Clinical Case Study

A 24-Year-Old Man with Previously Diagnosed Hemophilia

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CASE

A 24-year-old Middle Eastern man diagnosed with hemophilia at the age 4 or 5 years presented to the hematology clinic for follow-up after a recent hospitalization for excessive bleeding from an accidental knife cut. The patient reported a history of prolonged bleeding after teeth extractions, an upper gastrointestinal bleed 3 years previously, and excessive bruising since childhood. He denied hemarthroses but reported chronic pain in his ankles and joints. The patient reported having been treated with fresh frozen plasma (FFP)[3] and factor VIII during past hospitalizations. Because of poor continuity of care, his disease had not been monitored or treated on an ongoing outpatient basis. The patient's family history is noteworthy for consanguineous parents (first cousins) and a sister who also experienced excessive bleeding, although her diagnosis was uncertain. Initial laboratory test results included normal complete blood count, including platelets, a prolonged activated partial thromboplastin time (aPTT), and a prolonged prothrombin time (PT) (Table 1). Fibrinogen activity was normal. A 1:1 mixture of the patient's plasma with pooled normal plasma demonstrated full correction of the PT and aPTT, a result consistent with factor deficiency.

Questions to Consider
• How do the coagulation studies for this patient differ from those typically seen for patients with hemophilia?
• What are possible causes of simultaneous prolonged PT and prolonged aPTT?
• What further coagulation studies would you recommend in evaluating this patient?

DOI: 10.1373/clinchem.2011.166728

Table 1. Patient's laboratory results (citrated plasma).

	Result	Reference interval
aPTT, s		
Patient	70.6	22.5–31.3
Pooled normal plasma	26.3	22.5–31.3
Mixture (1:1), s	29.3	22.5–31.3
PT, s		
Patient	19.2	9.4–11.5
Pooled normal plasma	10.7	9.4–11.5
Mixture (1:1)	11.5	9.4–11.5
Factor activities, %		
FII	139	69–140
FV	5	70–154
FVII	145	72–131
FVIII	4	56–172
FIX	113	69–176
von Willebrand factor	59	50–125
von Willebrand factor antigen, %	81	≥50
Fibrinogen, mg/dL (μmol/L)	308 (9.0)	180–400 (5.3–11.8)

Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the July 2012 issue of *Clinical Chemistry*. To view the case and comments online, go to http://www.clinchem.org/content/vol58/issue7 and follow the link to the Clinical Case Study and Commentaries.

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