Opinion

Mixed gonadal dysgenesia in pediatric patient. About a case

Volume 15 Issue 3 - 2024

Alfonso Martínez Villamandos, Eduardo López Candel, Tomás Ferraris, Cristina Domínguez, Paloma Ramos, Carolina Marañés Gálvez, Luis Alonso Jiménez
Hospital Universitario Torrecárdenas, Almería, Spain

Correspondence: Alfonso Martínez Villamandos, Hospital Universitario Torrecárdenas, Almería, Spain, Email fronchomartine@gmail.com

Received: April 30, 2024 | **Published:** May 13, 2024

Introduction

We present the case of a 6-year-old patient, female phenotype, referred to pediatric surgery for mixed gonadal dysgenesis diagnosed during a study for short stature.

Anthropometrically, he was p3 for weight, p1 for height and with a BMI of 14.38 (p20).

After performing a cytogenetic study in peripheral blood, two cell populations were observed. The first, with 7 metaphases, has 45 chromosomes with a single X chromosome. The second, with 23 metaphases, has 46 chromosomes with an X chromosome and a Y chromosome. Chromosome formula 45X(7)/46,XY(23)

The phenotype of patients with 45X/46XY mosaic karyotype varies in a wide range from the female phenotype with classic Turner syndrome, through individuals with ambiguous genitalia, infertile men (Hypogonadism and oligo/azoospermia), to normal men. Therefore, both Turner syndrome and virilization can be expected. The gonads are usually dysgenetic with insu iciently di erentiated testicular tissue, which can occur in both gonads (mixed gonadal dysgenesis), with an increased risk of gonadoblastoma and other tumors on these gonads.

There is no clinical relationship between the symptoms and the proportions of the mosaic since its distribution in the di erent tissues of the body is not known.

To complete the study of this patient, ultrasound and gynecological magnetic resonance imaging were performed, which reported the presence of uterine rudiment without identifying gonadal structures, although the presence of bandellet gonads or gonadal striae could not be ruled out. These studies support the diagnosis of mixed gonadal dysgenesis

The management of these patients requires exploratory laparoscopy to confirm the presence of dysgenetic gonads or gonadal remains and to proceed with their removal if they exist, given the risk of tumor development.

In our patient, exploratory laparoscopy was performed, which confirmed the presence of dysgenetic gonads. It was removed and histologically studied, which reported the presence of uterine tubes with fimbriae, without significant histological alterations. In both gonads, no tissue macroscopically compatible with the ovary was observed; microscopically, remains of ovarian parenchyma were observed, composed only of stroma and rete ovarii, without follicular structures or findings of malignancy (Figures 1–3).[1–4]

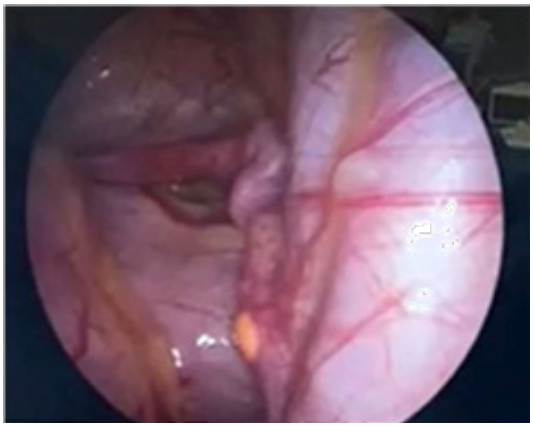

Figure 1 Note the presence of ectopic adrenal tissue in the gonad.

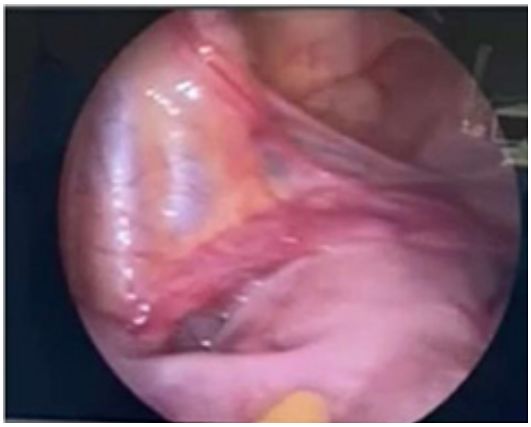

Figure 2 Left side.

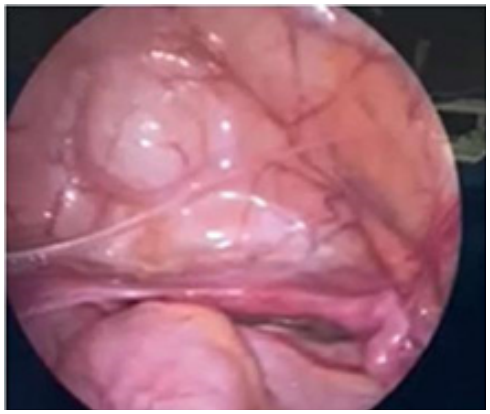

Figure 3 Uterus.

Acknowledgments

None.

Funding

None.

Conflicts of interest

All authors declare that there is no conflict of interest with respect to this manuscript.

References

1. de Marqui AB, da Silva-Grecco RL, Balarin MA. Prevalência de sequências do Y e de gonadoblastoma em síndrome de Turner [Prevalence of Y-chromosome sequences and gonadoblastoma in Turner syndrome]. *Rev Paul Pediatr*. 2016;34(1):114–121.
2. ALvarez-Nava F, Soto M, Sánchez MA, et al. Molecular analysis in Turner syndrome. *J Pediatr*. 2003;142(3):336–340.
3. Zelaya G, López Marti JM, Marino R, et al. Gonadoblastoma in patients with Ullrich-Turner syndrome. *Pediatr Dev Pathol*. 2015;18(2):117–121.
4. Shen W, Li Y. Gonadoblastoma in Turner syndrome with puberty delay: A case report and literature review. *Mol Genet Genomic Med*. 2023;11(12):e2300.