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(REVIEW ARTICLE)

Morris syndrome (Androgen Insensitivity Syndrome)

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Abstract

Morris syndrome, also known as Androgen Insensitivity Syndrome (AIS), is an intriguing and extraordinary genetic condition that exerts a remarkable influence on individuals with male sex chromosomes (XY). Within the realm of this rare syndrome, the body displays a captivating indifference to androgens, the very essence of male hormones like testosterone, leading to the emergence of physical characteristics typically

associated with the female gender. It is an occurrence that befalls a mere fraction of the population, affecting around 1 in 20,000 individuals.

Morris syndrome is caused by a genetic mutation located on the sex X chromosome affecting the AR (androgen receptor) gene.

Androgen insensitivity syndrome is characterized by the presence of external female phenotype, 46,XY karyotype, and intraabdominal testes. This syndrome is the third most frequent cause of primary amenorrhea, after gonadal dysgenesis and congenital absence of the vagina. In approximately 40% of AIS patients, there is no family history of the disease.

Morris syndrome unfolds with an intricate interplay of internal and nonfunctional testicles, which, despite their presence, fail to fulfill their intended biological purpose of sperm production. Meanwhile, on the external front, the genitalia manifest a striking semblance to that of the female anatomy, featuring a vagina and labia. Additionally, some individuals with Morris syndrome may witness the existence of an enlarged clitoris, bearing an uncanny. Comprehending Morris syndrome is vital for healthcare professionals to offer accurate diagnoses and appropriate treatment modalities. Increased awareness among the general populace can foster an inclusive and supportive environment for individuals navigating life with Morris syndrome.

Objective: The objective of this article is to provide a comprehensive understanding of Morris syndrome, also known as Androgen Insensitivity Syndrome (AIS), including its genetic basis, clinical manifestations, challenges faced by individuals with the syndrome, and the importance of accurate diagnosis and appropriate treatment modalities.

Methods: To achieve the objective, the article will employ a literature review approach, drawing information from scientific and medical sources. Peer-reviewed research articles, medical textbooks, and authoritative online resources will be consulted to gather relevant information on Morris syndrome.

Keywords: Morris syndrome; Androgen Insensitivity Syndrome (AIS); Rare genetic condition; Male sex chromosomes (XY); Insensitivity to androgens; Testosterone; Female physical characteristics; 1 in 20,000 individuals (3); Internal testicles; Nonfunctional testicles; External genitalia; Enlarged clitoris.

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1 Introduction

Morris syndrome, also known as Androgen Insensitivity Syndrome (AIS), is an intriguing and extraordinary genetic condition that exerts a remarkable influence on individuals with male sex chromosomes (XY). Within the realm of this rare syndrome, the body displays a captivating indifference to androgens, the very essence of male hormones like testosterone, leading to the emergence of physical characteristics typically associated with the female gender. It is an occurrence that befalls a mere fraction of the population, affecting around 1 in 20,000 individuals **(3)**.

The initial reports The initial reports of the SIA date back to the 19th century, and it is even supposed known personalities such as Queen Elizabeth I of England and Joan of Arc England and Joan of Arc had this affectation. **(10)**

Androgen insensitivity syndrome is characterized by the presence of external female phenotype, 46,XY karyotype and intraabdominal testes. This syndrome is the third most frequent cause of primary amenorrhea, after gonadal dysgenesis and congenital absence of the vagina. **(9)** In approximately 40% of AIS patients of AIS patients have no family history of of the disease. **(10)**

Morris syndrome unfolds with an intricate interplay of internal and nonfunctional testicles, which, despite their presence, fail to fulfill their intended biological purpose of sperm production. Meanwhile, on the external front, the genitalia manifest a striking semblance to that of the female anatomy, featuring a vagina and labia. Additionally, some individuals with Morris syndrome may witness the existence of an enlarged clitoris, bearing an uncanny resemblance to the male organ known as the penis.

A genetic analysis was performed for the androgen receptor (AR) gene. Two point mutations were found: in exon 1 there is a silent R210R mutation, in exon 7 there is a change from G to A at codon 866, which predicts the change of amino acid 866 (valine to methionine) in the protein, mutation V866M. **(8)**

The syndrome's severity showcases a spectrum of variations, with each individual's experience being unique. In certain cases, individuals may remain oblivious to their condition until adolescence or adulthood, when the full realization of their genetic distinction dawns upon them. Such delayed awareness can be attributed to the outward manifestation of physical traits commonly associated with femininity, thus concealing the underlying genetic makeup. Only through medical evaluation and diagnostic procedures can the veil be lifted and the diagnosis of Morris syndrome be unveiled.

