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Paediatrics

GLUTEAL SWELLING AS A PRESENTING FEATURE OF CHRISTMAS DISEASE: CASE REPORT

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ABSTRACT Christmas disease or Haemophilia B is an uncommon inherited bleeding disorder. It occurs due to deficiency or reduced activity of coagulation factor IX (Christmas Factor). Haemophila B is seven times less common as compared to Haemophilia A. Classic presenting features of this disease include spontaneous haemorrhage in tissues with trivial trauma and bleeding in joint cavities. We hereby report a case of 14 months old male child, who presented with a gluteal abscess, multiple bruises over right shin and on anterior abdominal wall. Diagnosis of haemophilia B was done after the coagulation factor level assay revealed markedly reduced plasma activity of factor IX.

KEYWORDS : Gluteal swelling, Multiple haematomas, Christmas disease, Factor IX Activity

INTRODUCTION :

It was recognized in 1947, that haemophilia bleeding disorder consists of two subgroups. Haemophilia B was first reported in the medical literature in 1952 in a patient named Stephen Christmas. It is an inherited, X-linked, recessive disorder that results in deficiency of functional plasma coagulation factor IX. It is relatively common in Ashkenazi Jews. The incidence is 1 in 30,000 live births.

The hallmark of this disease is bleeding in joints and soft tissues with minor injuries. The frequency of bleeding manifestations & the age of presentation depends on the plasma activity of factor IX. Of all hemophilia cases, 80-85% are hemophilia A, 14% are hemophilia B, and the remainder are various other clotting abnormalities.

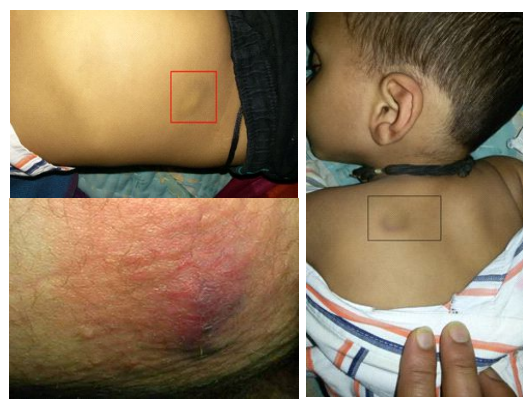
CASE REPORT :

A 14 months old male child born of non-consaguineous marriage was brought by parents with complaints of swelling over right gluteal region with history of 2 days. Parents gave history of appearance of a small lemon sized swelling with soft consistency over the right calf 4 days back, which was initially ignored. There was appearance of a similar lesion on right side of the buttock 2 days afterwards which was attributed to fall on the same side while walking.

The lesion started as a small bruise of 1x2 cm, and then grew in size to presenting level of 2x3.5cm over a day. It became firm & was associated with considerable pain & tenderness. Patient's father gave history of random appearance of 2-3 similar lesions on arms at the age of 4 months with a vague reference to bleeding continuing from the sampling site for a long time back then. Appearance of multiple similar lesions over anterior aspect of right shin, right knee joint & anterior abdominal wall in the following 4 days, without an apparent traumatic event warranted a full investigation work-up.

On examination, the baby was euthermic, euhydrated with pulse rate of 100/min & respiratory rate 26/min. Peripheral pulses were well felt, indicating normal peripheral perfusion. Systemic examination was within normal limits.

Local examination revealed a 2x3 cm ovoid swelling with soft to firm consistency, which was tender, non-reducible, nonpulsatile, associated with induration. The swelling had locally raised temperature & varying degree of erythema, pointing the diagnosis towards acute infective aetiology, such as gluteal abscess.



INVESTIGATIONS:

Ultrasonography of the swelling showed ill defined heterogenous hypoechoic lesion of dimensions 20x25x30mm in subcutaneous plane approximately 5mm deep to the skin, suggestive of an extension of ischiorectal abscess.

His haemoglobin was 6.8 g/dl with microcytic hypochromic peripheral blood picture. Platelet count was 5.2 lakh/cmm. Blood Glucose-6-phosphate dehydrogenase (G-6PD) levels were assayed to rule out haemolytic disease, occurring due to deficiency of the enzyme. The levels were normal.

