

Demographic Data about Glanzmann's Thrombasthenia and Bernard Soulier in Wassit , Iraq

Dr Ahmed Hatem Sabhan[1]
Dr, Safaa A Faraj [2*]
Dr. Jassam Kareem Al Lami [3]
1 MD Pediatrics, Welfare Children Teaching hospital, Medical City, Baghdad, IRAQ.
2 MD Pediatrics, Wasit university, College of medicine, IRAQ.
3 MD Pediatrics, Al Karma Teaching hospital, Wasit, IRAQ
*E-mail of the corresponding author: safaafaraj@uowasit.edu.iq

Abstract

Objective: Thrombasthenia is rare inherited bleeding disorder .Clinical presentation differs and the aim of this study is to determine the demographic Data for Glanzmann's thrombasthenia ,How it differs from Bernard soulier, the clinical presentation of patients with Glanzmann’s thrombasthenia (GT) and Bernard and to correlate their clinical presentation and laboratory findings.

Methods: A retrospective analysis of 32 patient having Bleeding disorder (Thrombasthenia -un specified,Glanzmann’s and Bernard) registered in The hematology center \ Al Karama hospital \ Al Kut city, Wassit state, Iraq. The data were collected from records from June 2003 to April 2016.The Demographic data of patient's age at time of diagnosis, sex, ABO blood group ,Chief complaint ,investigation values family history and the consequent state of patients were collected to be analyzed .

Results: From total of 32 patients , 65.6% from them were males, 21 males and 11 females. In this study The patients were from three provinces (most patients were from Wassit 25, Dhi-qaar 4 and Missan 3.We found that from total of 32 patients, the number of patient above 5 years of age were 20 (62.2%), where the patients below 5 years were 12 (37.5%).A positive family history were found in 21 (65.6%), negative family history were found in 11 (34.4%).In this study, most of cases were Bernard soulier (43.8%) with 14 patients , while Glanzmann's (34.4%) in 11 patients, and 7 patients with thrombasthenia (21.9%). As for blood groups, most cases were unknown (37.5%) in 12 pts ,the second most common were A+ in 8 pts (25%) , B+ group and O+ were equal in 5 pts (15.6%) each, the least common were O- in 2 pts (6.2%).Most cases presented as bleeding in 24 pts (75%) , known case in 5 pts (15.6%) and pallor in 2 pts (6.2%), accidental during surgical operations was the least common with only 1 patient (3.1%).Most cases were positive consequent state -21 patients with 65.6% percentage, negative consequent were 9 pts (28.1%) and unknown in 2 pts (6.2%) .

Conclusion: Bleeding disorders such as Glanzmann’s and Bernard are not that rare in Iraq. There is also female:male ratio that is higher than other researches. Most of the patients were above the age of 5 ,while in other researches the diseases are most likely diagnosed in infancy.

Key word: bleeding, platelet function, thrombasthenia

1-Introduction

Glanzmann a swiss pediatrician initially described Thrombasthenia in 1918, when noted patient had purpuric bleeding with normal platelets count. Glanzmann, and Bernard – soulier syndrome, are inherited disorder of platelet function, as well as deficiencies of platelet adhesion, aggregation and secretion, all these disorders are characterized by a lifelong bleeding tendency.Glanzmann thrombasthenia **:**It is an autosomal recessive rare genetic platelet disorder characterized by qualitative or quantitative deficiencies of fibrinogen receptor IIb IIIA GP leads to defective platelet aggregation and subsequent bleeding, with mucocutaneous bleeding at birth or early in infancy is the typical presentation, and the chromosome 17 is the site of the gen of both proteins and 50% activity of each protein enough for normal platelet aggregation(1,2,3,4,5).Countries such as Jordan, Iran,

reported families with high incidence where close relative marriage is not uncommon.(6)Bernard-soulier syndrome**:** It is rare autosomal recessive bleeding disorder characterized by absence or decreased expression of GPIB/IX/V complex on the surface of platelets, this complex is the receptor of von willebrand factor, usually present with gum bleeding, epistaxis, bruising or internal bleeding which can be started since birth.(7).

