

## AMELOGENIN Y NULL: A FIRST CASE REPORTED IN PUERTO RICO

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The Institute of Forensic Science of Puerto Rico currently performs STR analysis with AmpFISTR® Identifiler™ PCR Amplification Kit. Gender determination employs the Amelogenin locus from the kit. Previous cases of erroneous XX typing have been reported for Amelogenin locus from AmpFISTR kits. This study reports the first case of null Y typed in the Forensic DNA-Serology Laboratory in Puerto Rico.

A twelve-year old boy was murdered by accident while playing with a loaded gun in San Juan, Puerto Rico at the beginning of 2008. Evidentiary and reference samples, whole blood and hair, were organically extracted, quantified with Quantifiler® Human, amplified with AmpFISTR® Identifiler™, typed on ABI Prism 3100 Genetic Analyzer, and analyzed with GeneMapper® ID Software . A full profile was obtained for evidentiary and reference samples matching all STR'S genetic markers. The Amelogenin marker was typed as XX, with a complete drop-out of the Y allele on repeated extractions. The boy is phenotypically identified as a male despite the erroneous gender determination. These results suggest a possible deletion in the Amelogenin locus.

The samples were recently quantified using Quantifiler® DUO and amplified with AmpFISTR® Yfiler™ PCR Amplification Kit. Preliminary results show that while detection of the SRY region was successful with Quantifiler® DUO, only four markers were amplified with Yfiler™ on repeated extractions. These results indicate possible deletion in the SRY region amplified by AmpFISTR® Yfiler™. A sample from his father was analyzed with both AmpFISTR® kits and results will be discussed. Additional sequencing studies in the future will give a better insight about the type of mutations on the Y chromosome of the boy, as well as male family members.