARY

Severe pulmonary hypertension (PHT) in children is rare but potentially life-threatening, with diverse aetiologies. Clinical presentation is often non-specific and may be overlooked, particularly in children with neurodevelopmental disorders. A case is reported of an autistic boy with avoidant restrictive food intake disorder (ARFID) who presented with severe PHT secondary to scurvy which showed complete reversibility following vitamin C replacement.

INTRODUCTION

Paediatric PHT caused by Vitamin C deficiency (scurvy) is rare but may occur in children with highly restrictive dietary patterns.[1,2] Scurvy impairs endothelial function and promotes pulmonary vascular remodelling, which may culminate in severe PHT.[3] Although uncommon, micronutrient deficiencies should be considered early in the diagnostic work-up, particularly in patients with neurodevelopmental disorders and restrictive dietary patterns as described in the patient with autism spectrum disorder (ASD).[4]

CASE PRESENTATION

A 7-year-old boy with autism spectrum disorder presented with symptoms of gastroenteritis and features of right-sided heart failure. He adhered to a highly selective diet devoid of fruits and vegetables, and demonstrated failure to thrive (weight 18 kg, <3rd percentile; height 126 cm, 25th–50th percentile). Initially, he required multiple fluid boluses, inotropic support for hypotensive episodes, and non-invasive respiratory support for respiratory distress.

On examination, he was receiving continuous positive airway pressure with a positive end-expiratory pressure of 5 cmH_2O and a fraction of inspired oxygen of 60%, blood pressure of 100/60 mmHg and pulse rate of 120 bpm. He had bilateral pedal oedema, raised jugular venous pressure, and hepatomegaly extending 4 cm below the right costal margin. A left parasternal heave and a loud P2 were present, with bilateral lung crepitations on auscultation.

Transthoracic echocardiography demonstrated dilated right-sided chambers with evidence of severe PHT, severe tricuspid regurgitation (estimated right ventricular systolic pressure 62 mmHg), impaired right ventricular systolic function (TAPSE 13 mm), and preserved left ventricular ejection fraction (79%). With a history of improper dietary intake and clinical suspicion of scurvy, vitamin C was tested and reported a markedly reduced serum ascorbic acid level (<0.1 mg/dL; Ref: 0.4–2.0 mg/dL). Notably, the patient lacked the classic dermatological or musculoskeletal hallmarks of scurvy. Other medical conditions causing PH were excluded. He was commenced on oral sildenafil and therapeutic-dose vitamin C.

At the six-week follow-up, the patient was clinically well, with significant improvement in PHT and normalisation of right ventricular geometry and systolic function. Sildenafil was discontinued, while vitamin C supplementation was continued.

DISCUSSION

This case highlights a rare but important example of reversible severe PHT secondary to nutritional deficiency. Although uncommon and frequently overlooked, scurvy represents a clinically significant cause of PHT and may be fatal if unrecognised and untreated.[1-2] While often dismissed as a historical relic, scurvy remains a clinically relevant entity in the modern era. In regions where food fortification and nutritional supplementation are widespread, the diagnosis is frequently unanticipated, leading to significant under-detection and diagnostic delays.[5]

Scurvy has been described as 'the great mimicker' as its clinical manifestations are highly variable. Presentations may be as non-specific as constitutional symptoms such as fatigue and loss of appetite. Some patients exhibit cutaneous or haematological manifestations, whilst the vast majority present with musculoskeletal symptoms, reported in 92% of patients.[6] Scurvy may contribute to the development of PHT through several mechanisms, most notably impaired endogenous endothelial nitric oxide production, coupled with dysregulated pulmonary vascular remodelling — including hypoxia-inducible factor-mediated pathways — resulting in diminished opposition to pulmonary vasoconstriction.[3] Cardiopulmonary manifestations have been reported to range from exertional fatigue to cardiopulmonary collapse secondary to right-sided heart failure in the context of pulmonary hypertension.[2,6]

