CHIMERIC GERMLINE TISSUE: ALLEGED FATHER'S GENETIC CONTRIBUTION TO CHILD FOUND IN SEMEN SAMPLE BUT NOT IN BUCCAL SAMPLE

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Relationship testing is conducted when a biological relationship between individuals is in question such as child custody, child support and immigration cases. In the US over 382,000 legal relationship tests are ordered annually[1], using DNA based PCR assays that amplify short tandem repeat (STR) regions. Limitations of this assay have been noted[1,2]. We report a case of congenital chimerism revealed as a result of unusual paternity test results. Tetragametic chimerism is a condition that occurs when embryonic cells from dizygotic twins fuse early in development, leading to the birth of a singleton with two cell lines containing different genomes[2]. This condition was suspected in this case after a pregnancy conceived with intrauterine insemination revealed an exclusion of paternity, but ancestry testing revealed an avuncular relationship between the father and child. Multiple tissues from the alleged father were tested. The buccal sample from the alleged father was consistent with a single source DNA profile that excluded the child in question. Analysis of a semen sample from the alleged father revealed the presence of multiple sets of alleles at several loci, consistent with chimerism, and included the child in question. Additional tissues showed a similar multiallelic profile. A thorough review of the test types and tissue samples analyzed is provided. To our knowledge, this is the first reported case in which paternity was initially excluded by standard DNA testing methods and later included as the result of the analysis of different tissues. This case of chimerism yielding a false exclusion is thought to be unusual; however, with the uptake of assisted reproductive technology, this outcome could occur with increasing frequency[2].

Sources:
(1) Annual Report Summary For Testing in 2010. AABB Annual Report, pp. 1-9.
(2) Boklage, C. (2005). Embryogenesis of chimeras, twins and anterior midline asymmetries. Human Reproduction, 579-591.