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Multidisciplinary approach to Loeys-Dietz Syndrome in children

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OBJECTIVE
The aim of this study is to describe the clinical manifestations of pediatric patients with Loeys-Dietz Syndrome followed-up at our center.

METHODS
Epidemiology, genetics, clinical findings and follow-up were evaluated. All pediatric patients with Loeys-Dietz syndrome were included.

RESULTS
A total of 14 patients were evaluated, with a median age at diagnosis of 10 years (range 0 to 20 years), 7 boys and 7 girls. Genetics: 4 (+1) TGFBR 1, 8 TGFBR2 and 1 TGFB2. Positive family history on 9/14 cases (64%). All patients presented aortic root dilatation except 2, median z-score of the aortic root +3.87 (range 0.53-7.11), and 9 also had aneurysms or dilatations at other levels. All the affected patients were treated with ARA-II and/or Beta-blockers. Aortic root replacement was needed on 6 (42%): 5 David's technique and 1 Bentall technique. Age range at surgery was 9 to 17 years old, and the range of Valsalva sinuses diameter was 38.5 to 47mm. One patient required pacemaker implantation immediately after David's surgery.

Half of the patients had pectus excavatum/carinatum, almost half had scoliosis and 5 had hindfoot deformity. Most common neurological involvement was tortuosity of the cerebral vasculature (7/14), and one patient suffered frontal hypoxic-ischemic events. Bronchiectasis were found on 2 patients, 2 obstructive sleep apnea and one spontaneous pneumothorax. Ophthalmologic involvement with strabismus or optometric disorders was described on 5 patients, and 1 retinal detachment. Other manifestations were one failure to thrive, one gastro-esophageal reflux, one food allergy and one asthma. Almost half of the patients had inguinal/umbilical hernias.
Median follow up of 4 years (range 2-9). Three patients died (21,4%). Two due to aortic dissection and one to cerebral hemorrhage at 12, 19 and 20 years of age.

CONCLUSIONS

Loeys Dietz Syndrome is a complex multisystemic disease. The prognosis is determined by the cardiovascular involvement, even in early ages. A multidisciplinary approach and a coordinated transition to adult units is mandatory.