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A study on orthopaedic status of the patient with hemophilia in the Bundelkhand region

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Abstract
A major complication in patients with haemophilia is joint damage associated with recurrent intra-articular bleeds. Frequent haemarthrosis affecting target joints can culminate in irreversible, disabling arthropathy, impeding quality of life (QoL)[4]. Despite the availability of on-demand treatment with Factor (F) VIII or IX to manage acute bleeding episodes, haemarthrosis may occur when treatment for bleeding is not effected rapidly, or may proceed without detection. In both instances unchecked bleeding initiates a sequence of events leading to destruction of joint cartilage. The examination include following joints: hip, knee, ankle, shoulder, elbow and wrist. The gross configuration was studied and any deformity (valgus, varus, subluxation, rotation defect), increase in breadth of epiphyses, capsular thickening or muscular atrophy was noted. The range of passive motion of the joints was measured. Loss of range of motion was estimated, when possible, by comparison with the mobility of the contralateral joint. Minor surgery was more common in younger patients. The majority of surgeries in patients ≤5 years and 6–10 years were minor (72.7% [n=8/11] and 64.7% [n=22/34], respectively) and the majority of surgeries in patients aged 11–20 years and 21–30 years were Group 1 major surgery (62.7% [n=106/169] and 55.2% [101/183]). However, Group 1 major surgery was still recommended in 27.3% of patients aged ≤5 years and in 35.3% of patients aged 6–10 years. Also, 10.1% of patients aged 11– 20 years and 15.8% of patients aged 21–30 years were undergoing Group 2 major surgery.

Keywords: Orthopaedic status, hemophilia, Bundelkhand region

Introduction
Haemophilia is an inherited bleeding disorder caused by a deficiency of circulating coagulation factors, leading to a disrupted clotting mechanism. There are two common forms of hemophilia, hemophilia A and hemophilia B, which are the consequence of having insufficient amounts of coagulation factor VIII (FVIII) and factor IX (FIX), respectively. Both hemophilia A and B are X-linked recessive disorders. As a result, the disorders manifest almost entirely in males while females are the carriers of the mutated gene. Female carriers have a 50% chance of giving birth to a male affected with hemophilia. However, approximately a third of patients with hemophilia A have the disease sporadically (they have de novo mutations), without the presence of family history.

The incidence of hemophilia A is approximately 1 in 5000 live male births; hemophilia A is about four to six times more prevalent than hemophilia B [1].

The World Federation of Hemophilia (WFH) estimates that there are 400,000 individuals worldwide with hemophilia [2].

The Canadian Hemophilia Registry Report in January 2007 suggests that there are 2207 patients with hemophilia A and 506 patients with hemophilia B in Canada [3].

A major complication in patients with haemophilia is joint damage associated with recurrent intra-articular bleeds. Frequent haemarthrosis affecting target joints can culminate in irreversible, disabling arthropathy, impeding quality of life (QoL) [4]. Despite the availability of on-demand treatment with Factor (F) VIII or IX to manage acute bleeding episodes, haemarthrosis may occur when treatment for bleeding is not effected rapidly, or may proceed without detection. In both instances unchecked bleeding initiates a sequence of events leading to destruction of joint cartilage.

Intra-articular hemorrhage is clinical hallmark of haemophilia. A single hemarthrosis may precipitate low grade synovitis which predispose the involved joint to recurrent hemarthrosis and a cycle of chronic synovitis, inflammatory arthritis and progressive arthropathy. The weight bearing joints are the most common sites of haemophilic arthropathy with frequency being in decreasing order Knee, Elbow, Shoulder, Ankle, Wrist, Hip.

Synovium lining of joint has limited capacity for absorbing of blood, recurrent bleeding into joints results in a level of blood breakdown products that synovial membrane cant remove. Iron, constituent of erythrocytes is play major role in inflaming synovium. Presence of hemosiderin is thought to promote the production of pro inflammatory cytokines such as IL-I,6 and Tumour necrosis factor-alfa and induction of gene which causes cellular proliferation such as mdm-2. The marked inflammation and synovial hypertrophy in haemophilic arthropathy bear resemblance to the pathological mechanism seen in RA, while progressive degeneration of hyaline cartilage mimic osteoarthritis. So a vicious cycle of hemarthrosis-synovitis-hemarthrosis developed which leads to progressive degeneration of joints. Emphasis should be given to break this cycle before development of any articular destruction [5, 6].

