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CASE REPORT

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Is Gorlin–Chaudhry–Moss syndrome associated with aortopathy?

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Abstract

Gorlin–Chaudhry–Moss syndrome (GCMS) is a rare disorder consisting of craniofacial dysostosis, hypertrichosis, underdeveloped genitalia, and ocular and dental anomalies. Recently, GCMS has been reclassified together with Fontaine syndrome as Fontaine progeroid syndrome (FPS), after a common genetic basis was found. It was previously thought that GCMS/FPS was not associated with aortopathy, but in recent years 3 patients with aortic disease have been described. We describe the fourth case, who is the oldest patient with GCMS/FPS reported in the medical literature: a 45-year-old patient who presented with acute aortic dissection. We therefore recommend screening patients previously diagnosed with GCMS/FPS for aortic pathology to aid early detection and avoid patient presentation in an acute setting.

Keywords: Gorlin–Chaudhry–Moss syndrome • Fontaine progeroid syndrome • Aortopathy dissection

INTRODUCTION

Gorlin–Chaudhry–Moss syndrome (GCMS) is a rare disorder consisting of craniofacial dysostosis, hypertrichosis, underdeveloped genitalia, and ocular and dental anomalies [1]. It has thus far not been associated with aortopathy. Ehmke *et al*. [2], who demonstrated that the syndrome is caused by recurrent mutations affecting the 217th amino acid in SLC25A24, did describe 2 GCMS patients with aortic ectasia. The same mutation is associated with Fontaine syndrome [3]. These syndromes show overlapping clinical features but have different life expectancies: patients with Fontaine syndrome do not survive beyond a year, whereas patients with GCMS are assumed to have a normal life expectancy. The cause of the different life expectancies is still unclear. Fontaine syndrome and GCMS, due to their common genetic basis, have been reclassified as Fontaine progeroid syndrome (FPS, OMIM: Fontaine progeroid syndrome; https://www.omim.org/entry/612289). A 15-year-old male with FPS has since been described with aortic dilatation. We present the case of a 45-year-old woman, with GCMS, who presented with an acute type A aortic dissection. Written consent was obtained from the patient.

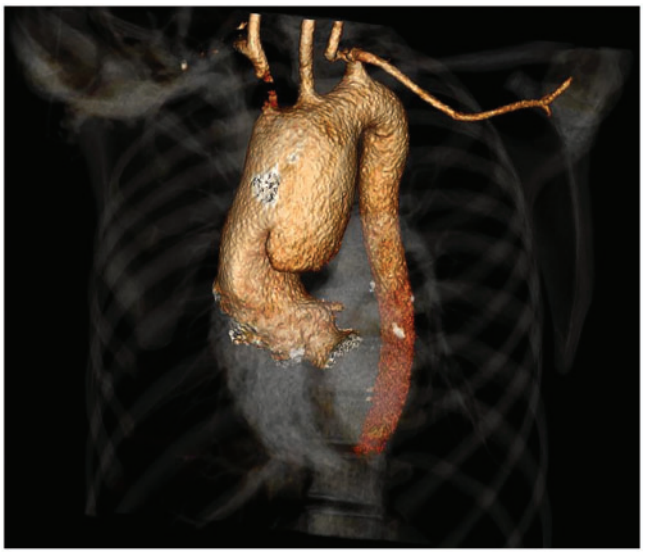

Figure 1: Three-dimensional reconstruction of the aorta.

CASE

A 45-year-old female, diagnosed with GCMS in childhood, displays typical features of the syndrome (Table 1). The patient had an otherwise unremarkable medical history. She was admitted to the hospital with sharp pains behind the sternum, that had been present for 10 days. The patient was in stable haemodynamic and respiratory conditions. Thoracic computed tomography (CT) showed an aneurysm of the ascending aorta and the proximal part of the aortic arch, with a maximal diameter of 51 mm (patient length 142 cm, weight 31 kg). Proximally, the aneurysm had a saccular aspect and a mural thrombus (Fig. 1). The patient was

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Table 1: Patient features of GCMS

Short stature
Orofacial findings
• Brachycephaly
• Midface hypoplasia
• Craniosynostosis
• Microphtalmia with impaired vision
• Downslanting palpebral fissures
• Narrow palpebral fissures
• Parietal alopecia
• Low frontal hairline
• Coarse hair and hypertrichosis
• High arched narrow palate
• Small prominent ears
Extremities
• Nail hypoplasia
• Bilateral distal phalangeal hypoplasia of the 4th–5th finger
• Cutaneous syndactyly of 4th–5th toes of the left foot
Hyperopia
Hypo- and microdontia
Conductive hearing loss.
Labia majora hypoplasia
Umbilical hernia

GCMS: Gorlin–Chaudhry–Moss syndrome.

transferred to a tertiary medical centre where she underwent urgent surgery.

Inspection revealed an aneurysmatic dilatation with a local partially chronic and partially acute dissection with fresh thrombus. A supracoronary ascending aorta and a zone 2 partial arch replacement (with reimplantation of brachiocephalic and left common carotid artery as an island) was performed using 18-mm Hemashield protheses. Surgery was performed under deep hypothermia, circulatory arrest, and bilateral antegrade cerebral perfusion.

The postoperative course was complicated by temporary respiratory distress caused by pulmonary oedema and pneumonia, which was treated with antibiotics. The patient was discharged from the hospital 2 weeks after surgery in a good clinical condition. Regular follow-up, including thorax CT, will occur in an outpatient setting.

Histopathological analysis on the resected aortic tissue showed an aortic dissection with fibrosis but no signs of connective tissue disorders (no cystic media necrosis) or vasculitis.

Genetic analysis revealed the recurrent mutation c.650G>A (p.Arg217His) in the SLC25A24-gene, confirming the diagnosis of FPS.

COMMENTS

This case report describes the oldest patient with GCMS in the medical literature. The 2 patients first described with GCMS were again described several years later, at 36 and 34 years of age, along with 2 new patients, including a 33-year old [1, 5]. No information was provided on whether these patients had aortic abnormalities. Other patients described were either children or adolescents. Ehmke *et al.* [2, 4] were the first to report 2 patients with aortic ectasia and a later case described a 15-year-old male with FPS and aortic dilatation. We now describe a 45-year-old patient with an ascending aorta aneurysm and a local dissection who underwent urgent surgery. Due to the small patient population, the association between the syndrome and aortopathy is difficult to prove but 4 patients, including our patient, have recently been described with aortic disease [2, 4]. We would therefore recommend screening patients previously diagnosed with GCMS/FPS for aortic disease.

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Conflict of interest: none declared.

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