Turner syndrome in childhood and adolescence – a multifaceted state of health

The internal genitalia, bone mineralization and aortic dilatation

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ABSTRACT

Conventional pubertal induction regimes with estrogen may be insufficient in Turner syndrome (TS) in terms of normal uterine growth. Magnetic resonance imaging (MRI) may provide more accurate measurements of uterine and ovarian size than ultrasound (US). Reduced bone mineral density (BMD), osteoporosis and an increased risk of fracture are features of TS. The relationship between BMD and bone markers in girls with TS might provide predictive information on fracture risk. Aortic dilation and aneurism formation is encountered in up to half of the adult TS population. The smaller body size in TS is a potential confounder when evaluating aortic diameters, which is corrected by adjusting the aortic diameters for body surface area (BSA). Recent studies suggest that MRI is a superior methodology to echocardiography.

The aims of the study were to compare uterine and ovarian size evaluated by US and MRI, to evaluate BMD by DEXA and the association to bone markers in blood, and to study the dimensions of the thoracic aorta by MRI to elucidate the aortic size and the potential predictors of the variation in aortic dimensions in 41 girls with TS ageed 10-25 years compared to agematched controls.

In TS, MRI was superior to US in detecting the ovaries. This finding could be important in future fertility treatments such as cryopreservation of ovarian tissue. The uterine size was subnormal by MRI in TS compared to controls despite seemingly age-appropriate estrogen treatment in the majority of patients. At the same time, we found estrogen treated girls and young women with TS to accrue bone mineral in a pace and time wise similar fashion as agematched controls in the spine and the hip, while accrual at the cortical part of the forearm seem deficient. In combination with the increased level of bone resorption markers and a low vitamin D status in TS, these results emphasizes the importance of focusing on general bone preserving measures like exercise and supplementation with calcium and vitamin D in addition to estrogen even in the young generation of TS. Aorta was not enlarged in TS. BSA predicted aortic size at the entire thoracic aorta and thus holds the promise of potentially more adequate identification of the pediatric TS patients facing an increased risk of dissection in childhood as well as in adulthood.

Longitudinal studies are needed to determine the optimal estrogen treatment protocol not only to induce puberty but also maintenance therapy. Studies defining the relationship between explanatory factors, aortic dilation and the risk of dissection in TS are also important. These future studies could potentially provide updated guidelines regarding cardiovascular surveillance including a threshold for aortic diameters in TS where therapy should be initiated.