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## How molecular biology can help in finding the origin of genetic defects in different populations

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Molecular biology has become an integral figure in the modern medicine, making DNA analyses as essential tools in the diagnosis, prognosis and management of diseases. Factor V Leiden (FVL) is the name of the most common genetic mutation which is associated with venous thrombosis, a disease that has high morbidity and mortality rates. Earlier studies on FVL found it in Europeans only, bringing speculations that FVL had occurred as a single event in the far past, in a single European ancestor who fixed the mutation in the current European carriers. However, our later research in Kuwait proved the presence of FVL in different non-European populations living in Kuwait. Does this mean these were originally from Europe? In this research, we showed how molecular techniques were used to explore the origin of FVL in different populations living in Kuwait. Was it the same European ancestor, or a separate one? A total of 512 healthy individuals were recruited from different populations (non-European) living in Kuwait: 360 Arabs (Kuwaitis, Lebanese, Syrians, Jordanians, Saudis, Iraqis, Palestinians, and Yemenites), 102 Armenians and 50 Afghans. A blood sample was collected from each case which

was used for DNA extraction. Real-time PCR was performed on the DNA samples to test for the presence of FVL. In the positive cases for FVL, real-time PCR was performed to explore 9 single nucleotide polymorphisms (SNPs) in the Factor V gene; these SNPs were previously reported to be associated with FVL in European carriers of the mutation (in linkage disequilibrium with FVL). The same was done on a randomly selected number of the negative cases (equals to the number of the positive cases). 99 of the 512 cases were found positive for the FVL mutation. When the 9 NPs were analyzed, all our positive cases had the same 9 alleles that were present in Europeans. However, this was not true in the negative cases. The results indicate that our positive cases in Kuwait had most probably descended from the same proposed European ancestor who had the FVL mutation event. Further studies are planned to perform additional molecular tests and combine our results with the available epidemiological data and anthropological knowledge to possibly determine how this mutation had reached Kuwait.

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