





Myocardial infarction in a 17-year-old patient diagnosed with MPOD II syndrome

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Brief Report

Cite this article: Clarac C, Karila-Cohen J, and Bonnet D (2024) Myocardial infarction in a 17-year-old patient diagnosed with MPOD II syndrome. *Cardiology in the Young* **34**: 2028–2029. doi: [10.1017/S1047951124025824](https://doi.org/10.1017/S1047951124025824)

Received: 24 May 2024

Revised: 11 June 2024

Accepted: 7 July 2024

First published online: 4 October 2024

Keywords:

Paediatric; dwarfism; microcephalic; myocardial; infarction

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Julie Karila-Cohen;

Email: julie.karila-cohen@aphp.fr**Abstract**

Introduction: Microcephalic osteodysplastic primordial dwarfism (MOPD) syndrome type 2, caused by a mutation in the PCNT gene (21q22.3), is a rare autosomal recessive disorder. Patients present with bone dysplasia, insulin resistance, kidney diseases, and cardiac malformations, making them prone to vascular diseases. Cardiomyopathy, hypertension, and coronary diseases are documented. The prognosis is associated with cerebrovascular complications. **Method:** We report a case of a patient with MOPD type II who suffered a myocardial infarction in our institution. Informed consent for publishing was obtained. **Result:** A 17-year-old female with MPOD II syndrome (20 kg and 86 cm) was referred for chest pain. Thoracic pains had been occurring for over a month, increasing in intensity, with an episode prompting emergency consultation. Initial tests revealed elevated troponin and an inflammatory response. Electrocardiogram (ECG) showed ST-segment depression and elevation. Echocardiography revealed hypokinetic inferior walls with moderate concentric hypertrophy. A coronary CT scan showed subendocardial hypodensity. Diagnostic coronary angiography revealed tri-branch lesions and almost complete stenoses or occlusions on the circumflex artery (Image). No indication for interventional treatment due to diffuse atheromatous lesions. Exclusive medical treatment was initiated. **Conclusion:** MPOD II syndrome is associated with cardiac malformations and neurovascular complications, including myocardial infarction. Regular ECG monitoring is advisable. Active surveillance for coronary diseases is necessary from adolescence. Recognising this complication allows for prompt intervention. This case highlights the need for specific monitoring and prompt management of chest pain in patients with MPOD II syndrome. Primary prevention could mitigate the occurrence of coronary events in this high-risk population.

Introduction

Microcephalic osteodysplastic primordial dwarfism (MOPD) syndrome type 2 is a rare autosomal recessive disorder characterised by a mutation in the PCNT gene (21q22.3),¹ with approximately 150 cases reported worldwide.² It represents one of the most common forms of primordial dwarfism with microcephaly.³ Commonly associated conditions include bone dysplasia often linked to scoliosis, dental anomalies, insulin resistance leading to diabetes, chronic kidney diseases, cardiac malformations, and overall vascular disease.² Cases of cardiomyopathy, hypertension, and coronary disease are described.^{2,4} The prognosis of this disease is strongly associated with cerebrovascular complications.

This article presents the case of a 17-year-old girl diagnosed with MOPD type II, who suffered an inferior myocardial infarction. Informed consent for publishing patient information and images was obtained from the patient's parents.

Case report

A 17-year-old female patient diagnosed with MOPD II syndrome was referred to our paediatric cardiology department for chest pain. Weighing 20 kg and measuring 86 cm in height, her physical characteristics were typical of the disease. Notable features included extreme microcephaly and facial dysmorphism (Figure 1), with a rounded face, short and stocky neck, retrognathia, dysplastic ear lobes, a broad and elevated nasal root, and a wide philtrum. Her surgical history included coxa vara surgery, craniosynostosis surgery, and the placement of transtympanic ventilators. She was on metformin for insulin-dependent diabetes related to her condition. She also had systemic hypertension but was not on antihypertensive therapy at admission. Cognitively, she attended a medical-social institute.

