

AN UNUSUAL TYPE OF CONGENITAL GENETIC CHIMERISM IDENTIFIED FROM A DNA RELATIONSHIP TESTING TRIO

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Genetic chimerism refers to a condition where an organism's cells, and therefore genetic material, are derived from more than one distinct fertilization product. Nowadays, chimerism in humans is quite frequently encountered due to allogeneic hematopoietic stem cell transplantation, medical procedures that can create individuals who are "artificially" chimeric. In contrast, chimerism that is naturally established during fertilization or early embryogenesis, giving rise to what has been referred to as "whole body chimeras", is a rare phenomenon, with a limited number of examples described in the scientific literature. Tetragametic chimerism is the most common and best described example of congenital, whole body chimerism, and it occurs as a consequence of the fusion of two distinct post-zygotic entities. Genome-wide DNA analyses of individuals who are tetragametic chimeras reveal the presence of up to four apparent alleles, due to double haploid maternal and paternal genetic contributions.

An example of a different type of congenital chimerism is reported here, identified coincidentally when performing DNA relationship testing on a paternity trio (biological mother, child, and alleged father). DNA extracted from multiple buccal swab samples obtained from the child consistently revealed a mixture of two DNA profiles. Subsequent analysis of DNA extracts derived from individual hair follicles made it possible to tease out the two individual profiles, which both were consistent with inheritance from the biological mother and alleged father. Surprisingly, the mode of inheritance was consistent with double paternal, but only single maternal genetic contributions. This condition, which presumably occurs extremely rarely, has been previously observed and referred to as parthenogenetic chimerism.