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Anomalies of the abdominal aorta in Williams-Beuren syndrome – another cause of arterial hypertension

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Abstract Vascular disease in Williams-Beuren syndrome is based on an elastin arteriopathy which may cause stenoses in small and great vessels. This study presents the pattern of stenotic lesions of the abdominal aorta and the incidence of arterial hypertension. From 112 patients with Williams-Beuren syndrome followed since 1975, 25 patients were studied by aortography. The diameter of the thoracic aorta and the change in diameter to the iliac bifurcation were compared with normal data. Renal artery stenosis was suspected when the proximal vessel diameter was less than 50% of the distal diameter. Of the 25 patients, 20 had vascular stenosis of whom 19 patients were affected by segmental narrowing either of the thoracic aorta (n=9) or the abdominal aorta (n=7) or both (n=3). Hypoplasia of the abdominal aorta was characterised by the smallest diameters at the renal artery level and an increased diameter of the infrarenal abdominal aorta. A total of 11 patients had renal arterial stenosis, associated with narrowing of other aortic segments in 10 cases. Only one patient had a solitary stenosis of the renal artery. Arterial hypertension was diagnosed in 17 patients, 2 of them had no vascular lesions; in the remaining 15 patients stenosis was present in more than one segment (aorta 6, renal artery stenosis 1, both 8). Conclusion: narrowing of the abdominal aorta in patients with Williams-Beuren syndrome is a frequent morphological manifestation of the arteriopathy. Isolated renal arterial stenosis was rare, since it was more frequently combined with a narrowed aorta. Hypertension is a common symptom in the affected group and must be regarded as a manifestation of generalised arteriopathy rather than renal hypoperfusion.

Keywords Abdominal aorta · Arterial hypertension · Renal arterial stenosis · Williams-Beuren syndrome

Abbreviations AH arterial hypertension · RAS renal arterial stenosis · WBS Williams-Beuren syndrome

Introduction

The main characteristics of Williams-Beuren syndrome (WBS) are typical facial abnormalities with mental retardation and vascular stenoses. The molecular defect is located on the long arm of chromosome 7 (7q11.23) including the gene for elastin, the major arterial wall protein [7]. Thus, elastin arteriopathy leads to thickening of the arterial wall resulting in stenosis of the great and small arteries. The pathology shows elastic disorganisation, hypertrophied smooth muscle cells and collagen bundles [8]. The most common locations of vascular stenosis are peripheral pulmonary arteries, aorta ascendens and the aortic arch [12]. Coarctation of the abdominal aorta has also been described [2, 11,12], but there is no information about prevalence and features of abnormalities of the abdominal aorta.

Patients and methods

In a retrospective analysis, we reviewed 25 abdominal aortograms of patients with WBS which had been performed between 1975 and 2000. At time of examination the patients were between 2.4 and 26.1 years old (median 11.5 years). The ratio of males to females was 2.4:1. The indication for the abdominal aortogram was a systolic abdominal murmur, or a otherwise suspected renal arterial stenosis (RAS) in nine patients. In the other 16 patients, in whom we performed the examination after 1990, the abdominal aortogram was part of the routine diagnostic programme in WBS patients, once catheterisation was necessary. To assess the segmental morphology of the aortic diameter, measurements were...
taken at several points: at level of the diaphragm, above the renal arteries and above the iliac bifurcation (Fig. 1).

Growth dependent normal values for the diameter of the thoracic aorta were calculated from measurements of left ventricular angiograms of 36 patients without aortic disease (age: 1 month to 26 years). Indication for angiography was preoperative evaluation of simple cardiac defects. Diameters were measured at the level of diaphragm and computed as a function of body weight (diameter of thoracic aorta = 2.45 x body weight^0.475; SD 1.5 mm). Since there are no normal values for diameters of the abdominal aorta in childhood, we calculated the change of diameter from the level of the diaphragm to the renal arteries and to the iliac bifurcation from the data published by Horob et al. [5] and Pearce et al. [10]. The data of both groups confirm that the aortic diameter at the level of renal arteries is about 80%, at the level of iliac bifurcation about 70% of the diameter of the thoracic aorta. This change in diameter was independent of age, gender and body surface area.

The expected aortic diameter was calculated for each patient. A measured diameter of less than 2 SD from the expected value was considered as hypoplasia. The individual reduction in vessel width was computed. Because the normal profile of the abdominal aorta exhibits a narrowing to 80% of the thoracic aorta at the level of renal arteries, for the suprarenal aorta we considered a narrowing to 70% of the thoracic aorta to be normal or mild, of more than 70% moderate or severe. RAS was diagnosed from the aortograms, no selective injections of contrast medium were performed. Since the stenotic lesion is usually very close to the orifice of the renal artery, there was a risk to inject behind the stenosis when the catheter would be placed in the renal artery. RAS was suspected when the vessel diameter distal to the stenotic area was constantly larger and the stenotic diameter was less than 50% of the vessel diameter.

Blood pressures were taken in all 25 patients as 24 h ambulatory measurements. Arterial hypertension (AH) was diagnosed when the mean of the arterial mean pressure during the daytime (7–22 h) exceeded more than 2 SD of the mean of healthy children. As reference, we used published data of age-matched control groups which were analysed by the same method [6, 13,19].

**Results**

To describe the abnormalities of the abdominal aorta we used four parameters (Fig. 1): the width of the thoracic aorta, the diameter change in the suprarenal and infrarenal segment and the presence of RAS. Only five patients had normal aortography. Thus, 20 of 25 patients had abnormalities in different segments of the abdominal aorta. Fig. 2 illustrates the combination of the hypoplastic thoracic aorta, suprarenal narrowing and RAS.

The thoracic aorta was hypoplastic in 12 patients. The measured diameter was between 2 and 5.2 SD below the expected value. In ten patients, narrowing of the aorta above the renal arteries was considered as moderate or severe. Of these patients with suprarenal narrowing, six also had a hypoplastic thoracic aorta or RAS.

Stenosis of the renal arteries was found in 11 patients. Only one patient had no other signs of aortic narrowing. The other ten patients with RAS had a combination with hypoplastic thoracic aorta or suprarenal narrowing or both (Fig. 2).

In contrast to normal, the aortic diameter increased below the renal arteries in 19 of 25 patients. The mean increase in diameter was 24%. There was no further narrowing of the aorta below the origin of the renal arteries. The mean diameter reduction from diaphragm level to iliac bifurcation was 18.4%. (range: −9%−44%). As mentioned above, a reduction of 30% we considered as normal. Diameter reduction was less severe in patients with hypoplastic aorta, so that the diameter at bifurcation in most patients almost returned to normal.

We found AH in 17 of our 25 patients. Only two of five patients with normal abdominal aorta, but 15 of 20 patients with vascular stenosis were hypertensive. (Fig. 3). Two of ten patients with RAS, three of ten patients with suprarenal narrowing and 3 of 12 patients with hypoplasia of the thoracic aorta were normotensive. RAS was not more frequently associated with hypertension than the other stenotic lesions.

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**Fig. 1** Outline of an abdominal aorta: normal (grey) and in WBS (black). *Arrows* depict points of measurement.

**Fig. 2** Stenosis and combinations of different segmental anomalies in normotensive (open circles) and hypertensive (closed circles) WBS patients.