Factor VII deficiency with Knee arthropathy. A case report.

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Abstract

Factor VII deficiency (or hypoproconvertinemia) is a rare autosomal-recessive bleeding disorder. Its clinical manifestations are heterogeneous, ranging from miscellaneous minor bleeding to severe life-threatening haemorrhages and joint haemorrhages, Here we present a case of congenital factor VII deficiency with chronic Knee arthropathy.

Case report

A 15-year-old girl with factor VII deficiency had recurrent hemarthroses involving the right knee, leading to chronic pain and leisure of functional score. Clinical examination found atrophy of the quadriceps, a swollen knee with moderate stiffness of this joint.

Radiographs plains showed bone cysts, joint space irregularity and global narrowing. Ultrasonography of the knee found synovial hypertrophy associated to hypoechogenic effusion in the quadriceps recesses.

The diagnosis of chronic arthropathy secondary to factor VII deficiency was retained. Our patient has had an intraarticular injection of Triamcinolone hexacetonide following fresh frozen plasma transfusion. Decreases of swelling and of joint stiffness were recorded after rehabilitation.

Discussion

The factor VII (FVII) deficiency is a rare inherited disorder of coagulation, accounting for one symptomatic individual per 500,000 population. Hemarthrosis is less common than hemophilia, although the characteristics of joint destruction are similar in the two conditions. For the case reported here, repetitive hemarthrois have lead to chronic arthropathy with a negative impact on functional score. Bleeding into joints require prophylactic replacement

therapy. Intraarticular injection of corticosteroids may be useful as a palliative measure for pain and inflammation particularly in countries with limited resources.

Keywords

Factor VII (FVII) deficiency, hemarthrosis, chronic arthropathy, fresh frozen plasma, Intraarticular injection, corticosteroids.

Introduction

Factor VII deficiency (or hypoproconvertinemia) is a rare autosomal-recessive bleeding disorder. More than 100 mutations have been identified in the Factor VII gene located on chromosome 13 [1]. Its clinical manifestations are heterogeneous, ranging from miscellaneous minor bleeding to severe life-threatening haemorrhages and joint haemorrhages [2]. Here we present a case of congenital factor VII deficiency with chronic Knee arthropathy.

Case report

Mrs G.H, 15 old years, suffered from FVII deficiency which was diagnosed during childhood. Her parents were consanguine and she had two brothers followed for the same bleeding disorder. The patient has not any health insurance coverage. This girl experienced recurrent spontaneous hemarthrosis in her right knee starting at years of age (figure1). Hemarthroses lead to chronic pain and use of analgesics. Clinical examination found atrophy of quadriceps with 7 centimeters decrease of the muscle perimeter compared to the left thigh. The affected knee was swollen with moderate stiffness of this joint. The prothrombin time was prolonged and the prothrombin and proconvertin test was 10% of normal. The activated partial thromboplastin time and the Stypven-cephalin clotting time were normal. The factor VII level was 1.4% of normal.

Radiographs plains showed osteoporosis, widening of the epichondral notch of the knee, epiphyseal overgrowth, bone cysts, joint space irregularity and global narrowing (figure 2). Ultrasonography of the knee found synovial hypertrophy associated to hypoechogenic effusion in the quadriceps recesses (figure 3).

The diagnosis of chronic arthropathy secondary to factor VII deficiency was retained. Our patient has had an intraarticular injection of Triamcinolone hexacetonide following fresh frozen plasma transfusion. Decreases of swelling and of joint stiffness were recorded after

rehabilitation. Furthermore, our patient expressed improvement of the Lesquenne and the scores.

Discussion

Congenital factor VII deficiency is a rare autosomal-recessive bleeding disorder, accounting for one symptomatic individual per 500,000 population [1,2]. Frequency is higher in countries where consanguineous marriage is more common as the case of Morocco. This inherited disorder of coagulation is the only congenital bleeding disorder characterized by isolated prolonged prothrombin time as observed in our case [3]. Clinical heterogeneity is the hallmark of this hemorrhagic disorder [2,3]. Bleeding manifestations and clinical findings vary widely, ranging from asymptomatic subjects to patients with hemorrhages that may cause significant handicaps [2,4]. It's to note that because of poor correlation between FVII levels and the bleeding tendency, FVIIC levels cannot be used to distinguish classes of severity [5]. Thus, severity of FVII deficiency is classified on clinical basis. Since chronic arthropathy has occurred in our patient, the FVII deficiency is qualified to be severe. As in hemophilia, recurrent hemarthrosis lead to chronic arthropathy source of functional disability [6]. Although hemarthrosis is less common than hemophilia, the characteristics of joint destruction are similar in the two conditions [4,6]. Management of factor VII deficiency bleeding disorder, in terms of substitution therapy and therapy schedules, is not yet optimal [7]. Treatments used for the treatment of FVII deficiency involve FVII replacement therapy using fresh frozen plasma, prothrombin complex concentrates (PCCs), plasma FVII concentrates and more recently Intravenous administration of recombinant FVIIa. Management of arthropathy could be assimilated to hemophilia case. Bleeding into joints require prophylactic replacement therapy. Beside surgical options, two basic types of synoviorthesis could be used in chronic arthropathy: chemical and radioactive. The materials most commonly used for chemical synovectomy are osmic acid, rifampicin, and

oxytetracycline clorhydrate [6,7]. Yet, intra-articular injections of steroids have been used to decrease pain and inflammation in patients with chronic synovitis, and may be useful as a palliative measure [8]. Evolution in our case is quiet satisfactory, at least regarding pain and functional status.

