

# INVESTIGATION OF POSSIBLE CORRELATION BETWEEN CLINICAL AND LABORATORY PHENOTYPE IN CONGENITAL FXI DEFICIENCY: RESULTS FROM A SINGLE CENTER

C. Santoro<sup>1</sup>, F. Barone<sup>1</sup>, G. Ferrara<sup>1</sup>, R.A. Bochicchio<sup>1</sup>, M.P. Bicchichi<sup>2</sup>, M. Acquila<sup>2</sup>, E. Baldacci<sup>1</sup>, F. Paoloni<sup>3</sup>, M. Chisini<sup>1</sup>, M.G. Mazzucconi<sup>1</sup>

<sup>1</sup> Hematology, Sapienza University, ROME, Italy

<sup>2</sup> Laboratorio Centrale Analisi. Istituto G. Gaslini. Genova

<sup>3</sup> Fondazione GIMEMA

## Introduction:

Bleeding phenotype (BP) in FXI deficient patients (pts) is not correlated with FXI:C level. There is no optimal predictive laboratory test to assess bleeding risk in such pts.

## Aim:

To analyze whether platelet function testing and global coagulation assays can serve as clinical tools in predicting BP in FXI deficient pts.

## Patients and Methods:

The ISTH-Bleeding Assessment Tool (BAT) was used to classify bleeders (score >3 for males, >5 for females) and no-bleeders. To explore platelet function, PFA-200 System and Light Transmission Aggregometry (LTA) were utilized. Thromboelastography (TEG) and Calibrated Automated Thrombography (CAT) were performed. The association between continuous variables and BP was tested by means of logistic regression model. Pts were evaluated also for molecular biology.

## Results:

- We enrolled 25 pts: 7 bleeders, 18 no-bleeders. Bleeders median FXI:C: 10.1% (4.3-46%); no-bleeders median FXI:C: 27% (6.2-67%).
- No significant association was found between FXI:C levels and BP (OR 0.968; 95% CI 0.92-1.01; p=0.19).
- **PFA-200:** a trend was observed, for association between BP and Collagen/ Epinephrine cartridge test results (p= 0.052).
- **LTA:** a trend was observed, for association between BP and ADP 5 μM (p 0.07) and Adrenaline 5 mM (p 0.07) test results.
- **CAT:** no significant associations were found with BP: endogenous thrombin potential, p 0.43; lag time, p= 0.37; peak, p= 0.28; time-to-peak, p= 0.42.
- **TEG:** no significant associations were found with BP: R, p= 0.58; α-Angle, p= 0.26; MA, p= 0.38; Ly30, p= 0.85; Ly60, p= 0.36.
- **Molecular biology:** 18 pts were studied. Seven had homozygous mutations, 3 compound heterozygous mutations, 6 heterozygous mutations. In 2 pts mutations were not detected.

## Conclusion:

We confirm variability in BP among FXI deficient patients, not significantly related to FXI:C levels. We found a trend for platelet dysfunction (PFA200 and LTA) and BP. We did not find any significant association between BP and TEG or CAT parameters.

Enrolled patients	25 (7 M, 18 F)
Median FXI:C (%)	24 (4.3-67)
Bleeders	7
Bleeders Median FXI:C (%)	10.1 (4.3-46)
No bleeders	18
No Bleeders Median FXI:C (%)	27 (6.2-67)

## INNOVANCE PFA – 200 System

Phenotype	n	Median FXI:C level (70-140%)	Median Coll/EPI (91-168 s)	Median Coll/ADP (61-123 s)
No - Bleeders	18	27.0 (6.2-67)	142.0 (89-166)	84.5 (66-223)
Bleeders	7	10.1 (4.3-46)	108.0 (81-207)	91 (56-125)

## Light Transmission Aggregometry (LTA)

Phenotype	n	Median FXI level (70-140%)	Median ADP (49-100%)	Median Epinephrine (53-100%)	Median Collagen (68-94%)	Median Arachidonic Acid (70-100%)
No - Bleeders	18	27.0 (6.2-67)	90.0 (69-100)	82.5 (55-97)	87.5 (62-97)	90.0 (68-100)
Bleeders	7	10.1 (4.3-46)	92.0 (34-99)	82.0 (8-92)	82.0 (61-94)	92.0 (54-96)

## CAT

Phenotype	n	Median FXI:C (70-140%)	Median Lag time (4.3 – 9.4 min)	Median ETP (379.2 – 909.5 nM x min)	Median Peak (27.1 – 90.7 nM)	Median tt-Peak (9.6-16.1 min)
No - Bleeders	18	27.0 (6.2-67)	5.2 (3.7-11.7)	522.8 (228.1-840.4)	47.8 (15.5-97.0)	10.7 (7.9-21.5)
Bleeders	7	10.1 (4.3-46)	5.3 (4.7-12.5)	464.6 (226.2-808.2)	41.7 (19.3-69.1)	12.2 (9.7-18.5)

## TEG

Phenotype	n	Median FXI:C level (70-140%)	Median R (4.3 – 6.3 min)	Median Alpha Angle (57.2 – 70.8 °)	Median MA (59.8 – 70.5 mm)	Median Ly30 (0.2 – 3.8 %)	Median Ly60 (3.1 – 14.6 %)
No - Bleeders	18	27.0 (6.2-67)	9.4 (6.1-68.2)	60.9 (14.8-70.1)	60.4 (34.9-70.6)	1.2 (0-11.8)	8.0 (0-38.8)
Bleeders	7	10.1 (4.3-46)	20.3 (5.2-60.1)	46.8 (11.7-67.3)	57.4 (27.1-69.1)	2.3 (1.2-5.7)	11.0 (6.9-25.8)



Poster Presented at:

DOI: 10.3232/jes.111.FF0210.2016

Rare bleeding disorders  
Cristina Santoro

202--MP-M  
9T0ZHM

9T0ZHM