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參展科別 **醫學與健康科學**

作品名稱 **Understanding the Modern Diagnoses of Protein C Deficiency "Pcd" with Unknown Gene Plays a Critical Role in the Inherited Thrombophilia**

得獎獎項 **三等獎**

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Abstract

Protein C deficiency (PCD) is found in 1 out of 200 to 500 persons in the general global population which is also one of the common conditions of Inherited thrombophilia, it's characterized by an increased tendency of blood to clot in human blood vessels. It is caused by several factors including mutations in the genes involved in thrombin binding, protein c activation and numerous clotting factors. This includes F5 (Factor 5 Leiden) gene on chromosome 1q24.2, F7 (Prothrombin) gene on chromosome 13q34, SERPINC1 (serpin peptidase inhibitor C) on chromosome 1q25.2, SERPIND1 (serpin peptidase inhibitor D) on chromosome 22q11.21, HRG (Histidine Rich Glycoprotein) on chromosome 3q27.3, PLAT (Plasminogen Activator) on chromosome 8q11.21 and THBD (Thrombomodulin) gene on chromosome 20p11.21. In the current study, a three Saudi families with inherited thrombophilia has been recruited to identify the underlying cause of this special condition. Whole exome sequencing, targeting all coding exons of the human genome, was performed using Illumina Nextera library preparation kits followed by paired-end sequencing on Illumina NextSeq500 instrument. Reads quality control was performed and reads were aligned to the reference genome using BWA software. Variants calling and annotation was performed using GATK. All known genes involved in causing inherited thrombophilia All known genes involved in causing PCD were excluded by whole exome sequencing. The genes that were previously reported to be involved in inherited thrombophilia were checked for any causative variant. No mutation has been identified in known genes. identifying a novel gene underlying PCD. The Result of this study will hopefully pave the way to better understanding the disease pathophysiology and help in developing DNA based diagnosis, carrier screening and somatic gene therapy.

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The presenter uses modern technology to study an inherited disease in Saudi Arabia, He has learned the techniques of DNA extraction, NGS sequencing, and DNA analysis. He also learned the analysis of patient's family pedigree and their disease association. Using a candidate approach method, he identified the mutation in one family. This is very nice work. The whole-exon sequencing technology is easy to perform nowadays because of the reduction of the cost of DNA sequencing. Bioinformatics analysis of the sequence data, however, requires more work. The presenter can try his future research toward the bioinformatics analysis. It will be very important work.