Methodology

The patients with hemophilic disorders attending the orthopaedic and pediatric department were included in the study (Prospective and retrospective study). A distinction is made between haemophilia A and haemophilia B, and within each of these types the degree of the disease is classified according to the plasma content of AHF (haemophilia A) or B-factor (haemophilia B) as severe**,** moderate or mild. All patients were being treated with on-demand FVIII or FIX for the management of acute bleeds. We also studied an evaluation of patient orthopaedic status and the requirement for orthopaedic intervention. Three definitions of surgical intervention were used: minor surgery (synoviorthesis); Group 1 major surgery (synovectomy, osteotomy or arthrodesis); and Group 2 major surgery (joint replacement with a prosthesis).

Clinical Examination

The examination include following joints: hip, knee, ankle, shoulder, elbow and wrist. The gross configuration was studied and any deformity (valgus, varus, subluxation, rotation defect), increase in breadth of epiphyses, capsular thickening or muscular atrophy was noted. The range of passive motion of the joints was measured. Loss of range of motion was estimated, when possible, by comparison with the mobility of the contralateral joint.

Hip. Flexion was measured with the knee bent. Abduction and adduction were measured with the hip joint extended. Normal Outward and inward rotation were measured with the hip joint flexed 90'.

Knee. Extension-flexion was measured.

Ankle (including the talo-crural and subtalar joints). Dorsal and plantar flexion as well **as** pronation and supination were noted. Normal ranges: dorsal flexion about 20°, plantar flexion about 40°, pronation-supination about 45'.

Shoulder. The purpose of the examination was to assess the mobility of the humeroscapular joint. Abduction with the scapula fixed normally about 90'. Rotation was measured with the arm abducted 90'.

Elbow (including the distal radio-ulnar joint with which it forms **a** functional unit). Pronation and supination was measured with the elbow flexed 90'.

Wrist. Volar and dorsal flexion as well as radial and ulnar deviation were studied.

Normal ranges: volar 80'-90', dorsal 50'-60°, radial about 20°, ulnar about 45'. Roentgen Examination As a rule, the patients were also examined roentgenologically

Results

Individual joints

HIP

Hip arthropathy occurred in 1 out of every 9 patients with severe haemophilia, and hardly ever in patients with moderate or mild haemophilia. In x-rays breakdown of acetabular roof may be seen which invite subluxation. Collapse of head, femoral head surface irregularity and erosin and other changes mimicking Perth's disease is usual presentation in hip arthropathy. Dislocation, Osteoarthrosis, Arthrokatalysis, Fibrous ankylosis and AVN of hip is some other presentation.

Knee

Patients had knee joint arthropathy, which was bilateral in many cases. This condition was found in 4 cases out of every 5 patients with severe haemophilia, in every other one with moderate haemophilia, and only occasionally in patients with mild haemophilia. In severe and moderate haemophilia the arthropathy, when present, was predominantly of grade 3 or 4. The joint changes were bilateral in three fourths of the patients with severe haemophilia and in half of those with moderate haemophilia.

There is general agreement that haemophilic arthropathy involves the knee more often than any other joint.

Changes in the epiphyses occur early: the trabeculation is increased and the epiphyses are enlarged. Cysts of varying size appear periarticularly. Disturbed development of the epiphyses can lead to deformation of the femoral and tibia1 condyles. The patella increases in size, especially in thickness, and its lower pole becomes squared off. Erosion of the joint cartilage often occurs early. In later stages the cartilage undergoes destruction with narrowing of the joint space, posterior subluxation of Tibia and the formation of osteophytes

Case-22yrs/M The right knee was increased in breadth and deformed. No exudates was demonstrable in the joint. The knee was flexed ' and there was varus deformity. Range of motion 20°. Roentgen examination revealed broadening of the femoral condyles, deformation of the patella and decalcification with decrease joint space. The left knee and the right elbow showed mild changes of the type seen in haemophilia arthropathy. Other joints examined were of normal appearance. Cartilage undergoes destruction with narrowing of the joint space and the formation of osteophytes

Case -10yr/M. Moderate exudates in the right knee joint. Extension defect of 60'. Range of motion 10' (120-110'). Roentgen examination revealed broadening of the femoral condyles, coarse trabeculation of the bones and erosion of the articular surfaces. The mobility of the elbow joint was somewhat reduced. Other joints examined appeared normal.

Case -17yr/M The left knee was increased in breadth and deformed. No exudates was demonstrable in the joint. The knee was flexed ' and there was a subluxation backward of the tibia. Range of motion 90'-60°. Roentgen examination revealed broadening of the femoral condyles, deformation of the patella and decalcification with decrease joint space. The right knee and the left elbow showed mild changes of the type2 seen in haemophilic arthropathy. Other joints examined were of normal appearance.

Elbow.
Patients with severe haemophilia were affected more, with moderate haemophilia, half were affected. None of the patients with mild haemophilia were affected. Grade 4 arthropathy was never observed before age of 40 in patients with moderate haemophilia. In patients with grade 3 or 4 extension-flexion as well as pronation supination was as a rule impaired.

