46XX male syndrome

R.L. Markosyan

Yerevan State Medical University, «Muracan» University Hospital, Yerevan, Armenia

OBJECTIVES

The XX male syndrome – Testicular Disorder of Sexual Differentiation (DSD) is a rare condition characterized by a spectrum of clinical presentations, ranging from ambiguous to normal male genitalia. The project "Genetics of Human Disorders of Sexual Development" is funded by Swiss National Science Foundation and fulfilled by the University of Geneva Medical School, the Medical Centers from Armenia, Poland and Ukraine.

The goal is to identify mutations underlying unresolved DSD phenotypes – in novel DSD genes, or regulatory regions that lead to atypical gene expression.

METHODS

During 2 years Armenian part collected 4 patients with SRY-positive 46, XX. The physical examination included the measurement of height, potential gynecomastia and the inspection of external sex organs. Bilateral volume was calculated as the sum of the volume of both testes. Serum levels of testosterone and Anti-Müllerian hormone (AMH) were assessed. Genomic DNA from peripheral blood of the patients using QIAamp DNA Blood Kits was extracted. The three discrete regions, AZFa, AZFb and AZFc, located on the long arm of the Y chromosome, were performed by multiplex PCRs (Polymerase Chain Reaction) amplification. The set of PCR primers for the diagnosis of microdeletion of the AZFa, AZFb and AZFc region included: sY84, sY86, sY127, sY134, sY254, sY255, SRY and ZFX/ZFY.

RESULTS

Our research reported that four patients had a female karyotype but were phenotypically male (46, XX males) and two of them were twins. They had normal external genitalia and masculinization, all males were SRY-positive, which translocated on the short arm of X chromosome, and absent of the spermatogenic factors encoding gene on Yq, such as AZFa, AZFb and AZFc region in Y chromosome. Endocrinological data indicated that the patients had a lower testosterone and a low level of AMH

CONCLUSIONS

Our reports adds cases on the four new 46, XX male individuals with sex reversal and further verifies the view that the presence of SRY gene and the absence of major regions in Y chromosome should lead to the expectance of a completely masculinised phenotype, abnormal hormone levels and infertility.

References

Text

Nef, S., Vassalli, J. D., 2009. Complementary pathways in mammalian female sex determina-tion. J Biol. 8, 74.

Ross, A. J., Capel, B., 2005. Signaling at the crossroads of gonad development. Trends En-docrinol Metab. 16, 19-25

Wainwright, E. N., Wilhelm, D., 2010. The game plan: cellular and molecular mechanisms of mammalian testis development. Curr Top Dev Biol. 90, 231-62.

Hiorta, O., Gillessen-Kaesbachb, G., 2009. Disorders of sex development in developmental syndromes. Endocr Dev. 14, 174-80.

55ESPE 431--P2

Gonads & DSD

Renata Markosyan

DOI: 10.3252/pso.eu.55ESPE.2016

Poster presented at: ESPE 2016 PARIS 10-12 SEPTEMBER