The role of molecular biology in the diagnosis of impaired haemostasis

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Routine genetic testing in inherited bleeding disorders is now mostly limited to relatively common disorders such as haemophilia A and B for which genetic counselling is well organized. With the advent of next generation sequencing technologies (NGS) the inclusion of genetic testing into the clinical care path of patients with rare inherited platelet and thrombotic disorders is coming closer.
In order to facilitate the introduction of NGS in the diagnosis of impaired haemostasis the ISTH and SSC have started a Thrombogenomics working party that brings together clinical medicine, genomics, computational biology and statistical genetics. The primary aim of this working party is to develop resources and NGS knowledge to support the introduction of DNA-based diagnosis.
This has resulted in a web-based database with 69 genes that are curated by gene experts around the world. Furthermore, a technology platform is being developed for selective capture of these 69 genes for NGS analysis. This platform that allows the simultaneous testing of all genes known to be associated with inherited bleeding disorders in one NGS test will be made available in the foreseeable future.