YOUR MOM IS NOT YOUR MOTHER - THREE INTERESTING CASES

Juan J. Yunis[1,2], Adriana Cuervo[1], Emilio Yunis[1]
[1]Servicios Médicos Yunis Turbay y Cia. Cra 24 # 42-24, Bogotá, Colombia
[2]Departamento de Patología, Facultad de Medicina, Universidad Nacional de Colombia. Grupo de Identificación Humana e Inmunogenética

Genetic chimerisms could be more frequent than we think. We have analyzed three different cases of tetragametic chimerism, the latest one during a routine paternity testing. In that case we found two maternal and two paternal exclusions, as well as an XY profile in the maternal sample (17 STR analyzed). Once verified, 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, and 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 were a genetic profile predominates in blood, different than that obtained from different tissues. Previously, we have reported a case of a 46XX/46XX phenotypically normal female (1) and worked on a very famous case featured on Discovery channel. Our results indicate that chimerisms could be more frequent than we think and that any unusual result should be investigated. These include XX males, XY females, the presence of additional alleles in different loci, since they could represent tetragametic chimerisms.

References:

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. ⌘