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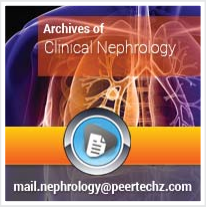

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Short Communication

Arterial hypertension in children with Williams-Beuren syndrome

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Williams-Beuren Syndrome (WBS) is a multisystemic disorder with prevalence of 1/7500 [1], affects both genders equally and it is caused by a microdeletion of 26-28 genes, including elastin and NCF1 genes, of the chromosome region 7q11.23 [1,2]. The disease is transmitted in an autosomal dominant fashion, but almost all cases are the result of de novo mutations [1].

The main clinical features include: elfin facies (100%), short stature, intellectual disability (75%), cardiovascular (80%) and genitourinary abnormalities and occasionally infantile hypercalcaemia (15-45%) [1,3].

This article mainly focuses in cardiovascular manifestations, essentially hypertension, diagnoses approach and treatment. Supravalvular aortic stenosis (SVAS) is the most common cardiac alteration (35-65%) [1], normally discreet or it can involve a long segment stenosis (15%) [1]. SVAS can worsen with time, especially during the first 5years; in 30% of cases it might need cardiac surgery or catheterization with a mortal rate of 6% [4]. Branch or peripheral pulmonar artery stenosis (60%) tend to improve or overcome spontaneously along time [1], essentially if occurs has a single event [4]. Supravalvar pulmonar stenosis is seen in approximately 10% of patients, demonstrating spontaneous improvement in many cases [1,5]. Other cardiovascular abnormalities are: stenosis of the thoracic or abdominal aorta, renal or intracranial arteries and/or vessels at other sites including the neck and limbs [1,7]. Septal defects, regurgitation or prolapse valve mitral, insufficiency or bicuspid aortic valve are seen in association with WBS. Children with WBS have higher risk of sudden death (1/1000 patients/year) by ventricular hypertrophy (biventricular outflow tract obstruction), coronary anomalies (ostial or difuse stenosis or dilatation) and prolonged corrected QT (13%) [1].

These patients are at high risk for systemic hypertension. Loss of function in the elastin gene is associated with increased intima-media thickness with thick irregular elastic fibres, swirling collagen and hypertrophied smooth muscle cells [2]. These vascular lesions are responsible for focal stenosis, generalized vascular narrowing and Hypertension (HTN) [3]. Vascular stiffness normally increases with ageing and comorbid conditions such as HTN and diabetes [3]. NCF1 is one of the deleted Williams genes, is a component of NAD(P)H oxidase complex and involved in generation of oxidase stress and reduced copy number of this gene are associated lower risk of HTN in WBS [6].

HTN occurs in about 50% of cases, at any age. Although in many patients it remains unexplained, most frequently found etiologies are: renal artery stenosis (50%) and/or diffuse aortic narrowing and/or aortic coarctation and high sympathetic activity [1,2].

Hypertension is also more common in subjects with a history of infantile hypercalcemia, even though it hasn't been found a correlation between them [2].

Severall genitourinary manifestations can occur in a SWB: multicystic kidney, renal hypoplasia or aplasia, ureterohydronephrosis and nephrocalcinosis (if hypercalcemia with hypercalciuria) [1]. It can cause hypertension and evolve to a renal chronic disease.

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Blood pressure measurement in both arms is recommended with use of a manual cuff at the end of the visit to minimize anxiety [3]. Consider cardiology or nephrology referral for hypertension when blood pressure is > 90th percentile for age and height [3], but HTN was defined blood pressure > 95th percentile for age and height. When these patients are diagnosed with hypertension, an investigation for vascular injuries passable of correction and evidence for end-organ damage should be performed [1]. Some experts, recommend an initial trial of antihypertensive medications and if HTN resists pharmacology therapy, it should proceed to investigate the renovascular causes [1,7]. However, other experts in the field would evaluate all patients with HTN for discrete vascular lesions that may be correctable and evidence for end-organ damage [1]. In HTN initial approach an abdominal ultrasound can be performed to mislead of genitourinary abnormalities and some serum blood (urea nitrogen, calcium, creatinine, 25hidroxyvitamin, 1,25-hydroxyvitamin and parathyroid hormone concentrations) and urinary tests (spot calcium/creatinine ratio). Renal ultrasound with doppler flow studies of renal arteries and abdominal aorta is performed because of the increased likelihood that renal artery stenosis is the etiology of HTN [1]. Patients with highly suspicion and/or inconclusive studies and/or refractories to antihypertensive therapy can perform a computed tomography or magnetic resonance angiography and/or arteriography to identify the renovascular cause [1]. The choice of study should be based on individual patient's characteristics including the need for/risk of sedation for the procedure, baseline renal function and clinical experience [1].

In patients whose initial work-up is negative or inconclusive, pharmacologic therapy initiates with Calcium Channel Blockers (CCB), namely amlodipine or nifedipine [1,2]. Whether blood pressure cannot be controlled, labetalol or beta-blocking drug can be added [8]. If hypertension remains difficult to control on both CCB and betablocker and if there is enough certainty of non-presence of renal artery stenosis, angiotensin-converting enzyme inhibitors or angiotensin-receptor blockers may be introduced with careful monitoring of renal function [1]. Other medications include diuretics, minoxidil or clonidine [1]. In patients identified with renovascular cause, corrective therapies such as surgical repair or percutaneus transluminal angioplasty can be performed; however the results are poor with persistent or recurrent HTN [1].

Conclusion

The patients with WBS have high risk of sudden death and for HTN, thus a multidisciplinary approach to patient is needed with cooperation and interaction between nephrologists and cardiologists.

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