2-Patients & methods

The study based on data collected from 32 patients(21 male and 11 female) ,age range from (7 months -17 years) who were diagnosed with Glanzmann's thrombasthenia and Bernard soulier syndrome they were registered in hematology center in al karama teaching hospital state of wassit,Iraq data were collected since 2010 till 2016 and were entered into statistical package for social science SPSS on April 2017 data include (age, sex, chief complaint, diagnosis, family history, sign ,HB, WBC ,platelet count ,blood group ,follow up) the following age groups were considered (patients < 5 and patients > 5) quantitative variables were summarized by finding mean and qualitative variables were summarized by frequency and percentage.

3-Results

From total of 32 patients , 65.6% from them were males and 34.4% females ,21 males and 11 females. In this study the patients were from three provinces (most patients were from Wassit 25 (78.1%) , Dhi-qaar were 4 patients (12.5%) and 3 from Missan (9.4%).

Almost equal numbers for patients in rural and urban areas.

About the age , we found that from total of 32 patients , the number of patient above 5 years of age were 20 (62.2%), while the patients below 5 years were 12 (37.5%). A positive family history was found in 21 (65.6%) , negative family history in found in 11 (34.4%). Most of cases were Bernard soulier (43.8%) with 14 patients , while Glanzmann's (34.4%) in 11 patients , and 7 patients with thrombasthenia (21.9%) .From total of 32 pts , 11 patients with Glanzmann's (34.4%), most (7 patients) were above 5 years of age ,10 were associated with positive family history , it was more common in males , in 7 males in comparison with 4 females. In Bernard soulier most were below 5 years of age (9 pts) , more negative family history in (8pts)than positive(6pts) ,as for sex of patients they were close as 8 males for 6 females , Bernard had the highest WBC count , for value of 21160 .As 7 patients of thrombasthenia (21.9%) , 5 of them were below 5 years of age , the majority are males , with more positive family history in 5 pts , the investigation indicated that the H.B was the highest value of 10.12. As for blood groups , most cases were unknown (37.5%) in 12 pts ,the second most common were A+ in 8 pts (25%) , B+ group and O+ were equal in 5 pts (15.6%) each.

the least common were O- in 2 pts (6.2%).Most cases presented as bleeding in 24 pts (75%) , known case in 5 pts (15.6%) and pallor in 2 pts (6.2%) ,accidental during surgical operations were the least common with only 1 patient (3.1%).

Most cases were positive for consanguinity marriage in 21 patients with 65.6% percentage , negative consequent were 9 pts (28.1%) and unknown in 2 pts (6.2%) . as demonstrated in table 2.

4- Discussion

This study reflects the prevalence of GT and Bernard soulier syndrome in the Southern Governorates of Iraq. It is clear that GT and BSS are not that rare in comparison to other areas of the world. In this study we analyzed 32 patient diagnosed with bleeding disorder in the Hematology center of Wassit in year of 2003 to 2016.
From total of 32 (21 males and 11 females) patients, most patients were from Wassit about 25, Dhi-qaar 4 patients and Missan 3 patients. Males are affected more than females, articles shows that males and females are almost equally affected.

Various studies done have shown that GT is a disease of children and young adults with majority of patients being less than 20 years (12, 13, 14). In our study patients were organized to: below 5 years and above 5 years in age. In GT a seven patients from11patients were less than 5 years, which is similar to that mentioned in other studies that usually diagnosed before 5years of age.(9). Most cases were positive for consanguinity marriage in

21 patients (65.6%), the incidence of both disease increase in communities were consanguineous marriage is a common behavior, which is more prevalent in some populations, such as Iranians, and Southern Indians.(9).
Consanguineous marriage between parents has been found to be very important risk factor for GT and other bleeding disorders . In the previous years incidence was higher in rural areas, however it is now almost in equal distribution, most likely the cause could be due to the acceptance and increased approval of cousin’ s marriage in urban areas too.

The clinical presentations of patients with GT was wide and mostly presented with mucocutaneous bleeding, mainly ecchymosis or petechiae, epistaxis and gum bleeding, this fact was found in other studies (9, 10).

In our study the investigations discussed : platelet count, HB and WBC, levels for both diseases are in normal range. However these investigation methods are not reliable in diagnosis specially for thrombasthenia in which the values are within normal range.
In Bernard platelet count is low(thrombocytopenia),However the mean value for platelet count is above 100,000 ,platelet count estimation is difficult, because the size of a platelet may reach that of RBCs.
No cases associated with organomegaly except for one, due to infection that misinterpreted the diagnosis and management of the patient.

5- Conclusions

Bleeding disorders such as Glanzmann’ s and Bernard are not that rare in Iraq.There is also female: male ratio that is higher than other researches.