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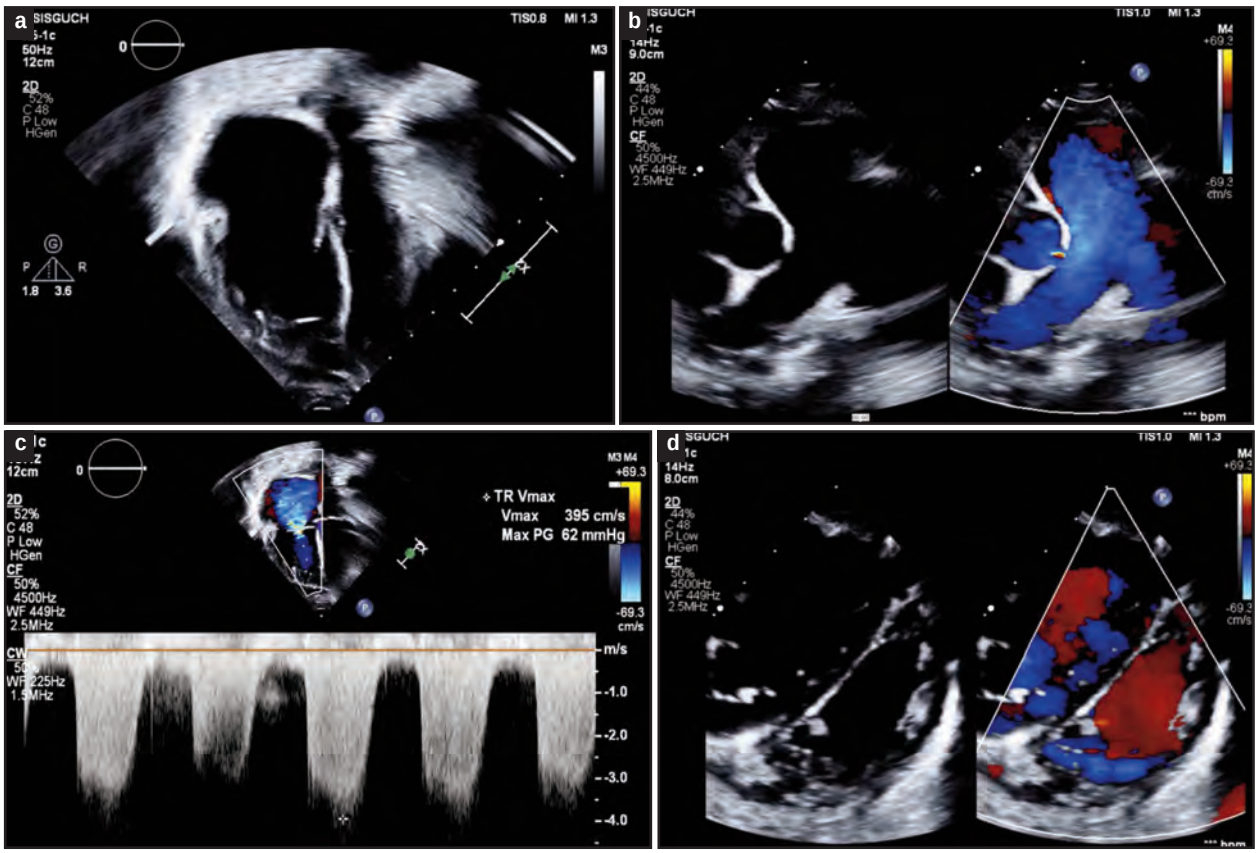

Fig. 1: Pre-treatment Echocardiogram; (a) Dilated right-sided chambers (b) Dilated pulmonary arteries (c) Severe tricuspid regurgitation with TR of 62mmHg (d) Interventricular septal flattening