Case-patient age-30yrs/m
O/E-supination and pronation restricted, with 15degree of fixed flexion deformity over Rt side, and 10 degree over Lt side.
X-rays –haemophilic arthropathy with decreased joint space, increased size of head of radius, deepening of the ulnar and radial incisures.

Grade-3 haemophilic arthropathy of elbow
CASE-22yrs/M with both elbow and knee involve, with flexion rt elbow (0-120) extion (0-30), Lf elbow-flexion (0-55) extension (0-35) also having one episode of Illio Psoas bleeding with sensory loss over lateral aspect of Lf thigh.

Ankle
2 case showing ankle affection, 6 patients have bleeding episode but no x-rays finding flattening of the superior articular surface of the talus was a common in x-rays, collapse of body of talus may also be a finding.

X ray-lf ankle AP view and Lateral view
Case-30yrs/m with sevre haemophilia having multiple bleeding episode over Rt ankle with involvement of Rt knee as well as Lf knee. X-ray finding shows flattening of talus with decreased joint space and sclerosis.

X-RAY Rt ANKLE-Grade 3 arthropathy of ankle
Bony Ankylosis
Bony ankylosis is rare in haemophilic arthropathy. The deficiency of fibrinolytic activity in the joints in haemophiliacs may thus help to explain why bony ankylosis is rare in such patients. In the present investigation 2 patients who had severe haemophilia showing ankylosis features believed that the joint had stiffened due to lack of treatment.
CASE-37yrs/m, with stiff knee joint Lf side, flexion of (10-20) on Rt side knee with severe haemophilia. X-ray finding shows ankylosed Lt knee joint

Wrist
The condition was equally common in moderate and severe haemophilia, and was seen in about one tenth of these patients. It was not seen in patients with mild haemophilia.
Case-32yrs/M with reapeated bleeding over Rt wrist, with restriction of movement of wrist. X-ray finding-multiple sub chondral cyst over distal end of radius and ulna with reduced joint space.

Fracture by Trival trauma
CASE-18yrs/M, having knee arthropathy, slipped by walking with support according to the patient. x-ray finding-Rt knee with leg show fracture rt mid shaft tibia. Patient was given spillage in the form of B/K POP SLAB and advised for surgery and referred to higher centre.

Malunited fracture RT mid shaft femure
CASE-20yrs/M with spontaneous bleeding over Rt knee and Rt shoulder, with chronic arthropathy of Rt knee, 3yrs back slipped while walking. Xray finding shows malunited fracture mid shaft rt femur.
The majority of patients (61%; n=52/85) required surgical intervention for joint disease. Most patients (n=38) had severe haemophilia (<1% factor activity), while 13 had moderate (1–5% factor activity) and one mild haemophilia (5– 40% factor activity). The age at which surgery was considered was relatively young: approximately 3% were aged ≤5 years; 9% were aged 6–10 years; 32% were aged between 11–20 years and 35% were aged 21–30 years. Less than one-quarter (21%) were aged ≥31 years. The number of surgical procedures per patient age group was also high in the younger cohorts.
Fourteen per cent of all required procedures were Group 2 major surgeries, 56% were Group 1 major surgeries (synovectomy, osteotomy or arthrodesis) and 30% minor surgeries (synoviorthesis). Assessment of the percentage of surgical procedures according to type of surgery and patient age highlighted that minor surgery was the predominantly recommended option in younger patients, while the need for Group 1 and Group 2 major surgery increased proportionately with increasing patient age (Figure 1).

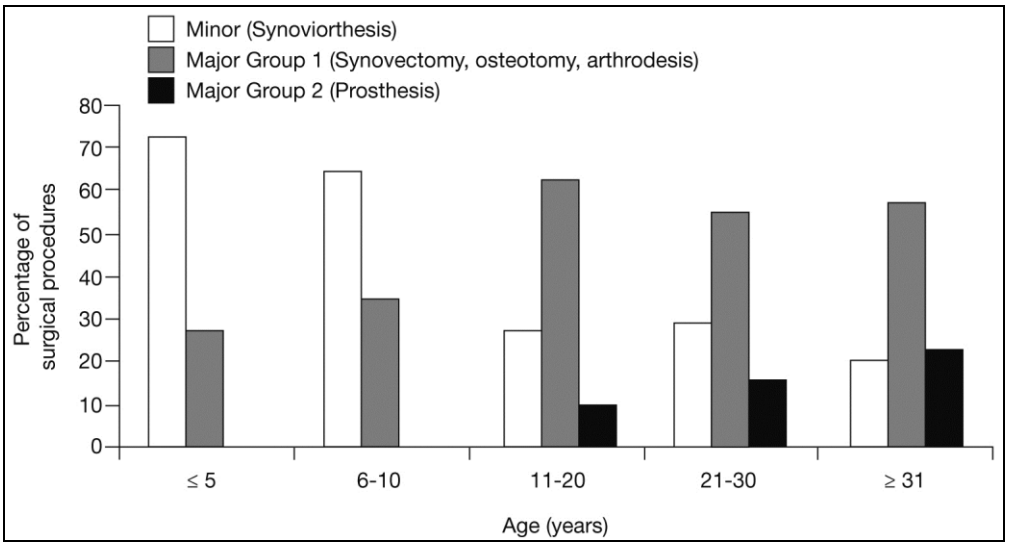

Fig 1: An assessment of the percentage of surgical procedures according to type and patient age.

