Rare disease makes people look like a woman, with male genitals

University of Granada researchers have designed a guideline for physicians and patients on the Androgen Insensitivity Syndrome (AIS), a rare disease that makes the subject develop reverse sex, which occurs when a subject looks like a woman but has male genes.

AIS has low prevalence (it only affects one in 2000 people), and it is characterized by the inability of tissues to respond to the action of male hormones. This prevents individuals with XY sex hormones (i.e. 46,XY) to develop male genitalia. This disorder is caused by a mutation in the gene that codifies the receptor of androgens, and diagnosis is confirmed by the identification of such mutation. This disease is transmitted y a recessive gene associated to gender i.e. it is transmitted by women but it is only developed by men.

The researchers examined the most relevant clinical and epidemiological data of AIS in a review study recently published in the journal *Gynecological Endocrinology*. The guideline for patients includes the follow-up protocols to be applied from birth to adulthood, through childhood and adolescence. The guideline

also includes additional information for patients.

Diversity of Symptoms

Clinical symptoms of AIS range from spermatogenic defects causing infertility in men with otherwise normal genitalia, to subjects who look female in appearance but have not menstruation or female internal genitalia. Sometimes, the gender of IAS subjects cannot be identified at birth and a more precise diagnosis is required to determine the sex of the newborn and plan potential treatments.

According to one of the authors of the study, a researcher at the University of Granada Department of Obstetrics and *Gynecology, therapy for AIS is based on three pillars:*

"The first step is reinforcing the sexual identity of the subject with the help of psychologists. In cases of sexual ambiguity and determination of female gender, the second step is to perform a gonadectomy (removal of testicles), as they may become cancerous. Finally, it is necessary to administer hormone replacement therapy in case the subject is assigned the female sex. "The prognosis of these patients is good if the testicles are timely removed", professor Mendoza states.

The Androgen Insensitivity Syndrome Guideline for physicians and patients is available at the web site of the Biomedical Research Network of Rare Diseases (CIBERER) www.ciberer.es, the Spanish Association of Human Genetics www.aegh.org and the research group on cancer in patients with polymalformation genetic syndromes (www.ct-csgp.org).

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