Congenital Thrombasthenia In Children Welfare Teaching Hospital: A Descriptive Study

* Lubna Foad Hussaia, *Obeida Amir Abid

ABSTRACT

Background: Thrombasthenia is an inherited genetic disorder affecting platelets, which is characterized by spontaneous muco-cutaneous bleeding and abnormally prolonged bleeding in response to injury or trauma. **Objectives:** The aim of this study was to assess the diagnosis and treatment of thrombasthenia in Children Welfare Teaching Hospital.

Type of the study: A cross-sectional study.

Methods: This descriptive study was performed on 66 patients with thrombasthenia from the first of October 2013 till the first of July 2015.The diagnosis of the disease was done by a wide spectrum of characteristics including family history, clinical manifestations, laboratory tests..

Results: The common manifestations of the disease at time of diagnosis were epistaxis in 23 patients (34.8%) and gum bleeding in 22 patients (33.3%) .Mostly established at age 1-10years.Family history was positive in most patients.hepatitis C was rare in our patients after platelet transfutions.

Conclusions: : It is a rare hemorrhagic disease in our center, mostly presented as minor bleeding involving mainly mucocutaneous regions which appears at an early childhood with positive family history in most cases.

Keywords: Thrombasthenia, epistaxis, platelet transfusion.

Al-Kindy College Medical Journal 2016: Vol.12 No.2
Page: 95-97

** Specialist Pediatrician, Children Walefare Teaching Hospital, Medical City , C.A.B.P. , F.I.B.M.S (Pediatrics).*

Received 4th April 2016, accepted in final 22th June 2016
Corresponding to Lubna Foad Hussaia

Thrombasthenia (Glanzmann's thrombasthenia and Bernard-Soulier syndrome) is a rare hemorrhagic disorder characterized by abnormal platelet function. It is inherited as an autosomal recessive trait, patients are born with a spontaneous mucocutaneous bleeding syndrome that is variable in both frequency and intensity but which on rare occasions is life-threatening [(1)]. Thrombus formation fails to form as platelets lack or have non-functional receptor of platelets that mediates the final step of platelet aggregation induced by physiologic agonists.It is caused by an abnormality in the genes for glycoproteins IIb/IIIa and Ib/IX/V.

The aim was to study the diagnosis and treatment of thrombasthenia in Children Welfare Teaching Hospital, Medical City, Baghdad.

Methods: It was an observational descriptive study. The study participants included all patients who were admitted and discharged from the hemophilia center in Children Welfare Teaching Hospital with the diagnosis of thrombasthenia from October 2013 till July 2015. Platelet count and initial coagulation screen PT ,APTT were found normal in all patients. Data collected includes : Gender, age at presentation, presenting complaint, family history, type of treatment (platelet transfusion, cyclokaprone), place , relation of gender with the type and time of presentation, hepatitis B ,C in patients and its relation to platelet transfusion. Diagnosis were confirmed by performing bleeding time which was prolonged; and positive platelet function test (PFA-100) which is a system for analysing platelet function in which citrated whole blood is aspirated at high shear rates through disposable cartridges containing an aperture within a membrane coated with either collagen and epinephrine or collagen and ADP.These agonists induce platelet dhesion, activation and aggregation leading to rapid occlusion of the aperture and cessation of blood flow termed the closure time.This time is prolonged in patients with thromasthenia [(2)].This analyzer was produced by BIO/DATA corporation.

Statistical analysis was done and results were expressed as frequencies and percentages and presented in tables and figures.

