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Glanzmann Thrombasthenia Mimicking Early Neonatal Sepsis

Nihat Demir[1], Sultan Kaba[2], Mesut Garipardiç[2], Erdal Peker[1], Oğuz Tuncer[1]

[1]Yüzüncüyıl University, School of Medicine, Neonatology, Van, Turkey
[2]Yüzüncüyıl University, School of Medicine, Pediatric Hematology, Van, Turkey

Abstract
Glanzmann thrombasthenia is an autosomal and recessively inherited disorder resulting from abnormality in the first step of thrombosis. In cases associated with hemorrhagic diathesis and ecchymotic mucocutaneous lesions, Glanzmann thrombasthenia should be taken into account in differential diagnosis if thrombocyte count is normal and bleeding duration is too long. In this study, we present the case of a pair of twins in which one of the siblings was lost in the uterus while the other had sepsis along with common intravascular coagulopathy and was eventually diagnosed with Glanzmann thrombasthenia.
Key Word: Glanzmann; Sepsis; Hemorrhagic Diathesis.

Erken Neonatal Sepsisi Andıran bir Glanzman Trombasteni Olgusu

Özet
Glanzmann trombastenisi pıhtı oluşumunun ilk basamağındaki anormalikten kaynaklanan otozomal resesif geçişli kalıtsal bir hastalıktır. Ekimotik mukokütenöz leziyonlarla ve kana diateziyle ilişkili olgularda, eğer trombosit sayısı normal, kanama zamanı uzun ise Glanzamann trombastenisi ayırıcı tanıda düşünülmelidir. Burada ikiz eşi olarak doğan, diğer ikizi intauterin exitus olan ve yaygın intravasküler koagulopati ve sepsis tablosuyla gelen ardından GT tanısı konan bir olgu sunuldu.
Anahtar Kelimeler: Glanzman; Sepsis; Kanama Diyatezi.

INTRODUCTION

Glanzmann thrombasthenia (GT) is an autosomal recessive disorder characterized by the absence or functional impairment of glycoprotein (GP) IIb-IIIa receptors, which were first defined by Doctor Eduard Glanzmann (1, 2, 3). In general, clinical findings emerge in neonatal and infancy period and they may delay until adult ages (5, 6). In twin pregnancies, mortality and morbidity risks are higher compared to singleton pregnancies. Among all twin pregnancies, loss of one sibling is seen at a rate of 3.7-6.8%. This may cause serious complications for both the mother and the other living baby. The living baby may suffer from complications such as preterm birth, central nervous system defects, early onset-late onset sepsis, renal anomalies, disseminated intravascular coagulopathy (DIC), and skin defects (8, 9). In this report, we are presenting the case of a surviving twin who developed DIC while the other had intauterin exitus and how underlying inherited hemorrhagic diathesis may be concealed causing diagnostic delay.

CASE REPORT

A 39-week-old baby girl was born to a 32-year-old healthy mother as the fourth child spontaneously delivered vaginally. Immediately after the birth, the infant started crying with a 1-minute Apgar score of 8 and 5-minute Apgar score of 9. At birth, the baby weighed 3200 grams (25-50 percentile); height was 50cm (25-50 percentile) and his head circumference was 34cm (50 percentile). We observed ecchymoses all over the whole body during the delivery and neonatal reflexes were weak. As its sibling died in the uterus in the 38th week of pregnancy, early sepsis was reduced and, sending the neonate to intensive care unit, we started sultamicilin-amicasin antibiotheraphy. Similar disorder was not detected in children of families with second degree kinship. On physical examination, we observed disseminated ecchymotic lesions ranging between 1cm and 3cm on the whole body (Figure 1).

The baby also had suction weakness. Other system examinations were normal. The laboratory results were as follows: complete blood count: haemoglobin (Hb) 11,5 gr/dL; haematocrit (Hct): 35%; white blood cell count (WBC): 20.000/mm^3; thromboctye count (PLT): 147000/mm^3; prothrombin time (PT): 14,5 seconds; actived partial thromboplastin time (aPTT): 30,7 seconds; aspartate transaminase (AST): 106 U/L; alanine aminotransferase (ALT): 24 U/L; total bilirubin (TB): 2.9 mg/dL; and C-reactive protein (CRP): 3 mg/dL. On the second day of the birth, the patient did not have any ABO, Rh, or other blood incompatibility issues and the direct coombs test was negative. We observed the following values: Hb 8.5 gr/dL, Hct %26, aPTT 30 secs, PT 15.2 secs, TB 16 mg/dL, AST 490 U/L, ALT 136 U/L, and CRP 3.2 mg/dL. We did not detect reproduction in blood or urinary cultures. We applied exchange transfusion due to anaemia and high TB. After the transfusion, TB was 6.5 mg/dL while other haematological parameters were detected normal. In

the meantime, echocardiography, cranial abdominal ultrasonography, and other examinations revealed normal results. As the patient had normal bilirubin values and recovered from ecchymoses, it was discharged in good health after five days (Figure 1).

