CASE REPORT

Progressive supra-aortic stenosis in a young adult with the findings of Singleton Merten Syndrome

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SUMMARY

Singleton Merten Syndrome is an autosomal dominant disorder of unknown origin. Patients often present with muscular weakness, failure to thrive, abnormal dentition, glaucoma, psoriatic skin lesions, aortic calcification and musculoskeletal abnormalities. In this case, we present a young girl with a history of aortic root replacement, who had an unusual progressive supra-aortic stenosis managed with urgent surgery during the course of the syndrome. Cardiovascular involvement needs special attention, since it is the major cause of mortality along with rhythm disturbances in the course of Singleton Merten Syndrome.

BACKGROUND

Singleton Merten Syndrome (SMS) is a genetically inherited syndrome of unknown origin.[1] It is an autosomal dominant disorder with variable expression of the symptoms with an onset during childhood. In this case, we would like to present a young girl with findings of SMS who had a history of operation for valvular aortic stenosis 3 years prior to admission to our clinic. We performed the replacement of the ascending aorta for severe and progressive calcification leading to critical supra-aortic stenosis.

CASE PRESENTATION

An 11-year-old girl was admitted to our hospital with recurrent aortic stenosis and syncope attacks. For the past 3 days, she had been experiencing attacks of dizziness, vertigo, deviation of the eyes and a presyncope-like situation with jerking and contraction of her arms, which lasted for a couple of minutes. Three years prior to her admission,

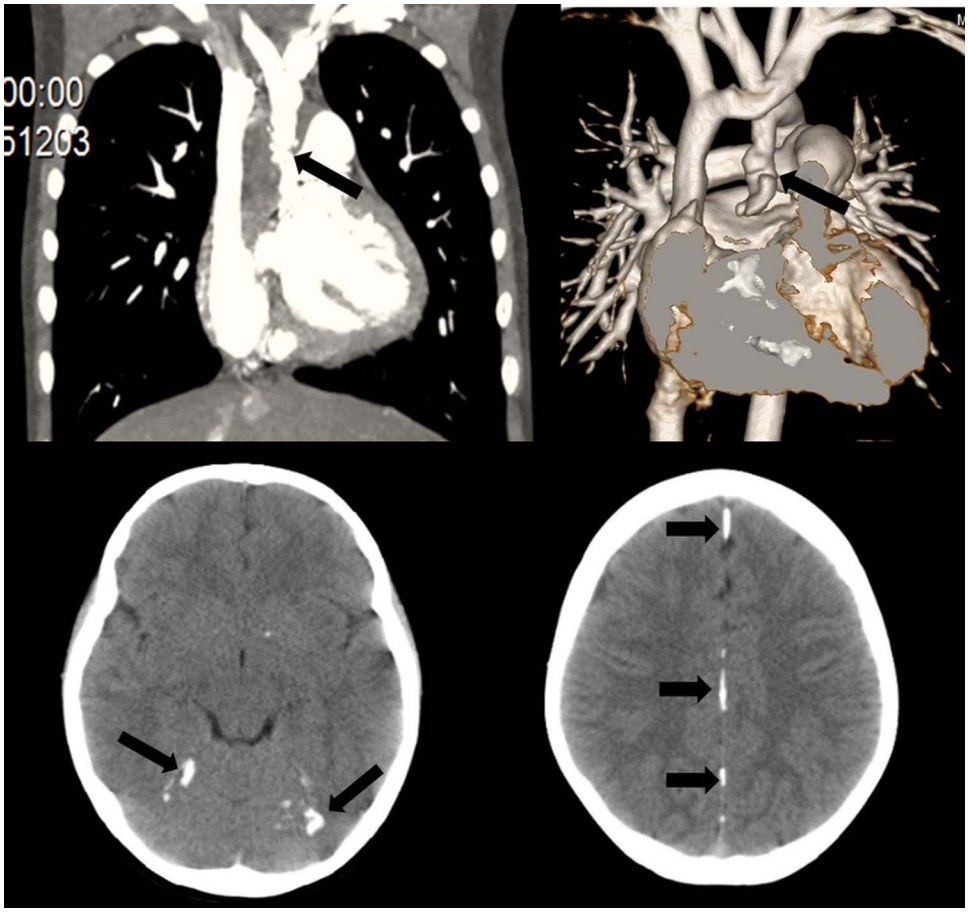

Figure 1 Critical supra-aortic stenosis at the ascending aorta (upper left side, arrow) at CT. Irregularity of the aortic wall and a possible haematoma or dissection formation can be identified at the MRI (upper right side, arrow). Scattered calcifications at the tentorium cerebelli and falx cerebelli at cranial CT are demonstrated in the lower images (arrow heads).

