Review

Isabel Rey Madeira


Turner syndrome affects 25–50 per 100,000 women. It is a chromosomal disorder that affects women who have one intact X chromosome and complete or partial absence of the second X chromosome and is associated with one or more clinical manifestations.

The classical definition of the Turner syndrome necessarily includes typical clinical characteristics such as classic facial features, neck webbing, and lymphedema. However, the clinical presentation may include only growth retardation and delayed puberty. Other clinical manifestations of the syndrome are early hearing loss, congenital cardiovascular disease, skeletal and renal anomalies, neurodevelopmental disorders, hypothyroidism, and celiac disease.

Dysfunction of the affected organs and systems occurs throughout life and requires a multidisciplinary approach. The care provided to girls with Turner syndrome has followed the advances in medical science, particularly in the fields of genetics, growth and development during childhood and adolescence, and congenital and acquired cardiovascular diseases. Among the comorbidities, neurocognitive issues and the transition from pediatric care to adult care have been given special attention.

Many necessary therapeutic interventions, such as the treatment of short stature and steroid replacement therapy, as well as the prevention of morbidities, such as hypertension, are of particular interest to the health professional caring for these girls from the first months of life through adolescence. Issues such as fertility should be explained to the families even before adulthood.

In this sense, the initial health assessment of patients with Turner syndrome is the pediatrician’s responsibility, who should refer these girls to other specialists in due time.

The project that was the basis of the recommendations listed in this article was an initiative of the European Society for Endocrinology and Pediatric Endocrine Society, in collaboration with the European Society for Pediatric Endocrinology, Endocrine Society, European Society of Human Reproduction, American Heart Association, Society for Endocrinology, and European Society of Cardiology. The document was endorsed by the European Society for Endocrinology, Pediatric Endocrine Society, European Society for Pediatric Endocrinology, European Society of Human Reproduction and Embryology, and Endocrine Society.

The importance of this document for pediatricians becomes evident, not only for those undergoing training in pediatric medical residency programs, but also for those who seek continuing medical education.

1 Coordinator of the Department of Pediatric Endocrinology. Adjunct Professor of Department of Pediatrics. Medical Sciences Faculty. Universidade do Estado do Rio de Janeiro (UERJ).