Most of the patients were above the age of 5 ,while in other researches the diseases are diagnosed in infancy.

The most common confirmed diagnosis is Bernard soulier,It is autosomal recessive disease however most of the cases with negative family history.

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figure No1 . geographic data of the patients

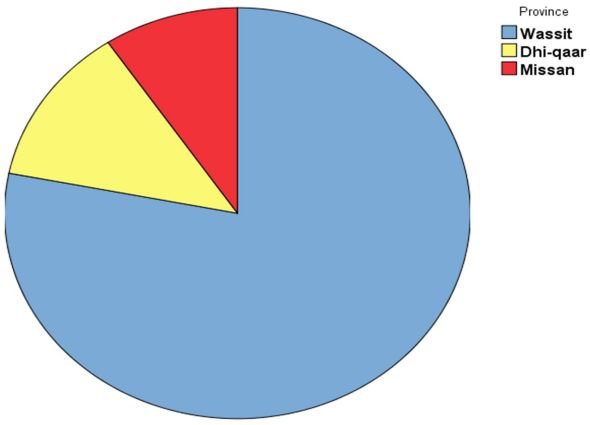

figure No2. blood group of the patients

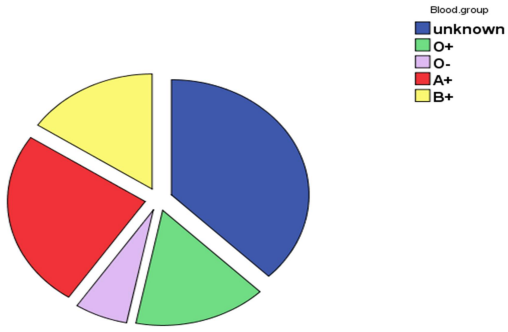

Table 1: demographic data of patients with Glanzmann's thrombasthenia and Bernard soulier diseases.

Viarabels		**Patient No.**	**Percentage %**
Age	≥ 5 years	20	62.2%
	<5 years	12	37.5%
Sex	Male	21	65.6%
	Female	11	34.4%
Address	Urban	15	46.87%
	Rural	17	53.125%
consanguinty	positive	21	65.625%
	negative	9	28.125%
	unknown	2	6.25%
Family history	negative	11	34.4%
	positive	21	65.6%
Chief complaint	known case	5	15.6%
	bleeding	24	75%
	pallor	2	6.2%
	accidental finding	1	3.1%
Diagnosis	thromasthenia	7	21.9%
	glanzmann	11	34.4%
	Bernard soulier	14	43.8%
Investigations			
1-Platelet count.	≥ 100,000	10	71.4%
	<100,000	4	28.5%
2-H.B count	maximum⇒16.70	minimum⇒2.80	mean⇒10.107
3-WBC	maximum⇒64000	minimum⇒ 4460	mean⇒13.021

Table 2: Number of Cases with thrombasthenia and Bernard soulier in association with age, sex ,family history and mean values of investigation.

Diagnosis	N. of p.	Age		Family history		Sex		Investigations mean values		
		$\geq$ 5 *years*	$\leq$ 5 *years*	positive +	Negative -	F	M	H.B	Platelet	WBC
Glanzmann' s	11	4	7	10	1	4	7	10.9	248333	8110
Bernard soulier	14	9	5	6	8	6	8	9.7	116000	21160
Thromasthenia	7	5	2	5	2	1	6	10.12	191400	8693
Total	32	18	14	21	11	11	21	----	----	----

Table 3: Blood groups ,presentation of patients with thrombasthenia and Bernard soulier disease and the consequent state of these patients.

ABO blood group			chief complaint			Consequent state		
Groups	Frequency	percentage	presentation	Frequency	percentage	State	Frequency	Percentage
A+	8	25%	Known case	5	15.6%	Positive	21	65.6%
B+	5	15.6%	Bleeding	24	75%	Negative	9	28.1%
O+	5	15.6%	Pallor	2	6.2%	Unknown	2	6.2%
O-	2	6.2%	Accidental during surgical operations	1	3.1			
Unknown	12	37.5%						

Table 4: LAB findings and age for patients with thrombasthenia and Bernard soulier in Wassit.

Values	Age	HB	WBC	Platelets
Minimum	7 months	2.8	4460	12000
Maximum	204 months	16.7	64000	398000
Mean	75 months	10.1	13020	171285