Conclusion

Our case illustrates an exceptional etiology of chronic arthropathy. Less known than hemophilia, factor VII deficiency should be suspected in front of prolonged prothrombin time with normal partial thromboplastin time. Bleeding into joints require prophylactic replacement therapy. Intraarticular injection of corticosteroids may be useful as a palliative measure for pain and inflammation particularly in countries with limited resources.

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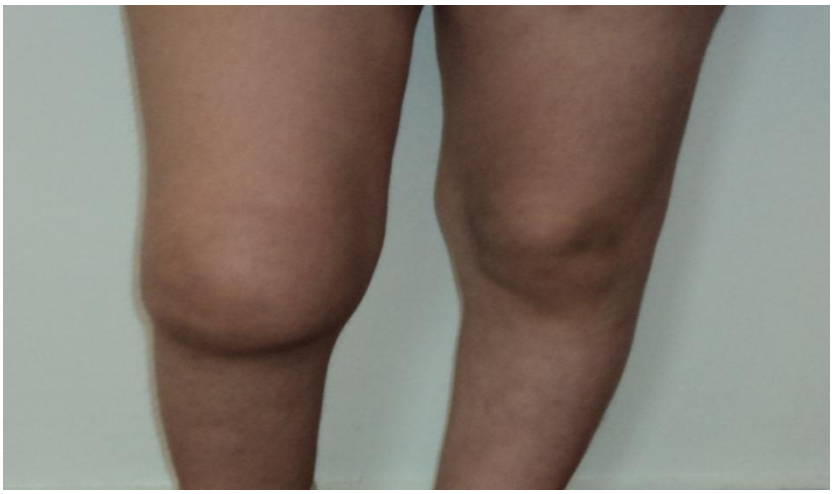

Figure 1: Swollen knee with a moderate flessum

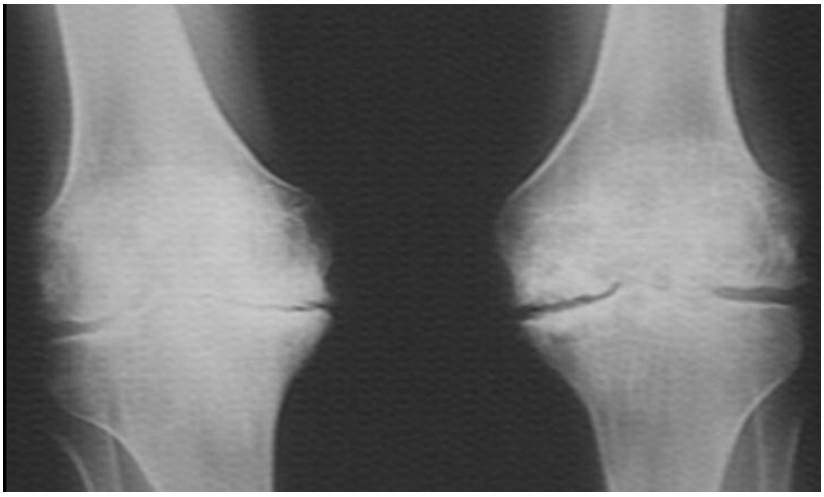

Figure 2: Radiographs plains of knee at right showing osteoporosis, widening of the epichondral notch of the knee, epiphyseal overgrowth, bone cysts, joint space irregularity and global narrowing

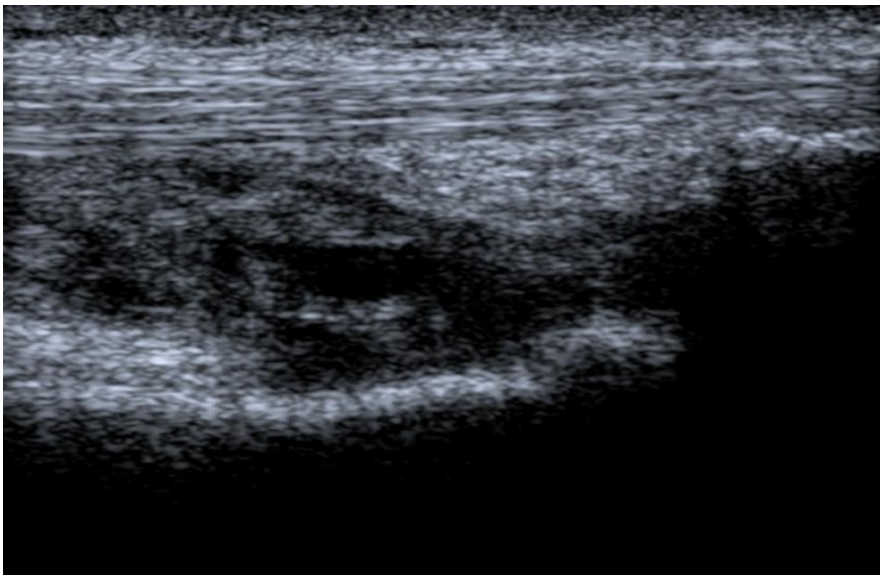

Figure 3: Ultrasonographic longitudinal scan of quadricipital recessus of knee at right showing synovial hypertrophy associated to hypoechogenic effusion in the quadriceps recesses