Results: Among 778 patients evaluated for bleeding tendency, 66 patients were diagnosed to have thrombasthenia. Out of these 66 patients, 36(54.5%) were males and 30 (45.4%)were females.The diagnosis of thrombasthenia was established mostly at the age of 1-10 years, 34patients (51.5%) .The relation of gender with the time of presentation is shown in **Figure1**

Family history was positive in 39 patients(59%) and negative in 27patients(40.9%). **Figure2**

Epistaxis was the major presenting complaint in 23 patients (34.8%) , followed by gum bleeding in 22 patients(33.3%) and ecchymotic spots in 14

patients(21.2%) ,while only 3 females (10%) with menorrhagia. The relation of gender with the type of presentation is shown in **Table1**

The products used in these patients included platelets transfusion. Testing for transmission of viral infections was also done in these patients and 4 patients (6%) were found with hepatitis C positive .**Figure3**

The type of thrombasthenia was diagnosed to be Glanzmann's thrombasthenia in 30 patients (45.4%) and 9 patients(13.6%) with Bernard-Soulier syndrome While it was not recognized in the remaining 27 patients (40.9%). **Figure4**

Figure1:The age of diagnosis in the studied group

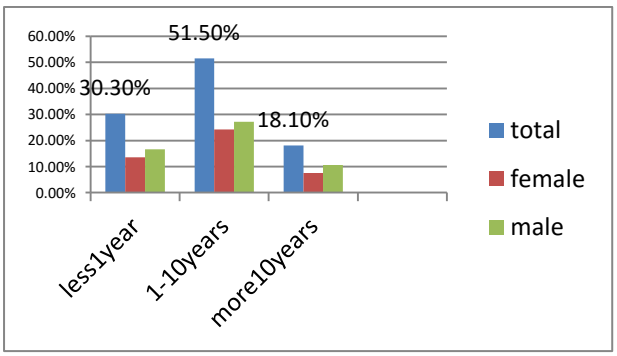


Figure2:The family history of the studied group

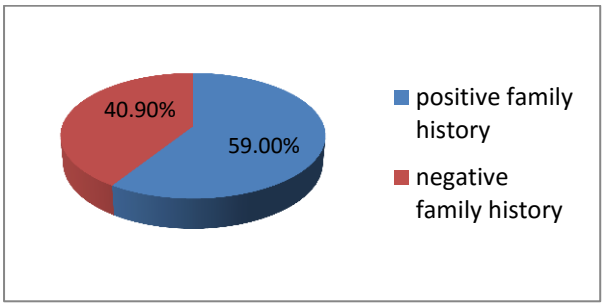


Table 1: The presenting complaints at the time of the diagnosis of the studied group

Bleeding type	Frequency(female,male)	percent
Epistaxis	23 (8,15)	34.8
Gum bleeding	22 (9,13)	33.3
Ecchymosis	14 (8,6)	21.2
Menorrhagia	3female	4.5
Tonsillar bleeding	1male	1.5
Gastrointestinal bleeding	2male	3
Bleeding from ear	1male	1.5
Total	66	100

Figure3: Hepatitis C in studied group

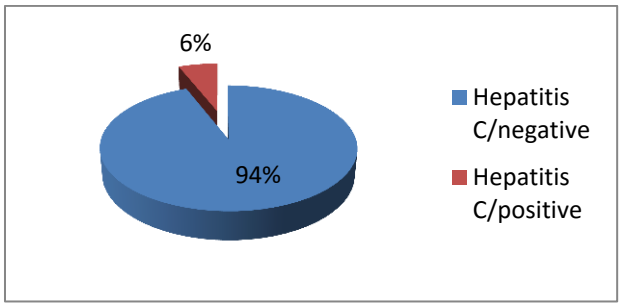
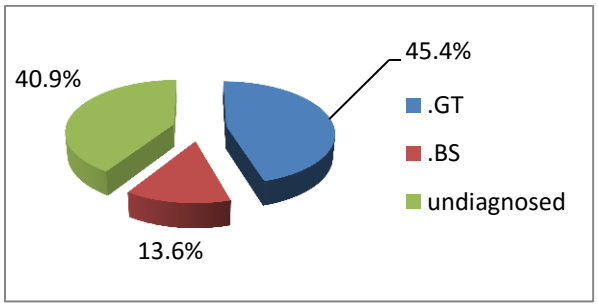


Figure4:The type of thrombasthenia diagnosed in 39 patients



Discussion: Thrombosthenia appears to be a rare bleeding disorder that to_ date only 66 patients have been identified in our haemophilia center in Children Welfare Teaching Hospital , while it is the second commonest inherited bleeding disorder among Saudis [(2)].