Bleeding time was 1 minute 20 seconds and clotting time 4 minutes 50 seconds. Prothrombin time was 13.5 seconds, with INR-1.0 and an aPTT of 26.1 seconds. Liver and kidney function tests were normal.

We advised the parents to get Mixing study/factor assay done in order to investigate the details regarding history suggestive of bleeding diathesis. By photo optical clot detection method, Factor VIII functional activity was 105.70% (Normal range is 60 to 150%), while Factor IX activity was found to be <1% (Normal range 70 to 120%).

This investigation clinched the diagnosis as severe variety of haemophilia B. The gluteal swelling was found to be a spontaneous subcutaneous haematoma due to the disease, which later got converted into an abscess because of secondary infection. Serial X-rays of large joints were taken later to rule out haemophilic arthropathy.

TREATEMENT:

Blood reports revealed evidence of microcytic hypochromic anaemia, for which an oral iron supplement was prescribed. The patient was referred to local haemophilia treatment center for education and genetic counseling of parents. Treatment was given as intravenous infusion of recombinant factor IX for preventing further bleeding episodes. Dose of infusion was determined as per weight of the patient and percentage increment needed in factor IX activity, to prevent bleeding. In this case, the desirable level was 50% factor IX activity to prevent major joint and muscle bleeds.

DISCUSSION :

The diagnosis of severity of hemophilia B is made by measuring the factor IX clotting activity. Severe hemophilia B has <1% factor IX, moderate hemophilia B cases show 1%-5% factor IX, while mild cases typically have 6%-40% factor IX.

Patients with mild hemophilia B never present with spontaneous bleeding episodes. Bleeding may start sometimes in an otherwise uneventful procedure like dental extraction. Diagnosis usually occurs in later decades of life.

Moderate variety of Christmas disease present earlier with bleeding manifestations, occurring with trivial traumas around 5 to 7 years of age.

Severe variety has the least plasma factor IX activity and it may present within 2 years of life with extremely frequent bleeding episodes. Manifestations may range from minor mucosal bleeds to huge subcutaneous/muscular haematomas. Intracranial bleed may occur following head injury. Neglected non-accidental traumatic events escape the attention until the parents start noticing random ecchymotic patches and bruises over various parts of the body.

As with other forms of haemophilia, the joints are the most common sites of spontaneous bleeding. With increasing frequency, this may lead to permanent joint erosion, loss of movement or deformity of joints. Molecular gene testing is the definite method to diagnose the condition. This is also important from the transmission point-of-view, as all males invariably transmit the disease, while carrier females have 50% chance to transmit mutated variant gene of factor IX.

PROPHYLAXIS :

National Haemophilia Foundation advises prophylaxis for all patients with severe haemophilia. The adequacy of prophylaxis is gauged by targeting certain factor IX levels for preventing spontaneous bleeds. For mild episodes, it is recommended that factor IX activity be maintained at 30%, for moderate like muscle bleeds- 50%, and for major life threatening bleeds, at 90%. For those with severe disease, prophylactic infusions of factor IX concentrate twice weekly to maintain factor IX clotting activity higher than 1% .

Sometimes, antibodies are produced in response to the exogenous factor IX preparations, which render the treatment less effective. Anti-factor IX antibody assay may help in such cases, and the dose needs to be raised.

Local measures to stop bleeding include pressure application, use of vasoconstrictors and anti-finbrinolytics. Special precaution to be taken in paediatric population involves avoiding intramuscular injections for immunization. Male children with a possible family history of haemophilia must not undergo circumcision, unless the disease has been excluded through other lab investigations.

CONCLUSIONS:

Haemophilia B is a rare X-chromosome linked bleeding disorder. Current products to replace factor IX are safe and effective.

Comprehensive history taking is of utmost importance. Any minor bleeding issues that would otherwise be attributed to non-accidental traumas must not ignored.

Timely infusion of recombinant factor IX prevents following episodes. Genetic counseling may be needed for prevention of further cases. Prenatal testing for pregnancies at increased risk is possible if the pathogenic variant has been identified in a family member.

Recombinant factors have an advantage over plasma-derived factors in preventing transmission of transfusion associated diseases.

Mis-sense mutation based factor IX gene therapy, using an adeno-associated viral vector is currently in clinical trials.

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