The multifaceted nature of Morris syndrome presents distinct subtypes. Complete AIS, rarely identified during childhood, may be stumbled upon during exploratory surgeries, when an abdominal or groin mass is discovered, only to reveal itself as an undescended or partially descended testicle **(7, 3)**. Most individuals with this variant remain undiagnosed until they experience a cessation of menstruation or face difficulties conceiving. On the other hand, partial AIS is frequently detected in childhood due to the presence of both male and female physical characteristics, acting as telltale signs. **(7, 11)**

The appearance of individuals with AIS varies throughout their lifespan. Newborns with AIS exhibit externally normal female genitalia, accompanied by undescended or partially descended testicles and a typically short vagina, often lacking a cervix. In some instances, the vagina may even be almost absent. **(1)** As these individuals reach puberty, the pituitary gland stimulates the testes, triggering testosterone production. Interestingly, due to the close chemical resemblance between testosterone and estrogen, a portion of the testosterone converts back to estrogen in the bloodstream, resulting in breast growth, albeit potentially delayed. Women with AIS do not experience menstruation and are unable to conceive. Since the development of pubic and armpit hair relies on testosterone, most women with AIS have minimal to no pubic or armpit hair, though some may possess sparse hair. **(5)**

The onset of symptoms associated with Morris syndrome exhibits a considerable degree of variability. Symptoms can manifest during different age ranges or occur at various stages of an individual's life, with some cases defying any predictable pattern. Pinpointing the onset of symptoms proves crucial for physicians to establish an accurate diagnosis, aiding them in providing appropriate medical care.

Comprehending Morris syndrome is vital for healthcare professionals to offer accurate diagnoses and appropriate treatment modalities. Increased awareness among the general populace can foster an inclusive and supportive environment for individuals navigating life with Morris syndrome. Further exploration of the genetic and physiological mechanisms underlying the condition is imperative to enhance diagnostic capabilities and refine treatment strategies. When there is vaginal shortening surgery or dilatation techniques are performed to avoid dyspareunia and allow for dyspareunia and allow sexual intercourse **(11)**

By illuminating the distinctive features, challenges, and treatments associated with Morris syndrome, we can foster understanding, empathy, and support for individuals living with this rare genetic condition. Through ongoing research, medical advancements, and societal awareness, we can strive toward improved outcomes and a higher quality of life for those affected by Morris syndrome and other related conditions.

Deep within, the testicles find an unconventional abode. Rather than residing in the familiar confines of the scrotum, they make their home within the body itself. However, their presence is often accompanied by a twist of fate. These internal testicles, while fascinating, are often nonfunctional, failing to fulfill their role in producing sperm. **(8, 10)**

In the realm of female reproductive health, a curious phenomenon emerges. Women with AIS experience a departure from the rhythmic dance of menstruation. Their bodies, due to their insensitivity to androgens, the hormones essential for typical female development, do not engage in the monthly shedding of the uterine lining. It is a unique aspect of their journey, a deviation from the norm that shapes their experiences.

As the symphony of hormones unfolds, estrogen takes center stage, casting its enchanting spell. Within the realm of AIS, this hormone holds the power to awaken breast growth, a remarkable transformation that varies in timing and extent. It is a testament to the harmonious interplay of the body, where femininity blossoms in its own unique way.

Androgen insensitivity syndrome has complex and varied clinical manifestations which pose many challenges for physicians and patients. The clinical manifestations may be specific to each phenotype, depending on the degree of androgen insensitivity **(4)**

The realm of hair, too, reveals its intriguing secrets. In the absence of sufficient testosterone, the orchestrator of hair growth, individuals with AIS may find their pubic and armpit regions adorned with subtlety or even devoid of hair. It is a distinctive aspect, a visual reminder of the intricacies within. **(8)**

Due to the absence of a functional uterus and ovaries, women with AIS often face challenges in conceiving and bearing children. It is a reality that calls for understanding and support, as they navigate the complex emotions and decisions that accompany their unique reproductive journey.

In exploring of Morris syndrome, we gain insight into the extraordinary experiences of individuals affected by AIS. Each aspect holds a story, a testament to the diverse pathways of human existence. It is through empathy, understanding, and ongoing research that we can offer support and foster an inclusive environment for those living with Morris syndrome and other related conditions.

2 Conclusion

In conclusion, Morris syndrome, or Androgen Insensitivity Syndrome (AIS), presents itself as an exceedingly rare genetic condition, entwined with captivating intricacies. Its impact predominantly affects individuals with male sex chromosomes, leading to insensitivity to androgens and the subsequent emergence of female physical traits. Occurring in a fraction of the population, the syndrome often evades detection until later

stages of life, showcasing varying degrees of severity. Effective management involves a combination of surgical interventions, such as testicle removal, and hormone therapy facilitating the development of desired physical characteristics.

Compliance with ethical standards

Disclosure of conflict of interest

No conflict of interest to disclosed.

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