Case Report

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 hemorrhages and joint hemorrhages. 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 presented with recurrent hemarthroses involving the right knee, leading to chronic pain and leisure of functional score. Clinical examination showed atrophy of the quadriceps and a swollen knee, with moderate stiffness of this joint.

Radiography revealed 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 intra-articular injection of triamcinolone hexacetonide following fresh frozen plasma transfusion. Decreases in swelling and joint stiffness were recorded after rehabilitation.

Discussion: Factor VII (FVII) deficiency is a rare inherited coagulation disorder, accounting for one symptomatic individual per every 500,000. Hemarthrosis is less common than hemophilia, although the characteristics of joint destruction are similar in the two conditions. For the case reported here, repetitive hemarthrosis led to chronic arthropathy with a negative impact on the functional score.

Bleeding into joints requires prophylatic replacement therapy. Intra-articular injection of corticosteroids may be useful as a palliative measure for pain and inflammation, particularly in countries with limited resources.

Key words: Factor VII (FVII) deficiency, hemarthrosis, chronic arthropathy, fresh frozen plasma, intra-articular 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 hemorrhages and joint hemorrhages [2]. Here we present a case of congenital factor VII deficiency with chronic knee arthropathy.
Case Report

Mrs G.H, 15 years old, suffered from FVII deficiency, diagnosed during childhood. Her parents were consanguineous, and she had two brothers followed for the same bleeding disorder. The patient did not have any health insurance coverage. This girl experienced recurrent spontaneous hemarthrosis in her right knee starting at years of age (figure 1). Hemarthroses lead to chronic pain and use of analgesics. Clinical examination found atrophy of quadriceps with a 7-centimeter decrease in the muscle perimeter, as compared to the left thigh. The affected knee was swollen and showed moderate stiffness. 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.

Plain radiographs 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 hypoechogetic effusion in the quadriceps recesses (figure 3).

The diagnosis of chronic arthropathy secondary to factor VII deficiency was retained. Our patient had had an intra-articular injection of triamcinolone hexacetone following fresh frozen plasma transfusion. Decreases in swelling and joint stiffness were recorded after rehabilitation. Furthermore, our patient expressed improvement of the Lesquene and the scores.

Discussion

Congenital factor VII deficiency is a rare autosomal-recessive bleeding disorder, accounting for one symptomatic individual per 500,000 [1,2]. Frequency is higher in countries where consanguineous marriage is more common, as in the case of Morocco. This inherited coagulation disorder 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 should be noted that, because of the poor correlation between FVII levels and the bleeding tendency, FVIIIC levels cannot be used to distinguish classes of severity [5]. Thus, severity of FVII deficiency is classified on a clinical basis. Since chronic
arthropathy has occurred in our patient, her FVII deficiency is qualified as severe. As in hemophilia, recurrent hemarthrosis leads to chronic arthropathy, a 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, intra-venous administration of recombinant FVIIa. Management of arthropathy could be similar to that in hemophilia cases. Bleeding into joints requires prophylactic replacement therapy. Besides 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 chlorhydrate [6,7]. However, 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 cases of prolonged prothrombin time with normal partial thromboplastin time. Bleeding into the joints requires prophylactic replacement therapy. Intra-articular injection of corticosteroids may be useful as a palliative measure for pain and inflammation, particularly in countries with limited resources.

Competing interests
The authors declared no competing interest.

Funding
None.

Provenance and peer review
Not commissioned; externally peer reviewed.

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