Molecular analysis of patients suspected of Fragile X Syndrome

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ABSTRACT. The aim of this study was to validate the molecular genetic diagnosis of patients suspected of Fragile X Syndrome (FXS) in the Laboratory of Human Cytogenetics and Molecular Genetics (LaGene) of the Department of Health of the State of Goiás, using polymerase chain reaction (PCR). Thirty-five patients referred by public health doctors to LaGene, indicating clinical diagnosis of FXS, were selected for this study. Two PCR analyses were performed using different primers, one for screening (PCR-T) and one for the detection of the pre-mutation (PCR-P). The products of both PCRs were subjected to polyacrylamide gel electrophoresis and then coloring. The visualization of amplicons was performed with the aid of an ultraviolet transilluminator. The diagnosis was confirmed in 88% of patients with PCR-T and 100% with PCR-P. The primer used in PCR-P was

found to be more sensitive and specific, allowing to identify the mutation in the samples, generating a more conclusive case for FXS, noting that the PCR-T is also required for the pre-classification of patients. Generally, the PCR technique is cheaper and easier to handle; therefore, we suggest the implementation of PCR in the genetics laboratory of the State of Goiás (LaGene) for the diagnosis of FXS.

Key words: Fragile X Syndrome; Intellectual disability; PCR; PCR-T; PCR-P

