A CASE OF TURNER SYNDROME ASSOCIATED WITH SEVERE COARCTATION OF THE AORTA

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A CASE OF TURNER SYNDROME ASSOCIATED WITH SEVERE COARCTATION OF THE AORTA (Abstract): Turner Syndrome (TS) is a chromosomal abnormality due to the complete or partial lack of chromosome X, with clinical polymorphic elements: small size, hypogonadism, various visceral abnormalities. About half of the cases may suffer from congenital heart abnormalities. We describe hereunder a case of TS diagnosed with coarctation of the aorta due to high blood pressure detected by accident. Key words: TURNER SYNDROME, COARCTATION OF THE AORTA

Turner Syndrome (TS) is a chromosomal abnormality due to the complete or partial lack of chromosome X. TS incidence is about 1 case/2500 new-born female babies. TS pregnancy frequency is much higher, and many foetuses are lost by miscarriages (10% of miscarriages have TS). The TS syndrome is a generic one, the most common chromosomal abnormality being homogeneous monosomy 45,X (55% of the cases), followed by structural abnormalities of chromosome X (25% of the cases) and one normal cell line mosaic in 20% of the cases.

The clinical symptoms vary, whereas the subjects’ psychological and motor development is within normal limits. At birth, this condition characterized by the subjects’ small size, upper and lower limb lymphedema, webbed neck; 5% of the cases have severe cardiac malformations (severe coarctation of the aorta) requiring urgent surgery. The characteristics that are constantly present during childhood, adolescence and adulthood are the subjects’ small size and, with age, delayed puberty, ovarian failure and infertility (1, 2).

These are accompanied with less common conditions such as: autoimmune diseases (thyroiditis, diabetes mellitus, celiac disease). 44.5% of the cases have congenital heart abnormalities (less severe coarctation of the aorta, bicuspid aortic valve, pulmonary and coronary artery abnormalities), whereas 40% of the patients suffer from early hypertension since their adolescence (3). The diameter of the aorta must be monitored the subjects’ whole life, as there is a risk of aortic dilatation. Hypoacusis due to frequent otitis and horseshoe kidney are also included in this category (4, 1).

Medical management consists of growth hormone therapy during childhood, then
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estrogen-progesterone supplementing during puberty. Trans-thoracic echography is recommended even when no clinical symptoms are present and is designed to detect possible aortic valve abnormalities or aortic dilatation (5).

The subjects’ life quality, self-esteem and social integration are much better when puberty is not induced too late. The prognosis depends on the associated abnormalities: congenital heart abnormalities, hypertension, diabetes mellitus, obesity, thyroiditis, osteoporosis (2).

CASE REPORT

Patient DRM, aged 14 years and 3 months, came for primary amenorrhea, repeated hypertension (highest value 150/80 mmHg) and systolic murmur.

Family medical history. The patient is the second child of an unrelated apparently healthy couple, the mother 31 years of age and the father 37 years of age (at the time of conception). No other congenital anomalies, genetic diseases or reproduction disorders run in the family.

Personal physiological history. The pregnancy had a normal evolution. The birth was a term birth, the delivery was natural, head first, APGAR score 7 (1), weight 2000g, upper limb lymphedema. No data on the size and head circumference at birth were available.

Personal pathological history. The patient’s first medical examination occurred when she was 13 years and 11 months old, in the endocrinology ward, where she went for primary amenorrhea. High blood pressure and systolic murmur are detected on this occasion, for which reason she was referred to the pediatric cardiology clinic.

Status upon hospitalization (14 years 3 months). The clinical examination reveals: small size (G = +1.16 DS, T = -2.33 DS, Pc = +0.88 DS), normally colored teguments and mucous membranes, lymphedema on the dorsal surface of the hands and feet, webbed neck, hard and soft palate dysmorphism, great inter-nipple distance, low hair level on the nape, many nevi. Slightly dysmorphic facies, ears positioned low and slightly flaring nose. Athletic constitution with broad shoulders and narrow hips. The patient also had cubitus valgus, clubfoot, shortening of the 4-5 metacarpal and metatarsal bones, hypoplasic nails and kyphotic-scoliotic posture. As concerns the cardiac system, we noticed the presence of collateral circulation in the thoracic cage, a III/6 degree systolic murmur in the left II-III intercostal space with interscapular-vertebral irradiation and in the epigastric region, clear peripheral pulse in the radial arteries, weaker pulse in the lower limbs (dorsalis pedis artery and posterior tibial artery, which is possibly also due to the lymphedema). The blood pressure was 160/80mm Hg in the upper limbs, and 140/70mm Hg in the lower limbs. The patient also complained of vertigo, fronto-occipital cephalalgia and bilateral hypoacusis.

