Prenatal Diagnosis of Severe Inherited Coagulation Factor VII Deficiency

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Objective
To establish a reference range for levels of FVII:C in cord blood in healthy human fetus. To provide prenatal diagnosis for fetus in a family with rare congenital factor VII deficiency, based on a combination of molecular genetic diagnosis and comparing the FVII:C levels of the fetus with unaffected pregnancies.

Methods
Fetal blood samples were obtained and the FVII:C levels detected in 31 unaffected pregnancies with a gestational age of 22-32 weeks. Abnormal karyotypes were excluded and a reference range was built. DNA sequencing was performed to detect mutations in the parents. Fetal FVII gene were examined by Sanger method and MLPA to exclude maternal cell contamination through linkage analysis. FVII:C levels of the target fetus were analysed at 26 weeks gestation and at birth.

Results
Average FVII:C levels in 30 normal pregnancies were 36.8% ranging from 21.5% to 64.6%. The FVII:C levels of fetus in study is 27.5%. A Paternal FVII gene was found; a splice mutation c. 681+1G>T. A missense mutation c. 1028G>A was detected in maternal FVII gene. The target fetus did not carry the familial mutation.

Conclusion
We have established a reference range of FVII:C levels in normal fetus (21.5%—64.6%), and fetal FVII:C levels increase slightly with increasing gestational age. The FVII:C level of the fetus in study lie in normal reference and mutation was not detected in the fetal FVII gene. This is the first report of prenatal diagnosis of inherited coagulation factor VII deficiency in our country.