Clinical, biological, and genetic features in an afibrinogenemia patient series in Algeria

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Introduction: The incidence of afibrinogenemia had not been previously reported in Algeria. Afibrinogenemia patients are prone to both haemorrhagic and thrombotic complications. Predictive markers of thrombosis in afibrinogenemia patients are not existent.

Aims and methods: Clinical and biological data from 46 afibrinogenemia patients are reported. Biological investigations included routine tests, genetics analysis and thrombin generation.

Results: FGA mutations (four novel and four previously described) and FGB mutations (seven mutations; five novels) were homozygous in all but one family as a result of 28 consanguineous marriages out of 30 discrete families. Incidence of afibrinogenemia in Algeria is at least 3 per million births. Umbilical bleeding was reported in 39/46 cases and was the main discovery circumstance. We also report post trauma or post-surgery (3/46) bleeding and spontaneous deep vein thrombosis (DVT) in adulthood (1/46), as discovery circumstances. The median age (10.5-year-old) of the population reported here explains why there are few hemarthrosis and obstetrical or gynaecological complications in this series. Thrombotic events were reported in seven patients (four spontaneous). Endogenous Thrombin Potential was significantly increased in thrombosis-prone patients compared to afibrinogenemic patients with and without personal or familial history (1118 vs. 744 and 817 nM IIa × min, respectively).

Conclusion: The incidence of afibrinogenemia in Algeria is the consequence of consanguineous marriage in families carrying private mutations. The thrombin generation test (TGT) could identify, among afibrinogenemic patients, those presenting a thrombotic risk.

Keywords: afibrinogenemia; bleeding complications; fibrinogen mutations; thrombin generation; thrombosis.