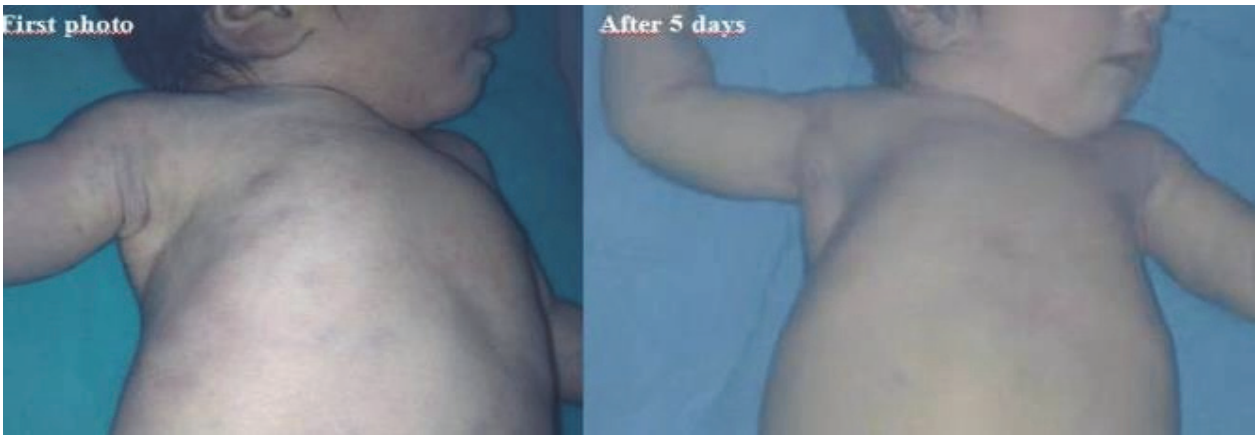

Figure 1. Photograph showing the lesions on the skin on the first (left) and fifth (right) days.

One week later, the patient was admitted with disseminated ecchymoses all over the whole body. The physical examination revealed disseminated partial and ecchymotic lesions. They were especially apparent on extensor surfaces of the gluteal area, back, and extremities. The abdomen and cranial ultrasonography results were normal but we detected disseminated micro hemorrhagic areas during fundus examination of the eyes. The haematological and biochemical examination results were as follows: Hb 11.5 gr/dL, Hct 36.7%, WBC 10.200/mm^3, PLT 354000/mm^3, PT 12 secs, and aPTT 32 secs. We observed that platelets did not constitute clumps in the peripheral smear and their sizes were morphologically normal. However, when we examined the patient with Ivy method, we found that bleeding period was 13 minutes (Normal: 1.9-5.8 mins); we also detected ristocetin and aggregation though ADP and epinephrine were not observed after the platelet aggregation analysis. Therefore, we performed a test and found out CD41 to be at 0,6% and CD61 at 0,2%. As a result, our patient was diagnosed with Glanzmann thrombasthenia and has been followed up in our clinic for the last 18 months.

DISCUSSION

Glanzmann thrombasthenia is an inherited platelet function disorder. This disorder is divided into three types according to GP levels. If GP level is below 5%, it is evaluated as type I; if it is between 5-20%, it is evaluated as type II; if GP value is normal or close to normal though with impaired function, it is evaluated as type III (4). Prolonged bleeding duration, low CD41 (GP IIb) and CD61 (GP IIIa) levels in flow cytometry, and the absence of aggregation with any substance except for ristocetin are diagnostic signs (5, 7). GT reveals itself in birth or infancy with various skin and mucosal bleeding so it should be kept in mind in diagnosis if bleeding duration is long despite normal thrombocyte count, PT, and aPTT values (3, 5, 7). In our case, petechia, purpura, and ecchymotic skin lesions detected in the examination shortly after the birth made us consider early sepsis (Figure 1).

Losing siblings in twin pregnancies causes serious complications. While intrauterine exitus of one baby does not cause any problems for the other baby in first trimester, this may cause serious complications for the mother and other baby in the second and third trimesters. The primary complications can be disseminated intravascular coagulopathy, anaemia, sepsis, nephrological problems, neurological damages, and premature birth (8, 9). Our patient was a term baby and the other sibling had died in the 3rd trimester; the findings detected on physical examination were primarily evaluated as sepsis and disseminated intravascular coagulopathy triggered by sepsis. This clinical picture recalls sepsis to mind; moreover, it shows that cases should be examined more deeply in terms of hemorrhagic diathesis. After a survey of the literature, we have found out that our case has been the earliest in receiving a diagnosis; the patient was diagnosed with GT when she was 15 days old. Therefore, we have aimed to present our case in order to emphasize the possibility that the diagnosis may be missed out by being masked due to sepsis in neonatal period of hemorrhagic diathesis. In other words, Glanzmann thrombasthenia may be mistaken for early and/or late neonatal sepsis. In cases with general ecchymotic eruption in birth, it might be observed that platelets do not constitute clump despite normal thrombocyte count, PT, and aPTT values through peripheral smear performed on finger. In such cases, GT or other platelet disorders should be considered.

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Correspondence/İletişim

Nihat DEMİR
Yüzüncü Yıl Üniversitesi Tıp Fakültesi Neontoloji Anabilim Dalı, VAN, TÜRKİYE
E-mail: demirnihat27@hotmail.com

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