Presentation of coarctation of the aorta in the neonates and the infant with short and long term implications

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Abstract

Coarctation of the aorta (CoA) can present with acute circulatory collapse during the neonatal period or in early infancy. This review article aims to cover the presentation and management of CoA; including the common signs and symptoms, pathophysiology, medical and surgical management. We will not cover the features of the disease presentation in older children or adults. A high index of suspicion of CoA is extremely important in infants presenting with circulatory collapse in the first 6–8 weeks of life. Decreased femoral arterial pulse volume as compared to the right brachial artery, raised upper limb blood pressure as compared to lower limb and differential cyanosis should raise suspicion of coarctation in an infant presenting with circulatory collapse. Prostaglandin (PGE1 or PGE2) infusions should be started as soon as possible to restore forward circulation.

Keywords coarctation of aorta; neonatal coarctation of aorta

Definition

Coarctation (*Latin: "drawing together"*) of the aorta (CoA) can be defined as narrowing of the thoracic descending aorta, usually at the site of insertion of the ductus arteriosus. In CoA, the narrowing is usually distal to the left subclavian artery and can vary in severity. If there is severe obstruction to blood flow, haemodynamic stability may rely on flow through the patent ductus arteriosus (PDA). CoA is therefore considered a critical congenital heart defect.

Prevalence

CoA accounts for 4–6% of all congenital heart defects, with a reported prevalence of about four per 10,000 live births. It occurs more commonly in males than in females (59% versus 41%). Most cases are sporadic. However, some chromosomal abnormalities increase the risk: 10% of patients with Turner's syndrome are affected, and the risk is thought to be higher in patients with abnormalities of the X chromosome (e.g. Fragile × syndrome).

30% of patients with CoA presenting in childhood have other associated congenital heart defects, including patent ductus arteriosus (PDA), ventricular septal defects (VSD), mitral valve abnormalities and aortic stenosis. A bicuspid aortic valve is present in nearly two thirds of infants with CoA.

Aetiology

CoA was once thought to be a simple discrete narrowing of the aortic isthmus. It has since become clear that the aortic arch can be affected in a highly variable manner and that it is associated with other left-sided heart lesions. It is also thought to be associated with a more widespread vasculopathy within the more proximal arterial tree; leading to increased prevalence of hypertension and subsequent risk of morbidity and mortality. The exact aetiology by which CoA is not clearly understood. Hypotheses include haemodynamic and ectopic ductal tissue theories.

Haemodynamic theory

Abnormalities decreasing the antegrade flow across the aortic isthmus lead to failure of the isthmus to grow, resulting in hypoplasia and coarctation ("form follows flow" theory). Spontaneous closure of the ductus arteriosus following birth completes the development of aortic obstruction. An increased incidence of CoA in patients with decreased antegrade aortic flow prenatally lends support to this hypothesis.

Ectopic ductal theory

Abnormal extension of ductal tissue into the aorta has been postulated as the cause of the formation of the coarctation shelf in the posterior aortic wall. When closure of the PDA is triggered after birth, this ectopic tissue will contract and sclerose in the same manner as the PDA resulting in narrowing and obstruction.

This theory is supported by the common finding of an aortic arch which looks normal while the PDA is open, which then develops coarctation once the duct shuts. Additionally, some neonates presenting with CoA when the PDA is shut will respond favourably to prostaglandin E1 or E2 infusion. This improvement occurs not by re-opening the PDA but by relaxing the sling of ductal tissue and allowing more forward flow. This theory, however, fails to explain the variable degrees of isthmus and aortic arch hypoplasia associated with CoA.

Neither of these theories adequately explains the widespread changes seen both in left heart structures (mitral valve abnormalities, bicuspid aortic valve) and upper body vascular structure (cerebral aneurysms) commonly associated with CoA. This leads to a further theory that CoA is a manifestation of a wider arteriopathic process. There is also likely to be a genetic component, as familial cases have been reported and association with various gene deletions described, along with the high incidence in patients with Turner's syndrome. Upregulation of certain genes has been postulated but not proven.

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Morphology

CoA classically manifests as a discrete localised constriction "coarctation shelf" of the aortic isthmus (area between left subclavian artery (LSCA) and aortic end of the PDA). However, it often exists as part of a spectrum of aortic narrowing, from this discrete localized narrowing to tubular hypoplasia, with many variations in between. Morphologists argue that although tubular hypoplasia and the localised constriction may coexist, they should be considered as separate entities although this has not been proven. The presence of associated arch hypoplasia is relevant to the longer term risk for the development of hypertension (Figure 1).

