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Short Communication

Genotyping of Single Nucleotide Polymorphisms in Down syndrome

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ABSTRACT

Buccal epithelial cells extracted from saliva have been used as an alternative source of human DNA, especially from newborns and patients with mental illnesses. To ensure that appropriate conditions for DNA extraction, storage, and genotyping were created, healthy volunteers with no history of genetic or neurological disease were included as controls. The research involved 100 children with Down syndrome (DS), 50 of whom had oral motor problems. To achieve high-quality genomic DNA for screening gene polymorphisms by traditional and real-time PCR, a buccal swab and/or mouthwash technique is used before teeth brushing. For routine evaluation of genetic variations in DS patients, extraction of genomic DNA from buccal epithelial cells proved to be an effective, convenient, inexpensive, and noninvasive technique.

KEYWORDS: Down Syndrome, Trisomy 21, Chromosome Abnormality.

INTRODUCTION

The sample included 100 people with Down syndrome who visited the Instituto de Puericultura e Pediatria Martago Gesteira (IPPMG/ UFRJ) and five stable volunteers who did not have any identifiable genetic diseases. The Ethics Committee (CEP-IPPMG) approved the report, and the participants gave their written informed consent. The most common hereditary type of intellectual disability is Down syndrome (DS). The most frequent cause of DS is nondisjunction, but the molecular events that underpin this mechanism are unclear. It is important to recognise genetic variations that may play a role in the aetiology of DS. Single nucleotide polymorphisms (SNPs) of folate metabolising genes have been studied in communities all over the world as maternal risk factors for DS. DNA of sufficient yield and high quality of immediate importance or to be stored for future study is needed for genetic studies. To isolate DNA, a number of methods have been developed. The aggregation of buccal epithelial cells originating from saliva is an alternate source of human DNA [1]. Saliva collection has a variety of benefits over blood collection, including being painless and noninvasive, as well as being more appropriate, particularly for newborns and mentally retarded patients. Buccal epithelial cells from saliva may be a suitable substitute for supplying vast amounts of high-quality DNA for regular genetic screening aimed at detecting risk factors and diagnosing genetic disorders such as Down syndrome. Technical advances in molecular biology are making a significant contribution to clinical applications, especially in genetic diseases [2]. The consistency and quantity of DNA derived from biological samples was determined by a variety of factors, including extraction methods and storage conditions. While blood is the most common source of DNA isolation, it has its own set of limitations. Invasive and uncomfortable collection; possible risk of disease transmission; extra treatment in collection; and qualified clinicians to administer the operation are the key restrictions [3].

CONCLUSION

Buccal epithelial cells isolated from mouthwash with TNE bufferr or oral swabs yielded high-quality, healthy DNA samples. His technique yielded enough DNA for molecular analysis to identify gene polymorphisms in children with Down syndrome using both RFLP and real-time PCR.

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