Genetic mechanisms of inherited bleeding disorders as a basis for personalised medicine approaches

Propositions belonging to the dissertation:

Genetic mechanisms of inherited bleeding disorders as a basis for personalised medicine approache

1. mRNA analysis has added value beyond DNA and protein analysis. (this thesis)
2. VWF propeptide persistence affects FVIII binding as well as VWF multimerization. (Casonato et al. Blood 2003 and this thesis)
3. A nonsense mutation is not necessarily a null mutation. (this thesis)
4. Plasma corrects the consequences of FV deficiency, but molecular therapies could correct its causes. (this thesis)
5. Since TFPI contributes to bleeding in several coagulation factor deficiencies, pharmacological modulation of FV-short splicing might be beneficial in multiple bleeding disorders. (this thesis)
6. Genetics plays a role, to a greater or lesser extent, in all diseases. (M. Jackson, *Essays in Biochemistry*. 2018)
7. Given the double role of FV in the coagulation process, deficiencies in FV may result in either haemorrhagic or thrombotic tendencies. (R. Asselta, *Semin Thromb Hemost.* 2009)
8. Rare diseases are rare, but rare disease patients are numerous. (Orphanet)
9. Actions speak louder than words; let your words teach and your actions speak. (St. Anthony of Padua)

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24th April 2024