Severe elbow arthropathy in a patient with congenital afibrinogenemia: a case report

Reidy, K; Brand, B; Jost, B
Severe Elbow Arthropathy in a Patient with Congenital Afibrinogenemia: A Case Report

Kerstin Reidy, Brigitte Brand and Bernhard Jost

This information is current as of March 17, 2011

Reprints and Permissions
Click here to order reprints or request permission to use material from this article, or locate the article citation on jbjs.org and click on the [Reprints and Permissions] link.

Publisher Information
The Journal of Bone and Joint Surgery
20 Pickering Street, Needham, MA 02492-3157
www.jbjs.org
Severe Elbow Arthropathy in a Patient with Congenital Afibrinogenemia

A Case Report

By Kerstin Reidy, MD, Brigitte Brand, MD, and Bernhard Jost, MD

Investigation performed at the Department of Orthopedics, Balgrist, University of Zurich, Zurich, and the Department of Hematology, University Hospital of Zurich, Zurich, Switzerland

Afibrinogenemia is a rare coagulation deficiency, inherited as an autosomal recessive trait. It can be manifested in homozygotes or compound heterozygotes by a severe bleeding tendency caused by deficiency of fibrinogen (factor I). The estimated prevalence in Italy is about 1:1,000,000, and 0.2% of the patients with a coagulation deficiency in the United Kingdom have afibrinogenemia. In Switzerland, nine patients are listed in the national hemophilia registry.

Among severe bleeding symptoms, umbilical cord bleeding is the most frequent and life-endangering event. Intracranial hemorrhage is the major cause of death and is the primary reason for prophylaxis. Less severe findings, such as spontaneous muscle hematomas, epistaxis, and menorrhagia, are also frequent.

Patients with afibrinogenemia have remarkably fewer spontaneous hemarthroses in contrast to better known bleeding disorders such as hemophilia A and hemophilia B.

We report the case of a patient with congenital afibrinogenemia who had severe elbow arthropathy and was treated with a total elbow arthroplasty. The patient was informed that data concerning the case would be submitted for publication, and he consented.

Case Report

A forty-seven-year-old man was referred to our institution with a severe arthropathy of the left, nondominant elbow. Afibrinogenemia was diagnosed at birth. For many years, he was administered 4 g of Haemocomplettan HS (human fibrinogen; CSL Behring, Bern, Switzerland) intravenously every two weeks. The causative mutation in his family (the parents were non-consanguineous, and his brother and two cousins were also affected) was identified as a homozygous deletion of approximately 11 kb of the fibrinogen alpha chain gene. Over the years, the left elbow became increasingly symptomatic secondary to recurrent hemarthroses. The elbow was always the primary joint involved, although mild osteoarthritic changes were noted in the ankles. In 1991, at the age of thirty-one years, he had an open elbow synovectomy, which decreased the frequency of joint bleeding substantially.

The elbow remained minimally symptomatic until 2006, when the patient was forty-six years old, and he presented with pain at rest, severe activity-related pain, and night pain. He also had difficulty performing the activities of daily living. Nevertheless, he was able to work full time in his profession as a telematics technician. Physical examination revealed a mild effusion. Range of motion was painful and severely limited, with flexion to 80° and a 45° extension loss. Forearm rotation was also painful and mildly limited. There were no neurologic deficits. The preoperative Mayo Elbow Performance Score was 40 points (15 points for pain, 5 points for motion, 10 points for stability, and 10 points for function).

Biplanar radiographs of the elbow showed severe degenerative arthritis according to the modified Arnold and Hilgartner classification of hemophilic arthropathy. Because joint salvage procedures such as arthroscopic or open débridement did not seem promising, a total elbow arthroplasty with use of a semiconstrained Coonrad-Morrey prosthesis (Zimmer, Warsaw, Indiana) was performed in 2006. Preoperatively, intraoperatively, and postoperatively, the patient was closely monitored and fibrinogen was administered under the supervision of a hematologist. Prior to anesthesia, the patient received 4 g of Haemocomplettan HS. This was his usual dose, given every two weeks, and was known to increase the fibrinogen level to >1 g/L.

At the time of surgery, the elbow joint was approached through a straight posterior incision. The ulnar nerve was exposed and mobilized, and at the end of the procedure it was transposed anteriorly. The triceps was reflected from the tip of the olecranon in a medial to lateral dissection with use of the technique described by Morrey et al. The joint was very contracted, and the radial head was resected to gain better exposure and adequate visualization. All of the joint surfaces showed complete loss of articular cartilage and eburnation of the bone. After humeral and ulnar preparation, the compo-
nents were implanted simultaneously with use of bone cement fixation. At the end of the procedure, flexion was 140° and loss of extension was 10°. The triceps was reattached to the olecranon. The postoperative radiographs showed proper positioning of the implant.

During surgery and in the early postoperative period, our aim was to maintain a permanent level of fibrinogen of 0.5 g/L. Therefore, 1 g of Haemocomplettan HS was administered intravenously every day, and the fibrinogen level was checked every other day.

There were no early or late postoperative complications, including nerve palsy or major bleeding.

Postoperatively, the elbow was immobilized in an anterior splint in full extension. From the second postoperative day, free active-assisted and passive mobilization of the elbow was allowed. On the fourth postoperative day, there was deep wound bleeding with slight swelling of the elbow, which did not affect the postoperative course. The fibrinogen level was 0.3 g/L.