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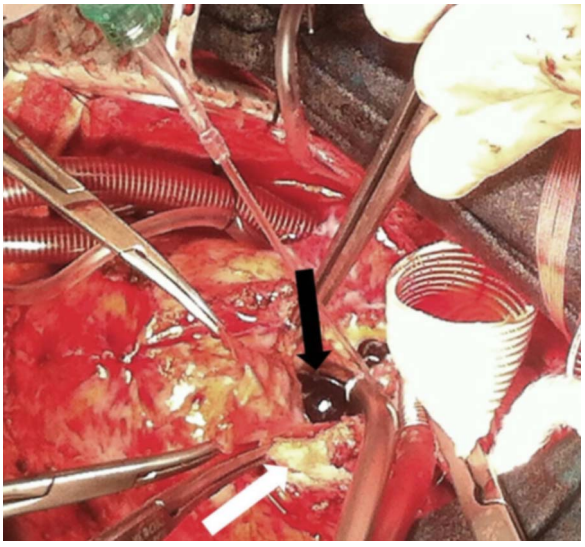

Figure 2 Operative view indicating the severe calcifications and dissection formation at the ascending aorta (white arrow). The previously replaced mechanical valve was functioning normally (black arrow).

anterior aortic root enlargement procedure (Konno-Rastan operation) and replacement of the aortic valve with a 17 mm mechanical valve had been performed. The aortic valve had been reported to be at bicuspid morphology at that time. She had been operated for glaucoma five times. Her parents reported easy cavity formation in her teeth. Her incisor teeth were abnormally developed with double line including caries. Her physical examination revealed systolic murmur at the right side of the sternum. Neurological examination was normal except for vertical nystagmus at the left eye and weakness of the lower extremity muscles. Transthoracic echocardiography revealed a supravalvular critical aortic stenosis with a gradient of maximum 90 mm Hg and a mean of 55 mm Hg. Mechanical aortic valve was functioning normally with a mild stenosis. Mitral valve leaflets were calcified and a mild mitral insufficiency and stenosis were noted. Cardiac MRI and CT were performed and severe calcification at the ascending aorta leading to a critical stenosis with a 7 mm passage at the narrowest point was noted (figure 1). Cranial CT was performed, which revealed severe calcifications at tentorium and falx cerebelli and bilateral punctate calcifications at the basal ganglia (figure 1). Her total and ionised blood calcium levels were 9.6 mg/dL and 1.16 mmol/L respectively. An urgent operation was scheduled. Re-sternotomy was performed following femoral arterial cannulation. Cardiopulmonary bypass (CPB) was established and low flow antegrade cerebral perfusion was initiated at 20°C. Aortotomy was performed at the ascending aorta, which was heavily calcified with a pin hole antegrade flow (figure 2). The ascending aorta was replaced with a 22 mm graft. The patient was weaned successfully from CPB on sinus rhythm. The postoperative course was uneventful, but episodes of severe bradycardia attacks without any finding of convulsions were observed. Although the patient was on sinus rhythm, on the 14th postoperative day a transvenous permanent pacemaker was implanted. The patient was subsequently discharged and was clinically doing well, without any symptoms, in the first year after surgery.

OUTCOME AND FOLLOW-UP

The patient is free of symptoms in the first year after surgery.

DISCUSSION

SMS is a very rare clinical entity that is thought to be a disorder of the calcium metabolism within a disturbed immune system.[2] At its first description, the symptoms of abnormal dentition, distal limb osteoporosis, marked calcification and intimal weakening of the aortic arch and valve were mentioned, which were all present in our patient. Besides these landmarks of the syndrome, our patient had glaucoma, psoriasis in the lower extremities, muscle weakness and atypical facial appearance. Vertical nystagmus and preoperative convulsions were probably related to central nervous system involvement, which included calcifications at the basal ganglion and tentorium cerebelli. Cardiac involvement and impaired cardiac function had been mentioned in a patient with aortic root replacement using pulmonary autograft in the literature.[3] To the best of our knowledge, this is the only case in the literature with aortic root enlargement due to severe valvular stenosis, followed by graft interposition of the ascending aorta for critical supra-aortic stenosis diagnosed 2 years after the first surgical intervention in a patient with SMS. The development of severe supra-aortic stenosis and recent central nervous system involvement may imply the progression of the disease in the puberty period.

Learning points

- Genetically inherited disorders of calcium metabolism may present with severe cardiovascular involvement.
- Urgent cardiovascular surgical interventions may be necessary in the natural course of these diseases.
- Multidisciplinary management of the acute exacerbations is mandatory in order to control the clinical outcome of complications.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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