

GENETIC CHIMERISM AND STEM CELL TRANSPLANTATION: HOW DOES IT AFFECT RELATIONSHIP & FORENSIC DNA TESTING?

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Genetically chimeric individuals are made up of cells that are derived from more than one distinct zygote lineage. Permanent chimerism can occur naturally as a consequence of the fusion of two zygotes or blastocysts into one organism early during development (tetragametic chimerism), or via the *in utero* transfer of blood stem cells between heterozygotic twins. Nowadays, with the continuously increasing medical use of allogeneic hematopoietic stem cell transplantations, procedures that are used mostly to treat hematologic and immunodeficiency disorders, artificial chimerism is the predominant cause of genetic chimerism in humans.

When performing relationship testing and forensic DNA analyses, the occurrence of genetic chimerism can have significant consequences for the interpretation of DNA profiles and the conclusions that are derived. DNA samples obtained from chimeric individuals often present with various degrees of profile mixtures, which can be highly dependent on the tissue of origin. Buccal swabs, blood, saliva, and hair are sample types commonly used for DNA profile analyses, and a meta-analysis of the observed relative donor/recipient chimerism will illustrate the variability within as well as across these four sample sources. Examples of DNA profiles obtained from artificial and natural chimeric individuals who submitted samples to the Identigene laboratory will also be presented.