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RESEARCH ARTICLE

A RARE CASE OF GLANZMANN THOMBASTHENIA

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Abstract

Background: Glanzmann's Thrombasthenia (GT) is a GPIIb/IIIa platelet surface receptor condition that is defined by a lack of platelet aggregation, either qualitatively or quantitatively. This receptor's physiological function is to bind various sticky plasma proteins that aid platelet adhesion and aggregation, ensuring the development of thrombus at sites of vascular damage. Patient of GT typically presents with menorrhagia, easy bruising, epistaxis, and gum bleeding.Prolonged untreated or unsuccessfully treated hemorrhagia associated with Glanzmann'sThrombasthenia may be life threatening.

Case Description: A 12 years old girl presented with bleeding since 12 days with complaints of generalized weakness. The patient was then investigated to find the cause of puberty menorrhagia. Several blood investigations were performed, and a final diagnosis of Glanzmann's Thrombasthenia was made.

Objective: To highlight a rare case of Glanzmann's Thrombasthenia with β-Thalassemia trait in a 12 years old girl with puberty menorrhagia.

Introduction:-

Glanzmann's Thrombasthenia (GT) is an autosomal recessive bleeding condition defined by a lack of platelet aggregation in the megakaryocyte lineage(1). It's a mild to severe hemorrhagic condition characterised by mucocutaneous haemorrhage(2). Quantitative and/or qualitative anomalies in GPIIb/IIIa (IIb3) integrin, the receptor that facilitates platelet incorporation into an aggregation or thrombus at locations of vascular damage, are connected to the molecular foundation.(1).

Inherited platelet abnormalities are uncommon and have received little attention until recently. Glanzmann's thrombasthenia is one of the most well-known and well-defined hereditary aberrant hemostasis syndromes today(3). GT affects males and females in equal numbers although some studies reported greater prevalence in females(4).

Menorrhagia, easy bruising, purpura, epistaxis, and gingival bleeding are all symptoms of GT(5). Patients with bleeding symptoms should be suspected of having a bleed, and accurate medical history, light transmission aggregometry, platelet function analysis, and flow cytometry are still valuable techniques(1).Local therapy, antifibrinolytics, and platelet transfusion are the most common treatments. Recombinant factor VII is a good substitute with high response rates(3). In individuals with severe GT who have failed to respond to standard therapy, bone marrow transplantation

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may be explored(6). In individuals with severe GT who have failed to respond to standard therapy, bone marrow transplantation may be explored.

Case Description

A 12 year old girl presented with complaints of excessive bleeding per vaginum since 12 days. She also had history of prolonged bleeding time on minor traumas. She had a history of one unit blood transfusion at the age of 8 months, when she has an episode of haematemesis. She also gave history of epistaxsis and gum bleeding. She was born out of a consanguineous marriage. She also gave history of a sibling being a diagnosed case of Glanzmann Thrombasthenia. On examination the patient had pallor. She was vitally stable. Her blood investigations revealed Heamoglobin- 4.8gm%, total leukocyte count – 3800, differential count: polymorphs - 40, lymphocytes - 54, eosinophils - 02, plateltes – 2.08lacs/mm3, red blood cell count RBC – 2.11lacs, heamatocrit- 15.9, MCV - 75.2, MCH – 22.7, MCHC – 30.2 , D-dimer – 0.60. peripheral smear showed, Predominantly Normocytic severely hypochromic RBCs with moderate anisopoikilocytosis showing few microcytes and pencil cells. Bleeding time was increased, >12minutes, activated partial thromboplastin time – 30.90s (control- 30s), prothrombin time – 12.60, INR – 1.0. After the preliminary blood investigations were done, a peadiatrician opinion regarding blood transfusion was made, she was advised blood transfusion at 10ml/kg over 4 hours. Glycoprotein 2B/3A test was sent for confirmation of diagnosis. She was transfused 320ml PRC during her hospital stay and was discharged on heamoglobin – 7.9 on request.

Discussion:-

Glanzmann thrombasthenia (GT) is a rare autosomal recessive bleeding illness characterised by a lack of platelet aggregation and affecting the megakaryocyte lineage.(7).It is found more common within populations that have a prevalence of consanguineous marriages although frequency of this is disorder is one in 1,000,000 individuals(1).Quantitative and/or qualitative anomalies of alphaIIb beta3 integrin are connected to the molecular foundation(8). This receptor is involved in the binding of sticky proteins that keep aggregating platelets together and assure thrombus development at blood artery damage sites. GT is linked with a wide range of clinical manifestations: some individuals experience just minor bruises, while others experience frequent, severe, and sometimes deadly haemorrhages(5). In GT, the source of bleeding is well defined: purpura, epistaxis, gingival haemorrhage, and menorrhagia are almost always present; gastrointestinal bleeding and hematuria are less prevalent(4). Antifibrinolytic treatment, such as tranexamic acid, aminocaproic acid, recombinant factor VII, and platelet transfusions, has been shown in studies to be effective for patients with GT(3). Allogeneic bone marrow transplantation performed in several severe casesof GT had led to the potential cure of this disease. This patient had typical sites of bleeding including: epistaxis, gingival bleeding, haematemesis and menorrhagia. She also had a typical history of parents in a consanguineous marriage. She was managed with PRBC transfusion, tranexamic acid, and iron supplements.

Since Glanzmann initially diagnosed the condition, our understanding of the pathophysiology and function of IIb3 has vastly advanced; nonetheless, GT therapy remains inadequate(6). Controlling and preventing bleeding in people with GT is critical yet difficult. The mainstay of therapy used to be local therapies, such as antifibrinolytic therapy with or without platelet transfusions(2). Recombinant factor VIIa, on the other hand, has seen a considerable surge in usage in recent years, with great response rates in treating and avoiding bleeding in GT patients(7). Gene therapy and stem cell transplantation are two treatments that have the potential to heal this condition, although they are both pricey and experimental at this time. In a few occasions, bone marrow transplants have been effective(8).

Conclusion:-

GT is a hereditary bleeding condition that affects just a few people. It is seen primarily in a small number of populations where consanguineous marriage is widespread. Patients with epistaxis and dental extractions frequently have simple bruising and bleeding. GT has a fairly excellent prognosis with adequate supportive treatment. When analysing any case of bleeding disease, GT should always be considered as a differential diagnosis.

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