The patient was hospitalized for ten days to closely monitor bleeding and fibrinogen levels. For the first six weeks postoperatively, 2 g of Haemocomplettan HS was given twice a week; then 3 g was given weekly for another twelve weeks before returning to the preoperative scheme of 4 g of Haemocomplettan HS every two weeks. Fibrinogen levels (a minimum of 0.3 g/L and a peak at a maximum of 1.4 g/L) were checked with every change of dose.

During this eighteen-week period, the patient received thromboprophylaxis with 5000 IU of low-molecular-weight heparin. We were very cautious because of the thromboembolic events in his family as well as case reports about thromboembolism in afibrinogenic patients and therefore chose this long period of prophylaxis.

By six months, the range of motion had plateaued with flexion of 135°, loss of extension of 30°, and full forearm rotation. At the one and two-year follow-up visits, the patient was very satisfied with the result and there was no loss of range of motion. Two years after surgery, the range of motion of the left elbow was unchanged compared with that at the time of the six-month follow-up. This is comparable with the results reported by Kamineni et al. and Chapman-Sheath et al. The patient was free of pain and able to perform all activities of daily living. The Mayo Elbow Performance Score was 95 points. Radiographs made two years postoperatively showed a satisfactory position of the implant with no signs of loosening.

**Discussion**

A fibrinogenemia is a rare hereditary coagulation disorder due to defects in the Aα, Bβ, or γ chains of fibrinogen, resulting in malfunction in the formation of cross-linked fibrin clots in the final common pathway of both the extrinsic and intrinsic blood-clotting cascades. Recently, genetic studies have substantially increased our understanding of the underlying...
molecular defects in afibrinogenemia; however, optimum treatment is not as well established as it is for hemophilia A and B because of the limited number of cases\textsuperscript{3,7,11,14,15}.

Compared with more common coagulation disorders, such as hemophilia A and B, some clinical manifestations, such as muscle hematomas, epistaxis, or central nervous system bleeding, are as frequent, whereas spontaneous hemarthroses are rare in afibrinogenemia\textsuperscript{3,4,14,15}. In hemophilia, arthropathy has been well described, with the elbow being the second most commonly affected joint besides the knee. To our knowledge, this is not only the first reported case of severe elbow arthropathy in a patient with afibrinogenemia but it appears to be the first reported case of a patient with severe arthritic joint involvement in this disease. The exact pathogenesis of arthropathy in afibrinogenemia is unknown. It is known that in hemophilia the pathogenesis is multifactorial and includes cartilage-mediated as well as inflammatory synovium-mediated components, resulting in a fibrotic and destroyed joint as in the patient reported here\textsuperscript{3}.

The cases of only a few patients with a coagulation disorder who had arthropathy and were managed with a total elbow arthroplasty are documented in the literature\textsuperscript{1,2,7,11,12}. Kamineni et al. reported on five patients with hemophilic arthropathy who were treated with a linked, semiconstrained total elbow replacement. Two of the five patients had severe complications (uncontrollable hemorrhage in one and a deep wound infection in both patients), and one patient had a minor complication (recurrent intra-articular hemorrhages). At a mean follow-up of 122 months, the patients had improved substantially in terms of pain compared with preoperatively. Elbow range of motion was also substantially improved at the time of the final evaluation. The postoperative Mayo Elbow Performance Score was 90 points, clearly showing an increase compared with the score of 24 points preoperatively\textsuperscript{12}. In another study, Chapman-Sheath et al. reported on seven total elbow replacements in five patients with hemophilia\textsuperscript{13}. They described major complications in three patients, including ulnar nerve palsy, axillary vein thrombosis, and septic loosening of the arthroplasty components in one patient each. The mean postoperative range of motion was 122° of flexion and an extension loss of 28.5° at a mean follow-up of forty-two months. The results are comparable with those in our patient, in whom flexion improved to 135° with an extension loss of 30°, an arc of motion that is sufficient to perform all of the activities of daily living\textsuperscript{17}. In patients with joint hemophilia, contractures are very common, resulting from recurrent intra-articular and intramuscular bleeding episodes. Correction of hemophilic contractures during total joint replacement can be very challenging; therefore, restoration of unlimited activities of daily living, rather than the achievement of anatomic or radiographic normality, is the primary goal\textsuperscript{18}.

Because of the repetitive bleeding episodes and the similar radiographic appearance of the arthropathy, we hypothesize a close similarity between the development of arthropathy in patients with hemophilia and in patients with afibrinogenemia. As in all bleeding disorders, patients with afibrinogenemia require a multidisciplinary team approach with close cooperation of the surgeon with a hematologist specialized in coagulation disorders.

---

**Severe Elbow Arthropathy in a Patient with Congenital Afibrinogenemia**

Kerstin Reidy, MD  
Bernhard Jost, MD  
Department of Orthopedics, Balgrist, University of Zurich,  
Forchstrasse 340, CH-8008 Zurich, Switzerland  
E-mail address for B. Jost: bernhard.jost@balgrist.ch

Brigitte Brand, MD  
Department of Hematology,  
University Hospital of Zurich,  
Rämistrasse 100, CH-8091 Zurich, Switzerland

---

**References**


