

Case Report

Haemophilic Arthropathy Presenting as JIA: A Case Report

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Introduction:

Haemophilia is a coagulation disorder caused by deficiency of factor VIII (FVIII) and factor IX (FIX), prevalence of which is about 1 in 10,000 male births¹. It is an "X" linked hereditary disorder. This results in males being affected by the disease while females are carriers. Haemophilia has been documented in female in whom X chromosome inactivation occurs at an early stage of embryogenesis, resulting in unusually low levels of FVIII². Spontaneous mutations account for about 5% of all haemophilia³. Haemophilic arthropathy is usually presented as monoarticular joint involvement and rarely presented as a polyarticular arthropathy. We are reporting a case of haemophilic arthropathy with involvement of multiple joints initially diagnosed and treated as Rheumatic fever and JIA.

Case Report:

Master ES was a 7 years old boy, 4th issue of non-consanguineous parents hailing from Madaripur, was admitted in the paediatrics ward of Bangabandhu Sheikh Mujib Medical University (BSMMU) on 13 April 2010, with the complaints of pain and swelling of both knee joints and right wrist joints for 5 years. Swelling first involved the left knee joint, subsequently right knee joint and right wrist joint were also involved. Joint pain and swelling was non-migratory in nature and not associated with morning stiffness. There was no history of fever associated with joint pain. For these problems, Master ES was diagnosed as a case of Rheumatic fever and Juvenile idiopathic arthritis by different physicians and was treated with NSAIDs including aspirin and naproxen on several occasions and oral methotrexate for few months. As there was no improvement of joint problems he was referred to BSMMU.

On further inquiry after admission he had H/O prolonged bleeding after cut injury. There was no H/O

significant trauma or surgery, contact with TB patient and muco-cutaneous bleeding from any site. Immunization was completed according to EPI schedule. The boy was not circumcised. His elder brother was suffering from similar type of illness for last few years. There was no history of similar problems among maternal uncles or cousins.

On examination, Master ES was mildly pale and all vital functions were within normal limit. Regarding locomotor system examination, both knee joints and right wrist joint were swollen and tender. Overlying skin was normal and periarticular muscle wasting was present. Flexion deformities of both knee joints were present. Movements of the affected joints were restricted. Skin survey showed few blue ecchymotic areas over thigh and abdomen (Fig: 1). Other systemic examinations were normal.



Fig-1: Shows swelling of both knee joints and right wrist joint of patient and swelling of left knee joints of his brother

Investigations were done which showed mild anaemia (Hb%-10gm/dl), normal total count and distribution with normal ESR (15mm in 1st hour). His bleeding time was 4.8 min (normal), clotting time was 5.6sec (normal), prothrombin time was 11.6 sec (control-11.8 sec) and activated partial thromboplastin time was 56.9 sec (control-28sec). Factor VIII assay was done which was 4 U/dL (4%). X-ray knee joints showed osteopenia, osseous erosions, joint space narrowing and bony ankylosis (Fig: 2). We diagnosed the case as moderate haemophilia A and consulted with

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paediatric haemato-oncologist. According to their advice, Master ES was treated with fresh frozen plasma and discharged after counseling.



Fig-2: X-ray Knee joint shows osseous erosions, joint space narrowing and bony ankylosis

Discussion:

The clinical presentation of haemophilia depends on the circulating levels of FVIII and is categorized as mild, moderate or severe. Severity of haemophilia is related to the blood level of FVIII⁴. Those with less than 1% of normal activity develop severe disease, levels between 1- 5% of normal are associated with moderate disease and patient with 6-30% of activity develop mild disease⁵. Patients with haemophilia A often give a history of skin bruising, joint swelling and unusual bleeding associated with minor trauma or surgical procedures⁶. In severe haemophilia individual bleeds spontaneously without any provoking trauma. A patient with moderate haemophilia may bleed after trauma and mild haemophilia usually bleeds after surgery⁷. One of the most severe complications of haemophilia is the development of a characteristic chronic arthropathy that results from repeated bleeding into joints⁸. In haemophilia, arthropathy is secondary to recurrent haemarthrosis and chronic synovitis⁹. The chronic arthropathy affects only a few joints, most notably the knees and elbows. Involvement of other joints like shoulder, wrist, hip and ankle is less common¹⁰.

JIA is the most common disease of chronic arthritis in children with involvement of multiple joints. Haemophilia usually presents as monoarticular joint involvement. Our patient Master ES had involvement of multiple joints and may be because of that, ES was diagnosed and treated as a case of Rheumatic fever and JIA by different physicians.

Haemarthrosis occurs in 70%-80% of haemophilia and incidence of bleeding into different joints are: Knee 45 %, Elbow 30%, Ankle 15%, Shoulder 3%, Wrist 3%, Hip 2% and Other 2%¹¹. The knee joints are most commonly affected and this was observed in our case as well. Mahajan et al in their case report showed osseous erosions, joint space narrowing and bony ankylosis of X-ray knee joint as in our case¹². Bellegard and Caetano had done x-ray in 4 patients with haemophilic arthropathy where osteopenia and reduction of joint spaces was present in all the cases¹³. Up to 25% of patients may have a normal family history or lack of males in the family tree⁶. In our case, elder brother had similar illness but no other member in family was affected. Our patient had H/O prolonged bleeding after cut injury and bruises were also present over the skin. But it was not disclosed previously. So for the diagnosis of haemophilia, a careful history and physical examination in details is very important.

In Haemophilia, haemarthrosis become a major problem throughout the patient's life and need immediate treatment. Bleeding into the joint irritate the synovial lining causing increased proliferation and vascularization. After repeated bleeding episodes, the synovium hypertrophies causing swollen joint and with further bleeding there is muscle weakness and loss of joint instability. Over a time, there is erosion of joint cartilage resulting in arthritis and the crippling deformity of haemophilic arthropathy⁷. In our patient peri-articular muscle wasting was present and there was flexion contracture deformity of both knee joints as proper diagnosis was very late. Correlating a good history, physical examination with characteristic imaging findings can result in prompt diagnosis and treatment.

Conclusion:

Haemophilic arthropathy is a joint-destroying disorder if not treated promptly. The aim of this paper was to highlight the importance of adequate history, thorough physical examination and proper investigations for early diagnosis and appropriate treatment of haemophilic arthropathy.

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