Our study group has shown slight male predominance (54.5%) .Like a study in Saudis were it was also a male predominance (56%).[(2)]

In our study it presents commonly at age of 1-10 years(51.5%), while it commonly presents with bleeding tendency which begins during the neonatal period and early childhood in Saudis.[(3)]

Although purpura, epistaxis, gingival bleeding and menorrhagia are nearly constant features, epistaxis is the most common cause of severe bleeding in our patients, it is typically more severe in children than in adults. It occurred in 34.8% of these patients. Gum bleeding is the next most common manifestation in this study, occurring in 33.3% of the patients, followed by ecchymotic spots in 21.2% of patients. These findings are similar to those reported in Sudia Arabia where epistaxis occurred in 88% and purpura in 75% of their patients. Gingival bleeding was observed in a higher percentage 71% in the series of Sudia Arabia. However, poor dental hygiene might have contributed to the increased prevalence of gingival bleeding in our patients. [(3)]

Menorrhagia was found to occur in only10% of our female patients while it has been reported as the most common feature in the group of George et al, occurring in 98% of their female patients[(4)]. It has also been reported by others to be one of the most common and most serious bleeding tendencies necessitating repeated blood transfusions [(5)]. The lower incidence of menorrhagia in our patients could be explained by their younger age at presentation, since all of them presented at age < 15 years and their age of menarche was not recorded. No intracranial hematomas or bleeding after circumcision were observed in this study.

The clinical and biological characteristics of 30 patients with Glanzmann's thrombasthenia and 9 patients with Bernard-Soulier syndrome who had been followed for many years but fully characterized only recently,when platelet aggregation tests became available for the first time in the country. While in Iran reliable diagnosis had been done in 2005 reveaed 23 patients with Glanzmann's thrombasthenia and of seven patients with Bernard-Soulier syndrome from southern Iran.[(6)] The study showed only one patient with resistance to platelets transfusion and use of rFVIIa instead. While in Iran 16% were treated with rFVIIa and 34% controlled by receiving both products.[(7)]

Coclusions: Congenital thrombasthenia is a rare hemorrhagic disease in our center, mostly presented as minor bleeding involving mainly mucocutaneous regions which appears at an early childhood with positive family history in most of the patients.All of the patients respond to platelet transfusion only.

References:

(1) Pittman M, Graham J. Glanzmann's thrombasthenia : An autosomal recessive trait in one family. Am J Medical Sciences. 1964;247:293.

(2) Walter A.Wuillemin, Katharina M.Gassera, Sacha S.Zeerleder.Evaluation of a Platelet Function Analyser(PFA-100®) in patients with a bleeding tendency.2002;132:443-448.

(3) Sahida K. Al-Barghouthi, Abdullah Al-Othman, and Amer Lardhi, Glanzmann's thrombasthenia_ Spectrum of clinical presentation on Saudi patients in the eastern province.1997 Jan;4(1):57-61.

(4) George JN, Caen JP, Nurden AT. Glanzmann's thrombosthenia: The spectrum of clinical disease. Blood.1990;75(7):1383-1395.

(5) Agarwal MB, Agarwal UM, Viswanathan C, etal. Glanzmann's thrombosthenia. Indian Pediatr. 1992;29(7):837-41.

(6) Afrasiabi A[1], Artoni A, Karimi M, Peyvandi F, etal. Glanzmann thrombasthenia and Bernard-Soulier syndrome in south Iran. Clin Lab Haematol. 2005 Oct;27(5):324-7.

(7) Eshghi P 1, Jenabzadeh A 1, Habibpanah B 2 .Hemorrhage reatment Report of Patients Suffering from Glanzmann's Thrombasthenia resulting Hospitalization from 2006 to 2011 at Mofid Children's Hospital. IJBC 2014;6(3): 127-131