

## **MURDERED MALE WITH AN INCOMPLETE Y CHROMOSOME**

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A 55 years old man was murdered with a knife, the suspect is a young man who was apparently his couple. After the crime the suspect killed himself.

During the autopsy, swabs from the oral, anal, penis and preputial cavity were taken and sent to the DNA lab. A bloody knife was found in a house where the suspect went after committing the crime.

In the preliminary tests, P30 protein was detected in both the anal and oral swabs. DNA extraction, quantification using Human and Trio DNA Quantification Kit and Global Filer DNA profiling from all the swabs, the knife and the victim's reference sample were performed .

On the analysed items, while plenty amounts of total DNA were detected, male DNA was almost in the detection limit. DNA profile from the oral, anal and preputial swabs, the knife and the reference sample did not amplified Y indel nor DYS391 markers, and the amelogenin amplified only the X allele: all the features of a female profile. Sample mixup was discarded, because the sample profile was obtained from several different evidences. Also, Costa Rican civil register was consulted for this person's sex definition and the custody chain of the evidence was corroborated. Pictures taken in the autopsy showed a person with no hair in its body nor in its face and anatomically presenting only a few male fenotypical characteristics.

Finally, samples were analysed using Y-filer Plus and from 27 markers, only 7 markers amplified (DYS627, DYS458, DYS19, DYS456, DYS449, DYS393, DYS481), corroborating the presence of the Y chromosome. Interestingly, all the markers that did amplify are located in the short arm of the Y chromosome, suggesting a possible deletion of a Y-chromosome region and/or a mutation near the AMEL Y binding site. Further analysis should be performed.

Additionally, a single source XY profile was obtained from the penis swab that excluded the principal suspect that leads to the hypothesis that a third person was present during the crime.

This uncommon phenomena must be remembered when working routine caseworks. Anticipated conclusions regarding sex assignment based only on amelogenin results should be avoided because confusion to the investigation can be introduced.