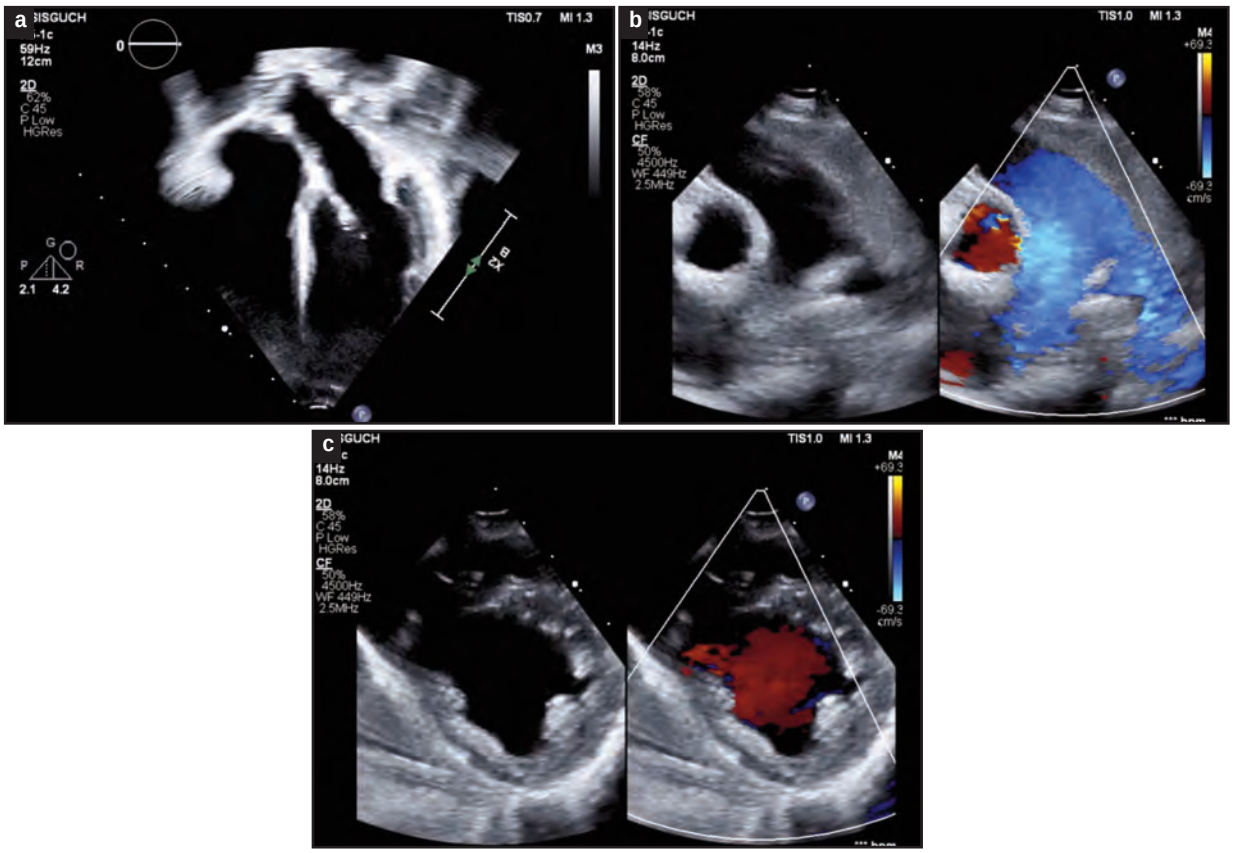

Fig. 2: Post-treatment Echocardiogram; (a,b) Right ventricle and pulmonary artery were much less dilated (c) Restored circular left ventricle

The clinical presentation in our case mirrors the findings reported by Satawiriya et al., characterised by decompensated heart failure and episodes of hypotension secondary to severe PHT.[2] Notably, the present patient lacked the hallmark mucocutaneous and musculoskeletal manifestations of scurvy, such as arthralgia, spontaneous haemorrhage, or anaemia which were present in previously documented cohorts.[1-2,6] This underscores that pulmonary vascular dysfunction can emerge as a sentinel or even isolated manifestation of ascorbic acid deficiency, preceding the more widely recognised classic signs of scurvy.