During her hospitalization we detected high blood pressure (150/80 mmHg); the results of the complete blood count, hepatic and renal tests, glycemia and urine tests were normal; the fist X-ray reveals that the bone age is in agreement with the chronological age, fertile growth cartilages; the pelvic echography reveals that both kidneys have normal morphology, uterus 25/9.9/26 mm, left gonad 15.5 mm, no right gonad is detected; the hormonal tests reveals subclinical hypothyroidism (TSH = 7.47 μUI/ml, fT4 = 17.95 pmol/l, AAT-PO = 8.82 UI/ml); thyroid echography – hypoechochogeneous thyroid.

Echocardiography revealed a severe aortic coarctation.
The genetic examination sets a certain diagnosis, namely Turner Syndrome, homogeneous monosomy X, karyotype 45,X.

As a coarctation of the aorta was suspected based on clinical grounds, a thoracic angio CT scan was performed, which revealed a significantly narrowed area (0.73 cm diameter) on the descending thoracic aorta 0.8 cm below the origin of the left subclavicular artery; the caliber of the ascending aorta was 3.27 cm (2.14 cm in the prestenotic area and 2.06 cm in the poststenotic area); considerable collateral circulation visible in the cervico-dorsal area, dilatation of the left subclavicular artery 1.42 cm, of the right brachiocephalic arterial trunk 2.16 cm, of the internal thoracic arteries and of the collateral branches of the descending thoracic aorta; partially aberrant pulmonary venous drainage with possible aterio-venous fistula, pulmonary perfusion disorders. The spine was also affected (spine compression at the level of the D3 and D12 vertebrae, D5-L1 multiple intrabody disk herniations and slight kyphosis.

The final diagnosis was Turner Syndrome by homogeneous monosomy X, tight coarctation of the descending thoracic aorta, subclinical hypothyroidism, lower limb lymphedema, kyphosis.

**Fig. 1.** Patient D.R.M.: echocardiography (suprasternal view) - severe coarctation of the descending aorta

**Fig. 2.** Patient D.R.M – Angio-CT: area narrowing descending thoracic aorta, the vertebral body subsidence cuneiform D3, D12, D5-L1 multiple intrabody disk herniations and slight kyphosis

**Fig. 3.** Patient D.R.M – descending aorta coarctation

**Therapy**

We recommended endocarditis prophylaxis, specific hypertension therapy and avoidance of physical exertion. Measures should be taken to prevent the complications of congenital abnormalities, either by dilating the stenosis area by means of a balloon and stent, or by specific surgical procedures (6). The proband will also be given Euthyrox 100 μg/day for her subclinical hypothyroidism.
Evolution and Prognosis
The asymptomatic forms of coarctation of aorta are detected due to high blood pressure values accompanied by cephalalgia, repeated nosebleed or after a stroke. The prognosis depends on the success of the cardiac abnormality treatment and on hypertension persistence after surgery.

Given the patient’s bone age, ~ 14 years, and her heart abnormality, GH therapy is tardy and will be avoided. The management of female patients suffering from Turner Syndrome proposed by Kavoussi et all. includes cardiology supervision in outpatient clinics even when there are no clinical signs, blood pressure measuring on each visit to the doctor, renal echography and aggressive urinary infection therapy, TSH, glycemia and cholesterol dosing, annual hepatic tests, early scoliosis detection, hyponacusis due to repeated otitis, growth hormone therapy until the bone age of 15 years, then estrogen-progesterone therapy, integration in a support group and psychological counselling designed to prevent any psychosocial integration difficulties (7, 8).

As literature reports a frequent association between TS and autoimmune diseases (Hashimoto thyroiditis, type 1 DM, celiac disease, juvenile rheumatoid arthritis, psoriasis, veal skin) and since the patient already has subclinical hypothyroidism, careful pediatric follow-up is required for the early detection of such complications (9).

The specificity of the case consists of the association between a coarctation of the descending thoracic aorta, a severe form diagnosed at the age of 14 years, and the Turner Syndrome.

CONCLUSIONS
The clinical manifestations, extremely suggestive of Turner Syndrome and of cardiac abnormality, require early genetic and cardiology exploration for a certain diagnosis setting, adequate therapy and prevention of serious possible complications of the coarctation of the aorta.

REFERENCES