FORENSIC CASE: SEX DETERMINATION

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A case was received at the genetics laboratory of the National Institute of Legal Medicine and Forensic Cienses in Medellín, Colombia of an individual that looked like a man and was born with ambiguous genitalia. The authorities requested that we *"...perform the necessary tests in order to establish its real gender…",* We performed a quantification with Plexor® and the genetic profile with the PowerPlex® 16HS system. The results were a feminine profile and autosomical (8,40E1) and Y-chromosome (1,66E-1) quantification.

Methods

A reference sample of saliva was received at the institute and the methodology of extraction was Chelex 100 to 20%, then a quantification with Plexor® was made to establish the presence of human DNA for the subsequent obtainment of the genetic profile made with the PCR kit PowerPlex® 16HS system. The sample was analyzed by the Analyzer Genetic ABI PRISM® 3130 with GeneMapper® Software. All the procedures have been validated at the genetics laboratory of the National Institute of Legal Medicine and Forensic Cienses in Medellín, Colombia.

Discussion

In this case we obtained a feminine genetic profile and Y-chromosome quantification, this is an unusual result. This could be explained by the translocation of the SRY gene in the X-chromosome, this gene is responsible for the development of masculine sexual genitalia. In the genetic profile, do not appear Y-chromosome because the marker of AMG-Y is not present on it, that could be explained by the presence of some regions of this chromosome but not a whole Y-chromosome.

Conclusion

There is evidence that explains male phenotype with the presence of regions from Y-chromosome (SRY). Its is necessary to do a medical diagnosis and other test like FISH SRY or karyotype that would explain which region is generating the problem with its sex determination.

References

Validation of a Multiplexed system for quantification of human DNA and human male DNA and detection of PCR inhibitors on biological samples. Maura Barbisin *et al.* Journal of Forensic Sciences, 2009
Genes del cromosoma Y, Significado Clinico, Rafael Oliva, Universidad de Barcelona 2003
"What do the X and Y chromosomes tell us about sex and gender in forensic case analysis?", Von Wurmb Schwark N *et al.* Journal Forensic Leg Med. 2006. ⌘