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SCREENING OF *Adenomatous Polyposis Coli* (APC) GENE POLYMORPHISM USING ALLELE-SPECIFIC POLYMERASE CHAIN REACTION (ASPCR) METHOD

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ABSTRACT

APC gene polymorphism is a type of hereditary colorectal cancer (CRC) which is associated with familial adenomatous polyposis (FAP). FAP is manifested as polymorphism of APC gene that is involved in regulation of cell growth. Therefore, polymorphism of the APC gene will lead to reregulation of cell which is lead to cancer. In this experiment, DNA was extracted from the whole blood. The DNA extractions were then amplified by PCR amplification method adapted from the Perry S.C and Steven L.G, (2001). The ASPCR method was a specific method that used to detect the gene polymorphism in healthy or descendent sample that were potentially related to CRC. The primers used were HGH-5 and HGH-3 which were used in amplification of growth hormone gene. They were used to detect the internal control at 434 base pairs. While, primers of APC-1, APC-W (wild) and APC-M (mutant) were used in amplification of APC gene at 97 base pairs. The objective of this research is to screen and detect the APC gene polymorphism in human samples that are high risk in developing CRC. Early detection and diagnosis were crucial for initial prevention of CRC. The result in this experiment showed bands appeared at 434 bp indicated the internal control used for ASPCR method (Perry S.C and Steven L.G, 2001). Other distinct bands at 97 bp showed that all the samples are the homozygous wild type variant. None of the samples showed neither heterozygous nor mutant variant type. In a nutshell, ASPCR method can be used as one of the variant detection for APC gene polymorphism.

CHAPTER 1

INTRODUCTION

1.1 Background of Study

Colorectactal cancer (CRC) is the most common cancer among men and the third most common cancer among women in Malaysia. There are many factors that will lead to CRC, such as food, lifestyle, genetic aspect, alcohol and awareness value. Five to ten percent of CRC is caused by heritable mutation. Therefore, early detection and screening of CRC is important for initial treatment and to reduce the possibility of getting CRC.

There are many screening methods and treatment procedures that are available. By earlier detection and treatment, the percentage of surveillance is increasing and severity become lower. Examples of screening method are fecal occult blood testing (FOBT), sigmoidoscopy, Sanger sequencing, polymerase chain reaction (PCR) and allele specific polymerase chain reaction ASPCR