SOA: Fibrinogen gene variants in monogenic and complex diseases

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Abstract

Fibrinogen is encoded by a three gene cluster, *FGB, FGA* and *FGG*, located in humans on the long arm of chromosome 4. More than 250 distinct causative mutations for the monogenic congenital fibrinogen disorders afibrinogenemia, hypofibrinogenemia (quantitative disorders), hypodysfibrinogenemia and dysfibrinogenemia (qualitative disorders) have been identified. This field of research has generated valuable and conclusive information on structure-function relationships for the individual fibrinogen chains, the soluble fibrinogen hexamer as well as fibrin. Fibrinogen gene variants have also been studied in association with the variability of circulating fibrinogen levels, which is linked with cardiovascular disease risk. This lecture will give a State of the Art overview of what is known on the role of fibrinogen gene variants in monogenic and complex diseases.