Pathophysiology and presentation

Depending on the severity of obstruction to aortic flow, development of collateral circulation and associated heart defects the presentation of CoA may be anywhere from acute circulatory collapse to an incidental finding of absent or weak femoral pulses or asymptomatic hypertension at an older age. As stated before, this article will focus on the presentation in the neonatal period.

Neonatal collapse

The classical presentation of coarctation occurs in a newborn with a discrete coarctation. Initially, while the PDA is open and the sling of ductal tissue is relaxed, there is ample forward flow from the aortic arch to the descending aorta. The baby is clinically well, there is no difference in saturations between the arms and feet (PDA flow will be bidirectional or predominantly left-to-right), and femoral pulses are likely to be either normal or slightly weak (Figure 2a).

At some stage, there may be a transition where the coarctation tissue starts to constrict while the PDA remains open (Figure 2b). The right ventricle will then be able to provide adequate perfusion to the body distal to the coarctation (remember, the right ventricle supports the systemic circulation in the fetus), but it will be shunting deoxygenated blood into the descending aorta, leading to differential saturations between the arms and legs. The baby is likely to remain haemodynamically stable at this point, with possibly normal or weak femoral pulses. The kidneys will remain perfused, so there is also likely to be good urine output. At this point, the left ventricle is exposed to a very large afterload which will eventually compromise function, and the upper limb blood pressure (BP) will climb. This phase may occur very transiently or not at all, meaning that routine oxygen saturation screening can often miss these patients.

Once the PDA restricts or closes (Figure 2c), there is inadequate perfusion of the organs distal to the coarctation. This results in severe acidosis from lower body ischaemia, oliguria due to poor renal perfusion and electrolyte imbalance. The acidosis will compromise ventricular function in the already pressure overloaded left ventricle and circulatory collapse can occur. This can present initially with tachypnoea due to acidosis and pulmonary oedema, often with rapid progression to hypotension, clinical signs of low cardiac output (grey colour, mottling, prolonged capillary refill time), worsening acidosis, coma and cardiac arrest. Due to impaired left ventricular function and reduced cardiac output, upper limb blood pressure may be low making four-limb blood pressure measurement an unreliable clinical sign. In any neonate (especially less than 1 week old) presenting this way, there must be a high index of suspicion of coarctation.

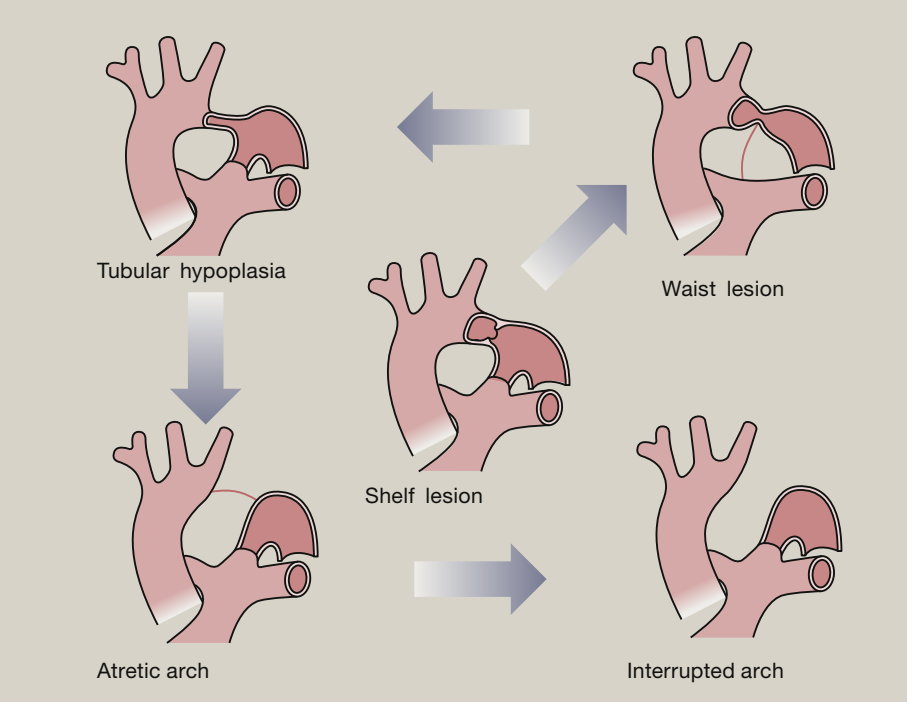

Figure 1 Morphological variants: the morphological spectrum of obstruction in the aortic arch.