TITLE:Hemophilia A prenatal diagnosis by factor VIII clotting activity using percutaneous umbilical blood sampling

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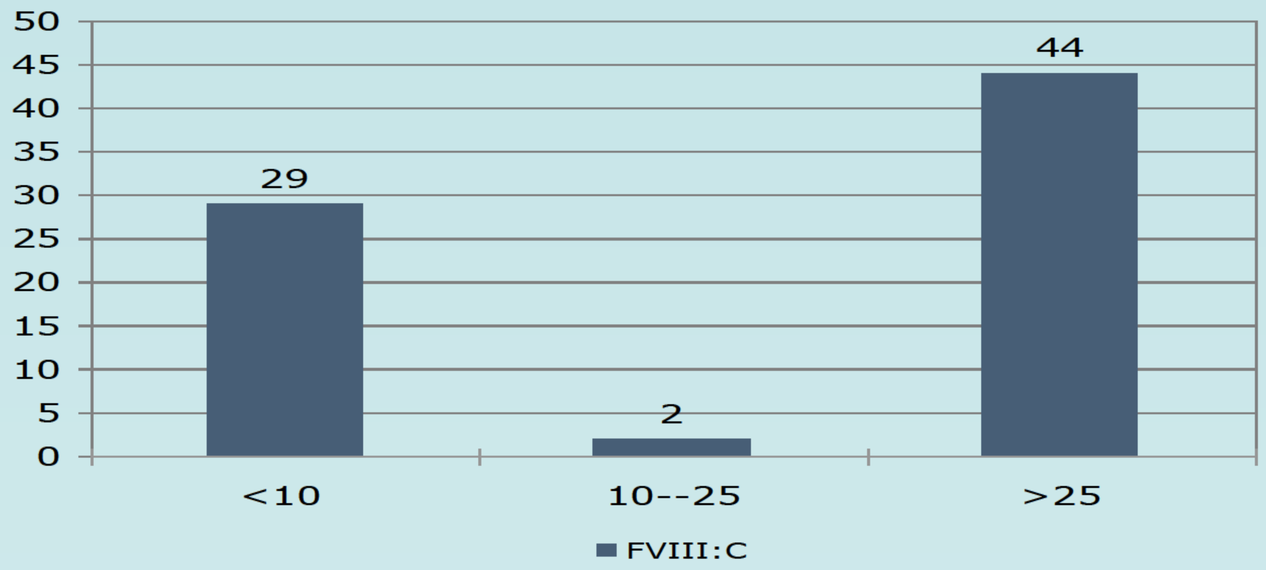
Objectives:

Prenatal diagnosis for pregnant hemophilia A carrier or putative carrier using phenotypic diagnosis with percutaneous umbilical blood sampling(PUBS).

Methods:

Seventy-five hemophilia A carrier or putative carrier with male fetuses at risk of hemophilia age from 21 years old to 42 years old at 20 to 34 weeks' gestation. Obtained fetal blood sample by percutaneous umbilical blood sampling (PUBS), fetal haemoglobin alkali denaturation test to prove fetal blood samples, abandon the first 0.5ml blood sample to avoid amniotic fluid contamination. Then spun the samples at 2500rpm for 20 min at 4°C for coagulation factor test. Measured factor VIII and IX coagulant activities in automated coagulometer immediately after spun by one stage assay method.



Results:

Twenty-nine abnormal prenatal tests (FVIII:C <10u/dl) in 75 of male fetuses at risk of hemophilia and fourty-four normal prenatal tests (FVIII:C >25u/dl) in 75 of male fetuses at risk of hemophilia. All the blood samples with FIX:C <20 u/dl, if fetal blood samples FIX:C were high, there may be maternal contamination. Only two fetuses in FVIII:C 10-25u/dl group, one fetus with FVIII:C 10.6u/dl, recommend test again one month later, but the pregnant woman insisted on termination of gestation. The other fetus with FVIII:C 21.3 u/dl at 25weeks, one month later FVIII:C become 50.2u/dl. All normal prenatal tests were followed up in all children and there were no misdiagnoses.

Conclusions:

When the linkage of F8 mutation is not informative or patients were referred to the laboratories late in their gestation, test factor VIII clotting activity using percutaneous umbilical blood sampling offer a quick and robust diagnosis method.

References:

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DOI: 10.3252/pso.eu.WFH2012.2012