

## **Pola Mutasi DNA Mitokondria Manusia Daerah HVI Yang Mengandung Poli-C**

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### **Abstract**

Determination of human mitochondrial DNA (mtDNA) sequence is essential for studying both normal variant polymorphism and variants disease related. Through direct sequencing can be obtained nucleotide sequence data of 16024-16383, the first hypervariable segment (HVSI) of the mtDNA control region. However, this method can not be applied to sampel containing poly-C stretch because of T16189C mutation. This thesis reported the nucleotide sequence of HVSI mtDNA for sampels containing poly-C stretch using cloning method. The strategy of this research included mtDNA amplification using Polymerase Chain Reaction (PCR) technique, recombinant DNA cloning, and sequencing using Dideoxy Sanger method. The result showed that that there are other mutation outside T16189C of three samples, they are XXAM, ESG, and GMR. Two substitution mutation at 16182 16183, and five mutation of insertion at 16059, 16074, 16041, 16188 and 16193. The existence of 16188 and 16193 mutation related to main mutation (T16189C) yield yield different pattern of poly-C. The differences are 10[C], 11[C], 13[C] dan 15[C] for C4B, GMR, ESG, and XXAM. Through this method have been obtained specific information of individual identity whose have T16189C mutation that can not read by direct sequencing. Hence, the database of HVSI containing poly-C stretch has been added for the nomenclature of human mtDNA.

***Keyword : Mitochondria DNA, Poli-C, direct sequencing, cloning, HVSI***