

## TETRAGAMETIC CHIMERISM. NATURE RARITIES OR NOT AN UNUSUAL FINDING IN PATERNITY TESTING STUDIES

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During a routine paternity testing case we found two maternal and two paternal exclusions, as well as an XY profile in the maternal sample (17 STR analyzed). Once we verified the STR results obtained from a new maternal blood sample, we reviewed the OB-GYN history, carried out a physical exam (phenotypically is a normal female with regular menstrual cycles), and obtained new samples from different tissues to carry out molecular and cytogenetic studies. We also obtained blood samples from first degree relatives (children, grandmother, siblings). She has had 3 children from three different relationships. In the other two children, two maternal exclusions were also found based on blood STR typing. The cytogenetic studies showed a 46XY/46XX constitution in a 83:17 of the metaphases analyzed. mtDNA analysis showed identical sequences between the mother and the first child analyzed. Y chromosome haplotype analysis showed a full Y-chromosome profile different from the Y chromosome profile found in her son. The X chromosome STR analysis showed heterozygosity for the majority of X-chromosome STRs analyzed. DNA obtained from hair follicles, saliva, mouth cells, menstrual tissue and urinary sediment were obtained. All tissues showed an XX constitution with amelogenin. The STR maternal exclusions found when the blood sample was analyzed, were not found in the other tissues analyzed and the obligated maternal alleles were identified.

Based on our results, we conclude that the present case represents a tetragametic chimerism with a 46XX/46XY composition where a genetic profile predominates in blood, different than that obtained from different tissues. These results indicates that ovotestes are not present (a usual finding in 46XX/46XY individuals). Previously, we have reported a case of a 46XX/46XX phenotypically normal female (1). Our results indicate that cases where 2 or more maternal exclusions are found, exhaustive analysis should be carried out in order to rule out tetragametic chimerisms. In addition, cases where alleged fathers are found to be XX based on amelogenin typing and an exclusion of paternity is identified, should be thoroughly analyzed to rule out tetragametic chimerisms.

1) N, YU.; M, KRUSKALL; JJ YUNIS; J, KNOLL; U, LYNNE; S, ALOSCO; M, OHARSHI; O, CLAVIJO; Z, HUSAIN; EJ YUNIS; JJ YUNIS; EJ YUNIS. Exclusion of maternity leading to the identification of a phenotypically normal XX/XX tetragametic chimera. New England Journal of Medicine, USA, v. 346, n. 20, p. 1545-1552, 2002.