She had experienced chest pain for over a month, occurring during exertion, resolving spontaneously, and of moderate intensity. An intense episode prompted an emergency consultation. The pain was described as retrosternal, oppressive, radiating to the left arm, and associated with dyspnoea. Initial laboratory tests showed elevated troponin at 318 ng/L and an



Figure 1 Facial dysmorphism in our patient with Microcephalic Osteodysplastic Primordial Dwarfism type II, frontal (a) and side profil photography (b).

inflammatory response with a C-reactive protein of 17 mg/L. The electrocardiogram (ECG) revealed ST-segment depression in the inferior territory and ST-segment mirror elevation in precordial leads V1 to V3. Chest X-ray was unremarkable with a normal-sized heart. Echocardiography showed hypokinetic inferior and inferoseptal walls, but overall cardiac function was preserved with a normal ejection fraction. The left ventricle displayed moderate concentric hypertrophy. A previous echocardiography showed a slightly hypertrophic left ventricle without other abnormalities.

A coronary CT scan revealed subendocardial hypodensity suggestive of ischaemic injury sequelae in the inferolateral territory. Follow-up laboratory tests showed a troponin increase, reaching a maximum of 7600 ng/L within 12 hours of the initial measurement. Diagnostic coronary angiography revealed tri-branch lesions and highlighted a diminutive left and right coronary network with almost complete stenoses or occlusions staggered along the circumflex artery (Figure 2). There was no indication for interventional treatment due to diffuse atheromatous lesions. Medical management was exclusively initiated, including dual antiplatelet therapy, management of cardiovascular risk factors such as dyslipidemia, and cardioprotective treatment.

Discussion

The primary cardiac malformations observed in MPOD II syndrome include persistent patent foramen ovale and atrial or ventricular septal anomalies, affecting approximately 25% of patients.⁵ Neurovascular complications are frequently documented, with recent reports including a case of moyamoya syndrome.⁶ Duker et al.⁷ reported on a cohort of 47 MPOD type 2 patients: eight of whom were diagnosed with myocardial infarction in adulthood, with a median age of 24 years (ranging from 17 to 33 years). Some individuals experienced multiple coronary events. A separate case report highlighted a brother and sister with the syndrome who suffered a myocardial infarction in adolescence.⁸ Despite the infrequency of coronary syndrome at this age, regular ECG monitoring is advisable. Vascular monitoring, particularly blood pressure monitoring at the cardiac and renal levels, is recommended due to potential complications associated with MOPDII syndrome, with particular attention to the risk of myocardial infarction approaching adulthood.⁶ The literature data underscores the necessity for active surveillance of coronary

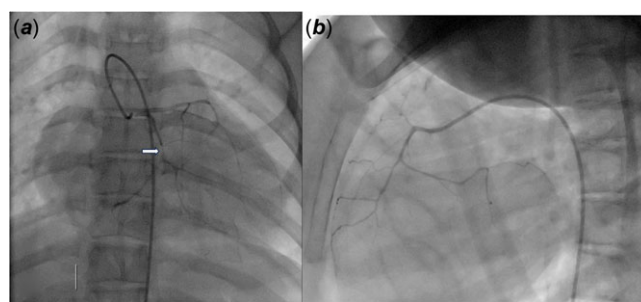


Figure 2 Coronary angiography (using a four French catheter) revealing tri-branch lesions. (a) Contrast injection in left coronary trunk: tiny left coronary network with almost complete occlusions staggered on the circumflex artery over 2,6 mm (arrow). (b) Contrast injection in the right coronary: tiny right coronary network.

diseases from adolescence onward, given that these patients share the same risk factors as the general population with coronary issues. Implementing primary prevention strategies for cardiovascular disease could mitigate the occurrence of these coronary events. Recognising this complication in the disease allows for prompt intervention at the onset of initial clinical signs.

Conclusion

This case underscores the need for specific monitoring and prompt management of chest pain in patients with MOPD II syndrome. The patient was diagnosed with myocardial infarction and received medical treatment. Primary prevention should be considered in this high-risk population for coronary artery disease.

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