Discussion

It is well established that early intervention in haemophilia is important in order to manage and prevent lasting joint damage, and priority should be given to minor surgery in patients under the age of 11 years, followed by Group 1 major surgery in patients aged 11–20 years. All case data recommended that Group 2 major surgery should be discussed in detail with an orthopaedic surgeon, taking into account of cast. In accordance with such recommendations, minor surgery was more common in younger patients. The majority of surgeries in patients ≤5 years and 6–10 years were minor (72.7% [n=8/11] and 64.7% [n=22/34], respectively) and the majority of surgeries in patients aged 11–20 years and 21–30 years were Group 1 major surgery (62.7% [n=106/169] and 55.2% [101/183]). However, Group 1 major surgery was still recommended in 27.3% of patients aged ≤5 years and in 35.3% of patients aged 6–10 years. Also, 10.1% of patients aged 11– 20 years and 15.8% of patients aged 21–30 years were undergoing Group 2 major surgery. There are some limitations associated with this analysis, including the lack of data on patient outcomes following the recommendation for surgical procedures (e.g. validation or success rates); There may also have been low implementation of primary prophylaxis in severe patients or insufficient use of rapid, on-demand FVIII and FIX, which has led to a situation where many patients with haemophilia have experienced haemarthrosis, leading to joint damage requiring surgery. Prophylaxis has been used to treat severe haemophilia for many years, and is recommended by organizations such as the World Federation of Haemophilia. Coppola *et al*. (2008) have recently discussed the benefits of prophylaxis in children, highlighting that primary prophylaxis should be started before the age of 2 years and after no more than one joint bleed; these recommendations are based on studies which showed that prophylaxis started in patients ≤2 years old was associated with fewer joint bleeds compared with patients who started prophylaxis later.

Comparisons of prophylaxis with on-demand treatment in Norway and Sweden showed that prophylaxis was associated with fewer surgeries; however, differences were found in factor-concentrate consumption, duration of use and speed with which on-demand treatment was given [7, 8] The importance of preventing long-term joint damage in order to ensure functional mobility and QoL in patients with haemophilia is essential. Ideally, joint health should be preserved and bleeds prevented so that surgery is not needed. There has been improved use of on-demand therapy in recent years. Surgery (as a method of slowing disease progression and improving joint health), may reduce the need for more major and costly surgery in later life. However, as 14% of all surgical procedures recommended or indicated in this study involved a prosthetic replacement in patients from as young as 11 years of age (due to advanced orthopaedic complications), there is a clear need for better and more comprehensive use of prophylaxis. This would help to reduce the levels of joint disease requiring major surgery in the haemophilia population, improve the orthopaedic outlook for patients and assist improved social integration. Furthermore, there is also a need for more physiotherapy and training, especially in children, as this can considerably reduce the need for orthopaedic surgery [9, 10]

Conclusion

The few cases treated with traction and extension brace do not of course warrant any definite conclusions concerning the value of different forms of substitution therapy during non-operative orthopaedic treatment. However, effective traction is possible without complicating haemorrhage into the joint undergoing treatment. The better the coagulation defect is controlled the smaller the risk of complications. It would appear that plasma transfusions are sufficient to suppress or prevent hemorrhage during traction treatment. The orthopaedic treatment regularly produced considerable improvement of walking ability. Treatment is facilitated if it is done at an early stage before contractures and muscular atrophy have become severe.

Previous experience has shown that substitution with blood and plasma is not sufficient for major operations on patients with severe haemophilia. Concentrated animal preparations produce good haemostasis, but they have antigenic properties and should therefore be used only in life-threatening situations. The only suitable substitution therapy is that using concentrated human preparations. Experience with such preparations in Sweden have shown that this preparation has good haemostatic properties and has no side effects. The preparation must be given for a long time after the operation, probably longer after operations on the skeleton than on soft tissue.

The operations should only be done in intimate cooperation with a coagulation laboratory, where the AHF respectively B-factor levels can be measured at short intervals. In view of the risk of complications such operations should only be done on strictly orthopaedic grounds.

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