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AMPLIFICATION OF EXON 11 OF CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR GENE (*CFTR*) USING POLYMERASE CHAIN REACTION (PCR)

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ABSTRACT

Cystic Fibrosis, a very common autosomal recessive disorder, is caused by defects of CFTR protein. This protein is coded by *CFTR*, which contains 27 exons. A primer pair was designed to specifically amplify exon 11 of the gene. The primers were tested on one subject, and the amplicons were sequenced. One SNP, at location 470, was detected producing a non-synonymous mutation from guanine to adenine. AS-PCR and primer pair designed can be used in the future to screen larger group of sample to test for the frequency of the SNP or other mutations in exon 11 of *CFTR* in Malaysians.

CHAPTER 1

INTRODUCTION

1.1 Background of Study

One of the earliest mentions of cystic fibrosis (CF) was from a literature from German-Switzerland that originates somewhere between seventeen to eighteen century when translated claims that when a child is kissed on the forehead and tastes salty, he is hexed and soon will be dead (Lubamba, Dhooghe, Noel, & Leal, 2012). This is because CF patients have irregular chloride channel function making his sweat taste saltier than a normal individual.

CF is an autosomal recessive disorder that has a multi-organ involvement that commonly causes symptoms such as lung diseases, gastrointestinal complications, pancreatic insufficiency and to some lesser extent, diabetes, bone disease and infertility (Bowen & Hull, 2015). CF is a condition where cystic fibrosis transmembrane conductance regulator gene (*CFTR*) undergoes mutation. Around 2000 mutations have been reported on the gene, and some are confirmed in the database as CF-causing. Mutations that are CF-causing can interrupt CFTR protein function. The result of this is the disruption in regulation of the chloride channel and inhibition of sodium transport in the epithelium, which is two of the main functions