

A 16-year-old boy is brought to the pediatrician with pain and limited motion of the right knee. He first noticed knee pain and swelling 6 months earlier, and the pain has gradually worsened since then. He has no history of fever, local erythema, or trauma. The patient reports that his right knee and right ankle have swelled several times before. He has a history of easy bruising since childhood and had an episode of excessive bleeding after a tooth extraction. His uncle had similar problems. Which of the following is the most likely cause of this patient's joint pain?

- ☐ A. Avascular necrosis
- ☐ B. Crystal deposition
- ☐ C. Hemosiderin deposition and fibrosis
- ☐ D. Immunologic tissue injury
- ☐ E. Occult traumatic injury
- ☐ F. Repetitive microtrauma
- ☐ G. Vector-borne illness

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- ☐ A. Avascular necrosis [3%]
- ☐ B. Crystal deposition [1%]
- ☒ C. Hemosiderin deposition and fibrosis [55%]
- ☐ D. Immunologic tissue injury [6%]
- ☐ E. Occult traumatic injury [12%]
- ☐ F. Repetitive microtrauma [22%]
- ☐ G. Vector-borne illness [0%]

[Proceed to Next Item](#)**Explanation:****User Id:** XXXXXXXXXX

Hemophilia A & B	
Inheritance	<ul style="list-style-type: none">• X-linked recessive
Clinical features	<ul style="list-style-type: none">• Delayed/prolonged bleeding after mild trauma or procedure<ul style="list-style-type: none">○ Hemarthrosis, hemophilic arthropathy○ Intramuscular hematomas○ Gastrointestinal or genitourinary tract bleeding
Laboratory findings	<ul style="list-style-type: none">• Prolonged activated partial thromboplastin time• Normal platelet count, bleeding time, prothrombin time• Decreased or absent factor VIII (hemophilia A) or factor IX (hemophilia B) activity

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Treatment	<ul style="list-style-type: none">• Administration of factor VIII or factor IX• Desmopressin for mild hemophilia A

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This patient's history of easy bruising since childhood, excessive bleeding after a dental procedure, and recurrent joint swelling, along with his family history, are most consistent with hemophilia. Hemophilia A and B, which are caused by deficiencies of Factor VIII and Factor IX, respectively, are bleeding disorders inherited in an **X-linked recessive** pattern. Although only **male patients** are severely affected, female carriers may be mildly symptomatic. Hemophilia can affect nearly every organ system and result in long-term complications such as hemophilic arthropathy.

Hemophilic arthropathy, a late complication that occurs in both types of hemophilia and is a significant source of morbidity, is thought to be caused by **iron/hemosiderin deposition** leading to **synovitis** and **fibrosis** within the joint. However, other mechanisms may also be involved. Hemophilic arthropathy typically presents with chronic worsening joint pain and swelling and can result in contractures of the joint and limited range of motion. It is most common in patients with a history of recurrent hemarthroses. Although severe hemophilic arthropathy may be visible on **plain films**, **magnetic resonance imaging** allows for much earlier detection and characterization of

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However, **early prophylactic treatment** with factor concentrates can significantly reduce the risk of developing arthropathy.

(Choice A) Avascular necrosis of the femoral head is seen in children with sickle cell disease and in Legg-Calvé-Perthes disease (LCP). LCP, or idiopathic avascular necrosis of the femoral head, typically presents with limp or hip pain and is most common in children age 4-12. It is rare in adolescents. Sickle cell disease may also cause recurrent vaso-occlusive episodes, which may affect the joints, but are not associated with excessive bruising or bleeding.

(Choice B) Urate and calcium pyrophosphate deposition are the mechanisms of arthritis in gout and pseudogout, respectively. Both conditions are uncommon in children.

(Choice D) Immunologic tissue injury occurs in a variety of conditions, such as rheumatoid arthritis and psoriatic arthritis. But while inflammatory arthritis may cause joint swelling, these conditions would not explain the patient's bleeding symptoms.

(Choice E) Although minor unnoticed trauma can result in episodes of hemarthrosis in patients with hemophilia, the prolonged course of symptoms in this patient does not suggest acute bleeding into the joint. Rather, it is more consistent with a chronic inflammatory process due to iron/hemosiderin deposition.

(Choice F) Repetitive microtrauma, also known as "the wear and tear" phenomenon, is thought to play a role the development of osteoarthritis, which is most common in adults age >55.

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(Choice G) Lyme disease is a vector-borne illness that can result in arthritis of the large joints as a late finding. Although Lyme arthritis can present with swelling and limited range of motion, it would be unlikely to account for this patient's other findings.

Educational objective:

Hemophilic arthropathy is a delayed consequence of recurrent hemarthrosis. It is associated with hemosiderin deposition leading to synovitis and fibrosis within the joint. The risk of hemophilic arthropathy can be significantly reduced by prophylactic treatment with factor concentrates.

References:

1. [Pathobiology of hemophilic synovitis I: overexpression of mdm2 oncogene.](#)
2. [Hemophilic arthropathy: a review of imaging and staging.](#)
3. [Pathogenesis of haemophilic arthropathy.](#)

Media Exhibit

hilar arthropathy

