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PERSISTENT MULLERIAN DUCT SYNDROME TYPE I: CAUSES, COMPLIKA TIONS, AND MODERN METODS OF DIAGNOSIS AND TREATMENT.

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Abstrakt

Currently, the number of males suffering from Persistent Müllerian Duct Syndrome (PMDS) is increasing worldwide every year. This syndrome is one of the rare disorders of sexual development in phenotypically male individuals with a 46XY karyotype. It is characterized by the presence of remnants (derivatives) of the Müllerian ducts and, in some cases, remnants of the uterus, fallopian tubes, and the upper part of the vagina. The only clinical manifestation of this syndrome is cryptorchidism. This article discusses the causes of Persistent Müllerian Duct Syndrome, its symptoms, complications, and modern methods of diagnosis and treatment.

Keywords

PMDS, cryptorchidism, Müllerian duct derivatives, Anti-Müllerian Hormone (AMH), testosterone, Sertoli cells, inguinal region.

Introduction

Persistent Müllerian Duct Syndrome type I is a rare disorder of sexual differentiation in which normal male external genitalia are preserved. The syndrome is clinically manifested by cryptorchidism (undescended testes). Because Müllerian structures persist, the testes cannot descend from the abdominal cavity into the scrotum. As a result, this condition may lead to infertility due to cryptorchidism and impaired spermatogenesis.

Causes

The condition develops due to impaired biosynthesis or function of Anti-Müllerian Hormone (AMH). The main function of AMH is to suppress the development of Müllerian duct derivatives in male fetuses. This hormone is produced by Sertoli cells. In about 45% of cases, the syndrome occurs due to

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mutations in the AMH gene, which reduce hormone production. In other cases, mutations occur in the AMH receptor gene, preventing the hormone from binding to tissues and performing its function. The disorder follows an autosomal recessive inheritance pattern.

Complications

Unilateral or bilateral cryptorchidism and inguinal hernia containing Müllerian remnants may lead to infertility. The abdominal cavity has a temperature slightly higher than the scrotum, and even a difference of 2–3°C negatively affects normal spermatogenesis. The presence of rudimentary female organs may also cause obstruction of the vas deferens.

Modern diagnostic methods

Diagnosis usually focuses on the presence of cryptorchidism. It often involves a multidisciplinary team and may be discovered incidentally during abdominal surgery. Diagnostic methods include ultrasound, MRI, laparoscopy, endocrine testing, and genetic testing.

Ultrasound (US): Used to detect Müllerian structures and locate undescended testes.

MRI: A highly informative non-invasive method for identifying Müllerian remnants and testicular position.

Laparoscopy/Laparotomy: Considered the gold standard because it allows direct visualization of internal structures.

Endocrine tests: Blood analysis for Anti-Müllerian Hormone (AMH).

Genetic testing: Identification of mutations in the AMH or AMHR2 genes.

Treatment

Treatment usually includes surgical removal of Müllerian structures (hysterectomy and salpingectomy). During surgery it is important to preserve the neurovascular bundle of the testes. After removal, orchiopexy is performed to move the testes into the scrotum. This procedure should ideally be performed early in childhood (before 1–2 years of age) to protect spermatogenesis.

Conclusion

Persistent Müllerian duct syndrome is a rare but clinically significant disorder of sexual differentiation in males caused by insufficient production of anti-Müllerian hormone or impaired tissue sensitivity to it.

The persistence of rudimentary Müllerian structures—the uterus and fallopian tubes—with a normal male phenotype creates the anatomical and functional preconditions for the development of cryptorchidism, inguinal hernias, and obstructive infertility.

Persistent Müllerian Duct Syndrome is a rare but clinically significant disorder caused by insufficient AMH production or impaired sensitivity to it. Early diagnosis and appropriate surgical treatment help eliminate anatomical abnormalities and preserve reproductive potential, which has important clinical and prognostic significance.

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