Congenital factor x deficiency
Association between coagulant factor activity and clinical bleeding severity in four cases

Zahra Badiei[1], Hamid Farhangi[2] , Mohammad Reza Keramaty[3], Masoud Shahroudian[4], Elahe Yasi[5], Ali Ghasemi[6]

[1] MD. Associate Professor of Pediatric Hematology-Oncology, Hemophilia-Thalasaemia Center of Mashhad (Sarvar Clinic),Mashhad University of Medical Science, Mashhad, Iran

[2] MD. Assistant Professor of Pediatric Hematology-Oncology, Hemophilia-Thalasaemia Center of Mashhad (Sarvar Clinic),Mashhad University of Medical Science, Mashhad, Iran

[3] MD. Associate Professor of Hematopathology, Hemophilia-Thalasaemia Center of Mashhad (Sarvar Clinic),Mashhad University of Medical Science, Mashhad, Iran

[4] Laboratory Specialist, Hemophilia-Thalasaemia Center of Mashhad (Sarvar Clinic), Mashhad University of Medical Science, Mashhad, Iran

[5] MD. Pediatrician , Hemophilia-Thalasaemia Center of Mashhad (Sarvar Clinic), Mashhad University of Medical Science, Mashhad, Iran

[6] MD. Assistant Professor of Pediatric Hematology-Oncology, Hemophilia-Thalasaemia Center of Mashhad (Sarvar Clinic),Mashhad University of Medical Science, Mashhad, Iran

Introduction and Objectives:

The congenital FX deficiency is a rare autosomal recessive disorder that usually presents with various hemorrhagic diathesis correlated with FX levels and prolongation of the PT and aPTT as seen in hemorrhagic disease of the newborn.

According to recent report of Peyvandi and her colleagues: patients with activity levels <10% of FX had a high risk of major spontaneous bleeding such as CNS and GI bleeding; suggesting this as a target level for prophylaxis. However, patients with levels >40% remained largely asymptomatic and those having levels between 10 and 40% only suffered minor spontaneous or triggered bleeding.

Materials and Methods:

Here we report 4 cases of congenital FX deficiency who are registered in our center during recent 10 years

1- Male, DOB 2004/04, parents are cousins. He is the second child of his family and has a 13 y old healthy sister. His mother has the history of 3 abortions (one in 2nd and two in 6th months of pregnancy). First sign: bleeding after BCG vaccination that was not controlled with local measures so coagulation screening tests were done and revealed PT > 120 and aPTT >140 sec, bleeding was controlled with FFP.

He experienced other bleedings: umbilical cord bleeding at 7 days old which controlled with local measures, intra-ventricular hemorrhage (IVH) at 26th day of life and control with transfusion of FFP. When he was 3 month old the diagnosis of FX deficiency was done but second ICH occurred at this time. He received treatment with FEIBA (there was no other PCC product in our centers at this time) and after improvement he put on prophylactic treatment by FFP 10cc/kg every other day. After 1 Y old he uses PCC (Beriplex® P/N 250 [PPSB], ZLB Behring) two times per week. At this time his condition is normal.

2- Female ,DOB 2007/03 , parents are not related , first sign : ICH at first month of life with PT > 50 , PTT=97 ,BT=4 , FX< 1% , prophylactic treatment with PCC was begun for her after resolving of ICH. Unfortunately her family didn't continue prophylactic treatment and she was died at 10 month old.

3- Male, Dob:2010/9 ,parents are cousins , history of GI & CNS bleeding in first child who was died and then she had two abortions .

This child had subgaleal hematoma and anemia at birth and after blood transfusion discharged, after 10 days he developed bleeding from umbilical cord and at this time diagnosis of his disease was done.

Unfortunately at 20 days old he experienced ICH but with proper treatment it was resolved and he put on prophylactic treatment by PCC (UMANNA COMPLEX®) two times per week. His Lab results include : PT>50 PTT=85 FX < 1%.

4- Female ,DOB 2011/07 ,parents are cousins ,first sign was bleeding after BCG vaccination, diagnosis was done at 7 days old with PT=32, PTT=60 , FX < 1% , Fibrinogen= Normal .When she was 1 month old developed ICH and after that prophylactic treatment was begun with PCC (UMANNA COMPLEX®).

Results:

They include 2 male, 2 female with age range 2 - 10 years. All of them had FX level <1 IU/dl and experienced **ICH** during first three month of life, three of cases are the first child of their family.

Three of them are result of consanguineous marriage and in one family there is history of GI & CNS bleeding and death of first child at 5th day of life.

Three of them are on prophylactic treatment with PCC /twice per week and one has expired at 10 month old because her family didn't continue prophylactic treatment. All of these families are concerned about having another FX deficient child, so avoid other pregnancy.

Conclusion:

Because marriage between close relatives is common in our country it seems that we must try to provide facilities of prenatal diagnosis for these families and their relatives who are at risk of having children with severe forms of bleeding disorders.

Key words :

FX deficiency, Congenital, Bleeding severity

References:

1) *Uprichard J, Perry DJ. Factor X deficiency. Blood Rev 2002; 16:97–110.*
2) *Herrmann FH, Wulff K, Lopaciuk S, Pollmann H. Two novel Factor X gene mutations in severe Factor X deficiency-Greifswald Registry of Factor X deficiency. Thromb Haemost 2001;1(S1) [Abstract P1126].*
3) *Wulff K, Herrmann FH. Molecular genetic analysis in the Factor X gene in patients with congenital FX deficiency. Thromb Haemost 2003; 1(S1) [Abstract P1201].*
4) *G. Auerswald. Prophylaxis in rare coagulation disorders—factor X deficiency; Thrombosis Research (2006) 118S1, S29—S31*
5) *Karimi M. et al. Phenotype and genotype report on homozygous and heterozygous patients with congenital factor X deficiency . haematologica | 2008; 93(6).*
6) *P. H. B. BOLTON-MAGGS.et al. The rare coagulation disorders – review with guidelines for management from the United Kingdom Haemophilia CentreDoctors' Organisation ; Haemophilia (2004), 10, 593–628*
7) *F . Peyvandi, D. DI Michele.et al. Classification of Rare Bleeding Disorders (RBDs) based on the association between coagulation factor activity and clinical bleeding severity. Journal of Thrombosis and Haemostasis,Vol 10, No 9,2012: 1938–1943*