

ARTICLE INFORMATION

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Tetragametic chimera in a patient with sexual ambiguity

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ABSTRACT

Introduction: Human chimerism is the presence of two (2) or more cell populations in an individual that contain genetic material from more than one zygote. Phenotypes range from phenotypically normal males, sexual differentiation disorders, or phenotypically normal females. We present a case of tetragametic chimera in a patient with sexual ambiguity.

Materials and methods: 1-year-old patient with given urogenital malformation, sexual ambiguity, severe hypospadias, interscrotal urethral meatus, testicles in scrotum, renal ultrasound, and normal urinary tract. Cytogenetic study, microdeletions in Y chromosome, and analysis of STR in blood, buccal cells, hair follicle of the patient and samples of the parents.

Results: Chromosome study 46,XX[18], 46,XY[10]. STR analysis confirmed the presence of 2 cell lines in the different tissues. Analysis of segregation of STR from parents showed an identical maternal profile in all the samples analyzed, and a paternal profile represented by the contribution of gametes with different genetic constitution, which allows us to conclude that the tetragametic chimera originated by independent fertilization of an ovule and a second body. polar.

Conclusions: A case with sexual ambiguity due to tetragametic chimera with chromosomal constitution in peripheral blood 46,XX[18], 46,XY[10] is reported. Analysis of the segregation of STR from the parents allows us to conclude the origin of the tetragametic chimera by independent fertilization of an ovule and a second polar body.

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