Comprehensive reviews indicate that the narrow dietary variety common in children with ASD significantly elevates the risk of micronutrient deficiencies; notably, scurvy is a recognised sequela of avoidant restrictive food intake disorder (ARFID) in this high-risk group.[7] ARFID, recently introduced in the Diagnostic and Statistical Manual of Mental Disorders, 5th Edition (DSM-5). It is characterised by persistently insufficient nutritional and/or energy intake and frequently co-exists with ASD, with reported prevalence ranging from 12.5% to 33%.[8] Children with ASD and co-occurring ARFID often exhibit more severe restrictive eating behaviours attributable to sensory sensitivities; such behaviours have been reported at up to 15-fold greater frequency than in typically developing children,[8] thereby heightening the risk of micronutrient deficiencies, growth faltering, and impairments in psychosocial functioning, despite adequate caloric intake.[9]

This case underscores the critical need for a systematic evaluation of secondary causes of PHT, including nutritional deficiencies, particularly in children with neurodevelopmental disorder.[10] Routine screening is strongly recommended of serum ascorbic acid, alongside other key micronutrients such as vitamin D and iron, in children presenting with PHT who exhibit selective eating patterns or are at high risk for malnutrition. Notably, scurvy-induced PHT in the paediatric population is typically transient; high-dose vitamin C supplementation achieves rapid clinical and haemodynamic resolution, thereby preventing long-term cardiopulmonary sequelae.[1-2]

Integration of comprehensive dietary and micronutrient assessments into the diagnostic algorithm for paediatric PHT is essential, particularly in children with autism and ARFID. Scurvy-induced PHT is a life-threatening yet fully reversible condition; early recognition and prompt treatment with ascorbic acid ensure complete haemodynamic resolution and prevents irreversible cardiopulmonary sequelae.

REFERENCES

1. Azar J, Ayyad M, Jaber Y, Ayasa LA. Scurvy-induced pulmonary arterial hypertension. BMJ Case Reports CP 2023; 16(4): e254730.
2. Satawiriya M, Khongphatthanayothin A, Limsuwan A. Reversible severe pulmonary hypertension related to scurvy in children. BMC Cardiovascular Disorders 2024; 24: 24.
3. Humbert M, Morrell NW, Archer SL, Stenmark KR, MacLean MR, Lang IM, et al. Cellular and molecular pathobiology of pulmonary arterial hypertension. J Am Coll Cardiol 2004; 43(12, Suppl S): S13–S24.
4. Sharp WG, Berry RC, Burrell L, Scahill L, McElhanon BO. Scurvy as a Sequela of Avoidant-Restrictive Food Intake Disorder in Autism: A Systematic Review. J Dev Behav Pediatr 2020; 41(5): 397-405.
5. Velandia B, Centor RM, McConnell V, et al. Scurvy is still present in developed countries. J Gen Intern Med 2008; 23: 1281-4.
6. Trapani S, Rubino C, Indolfi G, Lionetti P. A narrative review on pediatric scurvy: the last twenty years. Nutrients 2022; 14(3).
7. Inoue, T., Otani, R., Iguchi, T., et al. (2021). Prevalence of autism spectrum disorder and autistic traits in children with anorexia nervosa and avoidant/restrictive food intake disorder. BioPsychoSocial Medicine 15: 9.
8. Mayes SD, ZickgrafH. Atypical eating behaviors in children and adolescents with autism, ADHD, other disorders, and typical development. Research in Autism Spectrum Disorders 2019; 64: 76-83.
9. Bandini LG, Anderson SE, Curtin C, Cermak S, Evans EW, Scampini R, et al. Food selectivity in children with autism spectrum disorders and typically developing children. J Pediatr. 2010; 157(2): 259-64.
10. Humbert M, Kovacs G, Hoeper MM, Badagliacca R, Berger RMF, Brida M, et al. 2022 ESC/ERS Guidelines for the diagnosis and treatment of pulmonary hypertension. Eur Heart J 2022; 43(38): 3618-731.