On-line Case Report

Turner syndrome – an accidental finding?

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ABSTRACT

We report the case of a 9-year-old girl with bilaterally reduced pedal pulses following emergency surgery for unilateral subtrochanteric femoral fracture. This incidental postoperative finding of differential blood pressures between the upper and lower limbs led to the diagnosis of aortic coarctation and subsequent karyotype analysis confirmed the rare diagnosis of Turner syndrome.

Keywords: Turner syndrome – Aortic coarctation – Differential blood pressures

Turner syndrome is a genetic disorder of girls with specific physical features and reportedly high incidence of congenital cardiovascular abnormalities, especially that of aortic coarctation. Despite these clinical findings, the diagnosis of Turner syndrome is often delayed because of lack of suspicion.

We report the case of an incidental finding of aortic coarctation and subsequent diagnosis of Turner syndrome, following trauma surgery.

Case report

In 2004, a previously well 9-year-old girl was admitted through the emergency department having ridden her bicycle into a moving van. The patient sustained fracture of the right femur.

On admission, the patient was alert, orientated and haemodynamically stable with regular pulse of 112 beats/min and blood pressure of 105/45. On examination, no head, spine, chest, abdominal or pelvic injuries were found. Painful swelling to the right thigh was seen, with no breach of the skin. Pedal pulses were found to be present bilaterally, with no neurovascular deficit distally. X-rays revealed a displaced subtrochanteric fracture of the right femur. The patient remained comfortable in skin traction for

management of her femoral fracture until the time of surgery.

The patient underwent open reduction and internal fixation of the right femur with a blade plate.

The surgery had been uneventful and she required an intra-operative blood transfusion and fluid replacement for estimated losses of only 800 ml. At the end of the procedure, the patient was noted to be pale and cool, with poor capillary refill. No peripheral pulses were palpable in either lower limb below the femorals. Vascular surgical assessment in theatre noted poor Doppler signals at the popliteal, dorsalis pedis and posterior tibial pulses, to our shock, symmetrically. There was, however, good capillary refill found.

On the first postoperative day, despite haemodynamic stability, normal distal lower limb movements and sensation, and capillary refill time of < 2 s bilaterally, the dorsalis pedis and posterior tibial pulses remained difficult to palpate.

On review, the vascular surgeons noted lower limb pressures of 85–90 mmHg bilaterally compared with upper limb pressures of 120 mmHg, suggestive of aortic coarctation.

Angiogram of the aorta revealed coarctation of the aorta just distal to the left subclavian artery (Fig. 1). Given these

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Figure 1 Coarctation distal to left subclavian artery.

findings, she was referred to a paediatric cardiologist who found features of short in stature (< 0.4th centile for height) with neck webbing, increased carrying angle, pectus carinatum and wide spacing of the nipples, suggestive of Turner syndrome on examination. Cardiovascular examination revealed soft ejection systolic murmur. No cardiac abnormalities were noted on echocardiogram. Abdominopelvic ultrasound showed normal kidneys and reproductive organs.

Given a clinical diagnosis of Turner syndrome, the patient was referred on to the genetic service and chromosome analysis, revealed mosaic Turner syndrome.

Discussion

Turner syndrome is a disorder of girls who are found to have an abnormal or missing X chromosome on karyotyping. Clinically, these patients have features of short stature (height < 5th percentile), delayed puberty, congenital lymphoedema and dysmorphic features of webbed neck, nail dysplasia, high palate and short fourth metacarpal.¹

The prevalence of various cardiovascular malformations in patients with Turner syndrome have been reported between 17–45%.^{1,2}

Sybert² found cardiovascular abnormalities in 56% of patients with karyotype-proven Turner syndrome. This study reported 71% of these to be structural defects and 29%

functional. Coarctation of the aorta, a discrete aortic narrowing distal to the left subclavian artery, is the most common cardiac malformation reported in Turner syndrome.²

Pathognomonic clinical features of aortic coarctation are differential blood pressures between the upper and lower extremities with systolic hypertension in the upper limbs and diminished or absent femoral pulses,³ as described in this case.

The presence and quality of peripheral pulses to the lower leg in the general population is variable, especially the presence of the anterior tibial artery. The posterior tibial and peroneal vessels are however invariably present.

Robertson *et al.*⁴ found the incidence of impalpable pedal pulses in the general population to be 4.7% and, when also absent on Doppler examination in the paediatric population, to be an indicator of underlying congenital abnormality.

The absence of pedal pulses is, however, valueless in the diagnosis of Turner syndrome, as the degree of severity of aortic coarctation cannot be correlated with presence of lower extremity pulses, which result from variation in collateral blood supply, unless corroborated by other physical signs.⁵

Despite specific physical findings of short stature in the presence or absence of lymphoedema and web neck, the diagnosis of Turner syndrome is, however, often delayed in many affected girls, as in this case.^{1,3} This is because, although these clinical findings are frequent in patients with Turner syndrome, they occur relatively infrequently in the general population.

In the case presented, the diagnosis of Turner syndrome was made only after the unexpected postoperative finding of reduced pedal pulses, bilaterally.

This case highlights the need to consider the possibility of a secondary diagnosis in patients presenting with a seemingly unrelated problem, and that the finding of a possible postoperative complication may be explained as a feature of a congenital, dual diagnosis.

This also demands the assessment of the patient as a whole for specific physical features, not only of their primary problem. Retrospectively, Turner syndrome may have been suspected from physical features present in this case, especially on the finding of aortic coarctation, before confirmation with special cardiovascular investigations and chromosome analysis.

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