

A 3-year-old boy is brought to the physician by his stepfather with right-knee swelling that started after he fell while running. The boy is generally healthy but has always bruised easily. The mother does not have a history of easy bruising or bleeding problems; the history of the biological father is unknown. Examination shows an uncomfortable-appearing boy with significant swelling of the right knee with a large joint effusion. The patient has pain with movement at the knee and there is limited range of motion. Multiple ecchymoses are noted on his anterior shins bilaterally. The remainder of the physical examination is normal. Radiograph of the right knee shows a large effusion but no fracture. Which of the following is the most appropriate next step in management of this patient?

- ☐ A. Complete blood count and coagulation studies
- ☐ B. Complete skeletal survey
- ☐ C. Liver function tests
- ☐ D. Reassurance and supportive management
- ☐ E. Referral to child protective services
- ☐ F. Type 1 collagen assay

Submit

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- ✓ ☒ A. Complete blood count and coagulation studies [89%]
- ☐ B. Complete skeletal survey [4%]
- ☐ C. Liver function tests [0%]
- ☐ D. Reassurance and supportive management [3%]
- ☐ E. Referral to child protective services [3%]
- ☐ F. Type 1 collagen assay [1%]

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Explanation:

User Id: XXXXXXXXXX

Bleeding disorders			
Type	Symptoms	Examples	Laboratory results
Clotting defect	<ul style="list-style-type: none">• Hemarthrosis• Deep tissue hematomas	<ul style="list-style-type: none">• Hemophilia A• Hemophilia B	↑Activated partial thromboplastin time
Platelet aggregation defect	<ul style="list-style-type: none">• Easy or prolonged	<ul style="list-style-type: none">• von Willebrand disease• Bernard-Soulier	Abnormal platelet function testing

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Platelet aggregation defect	<ul style="list-style-type: none">• Easy or prolonged mucosal bleeding• Ecchymoses• Petechiae	<ul style="list-style-type: none">• von Willebrand disease• Bernard-Soulier syndrome	Abnormal platelet function testing
Thrombocytopenia		<ul style="list-style-type: none">• Idiopathic thrombocytopenic purpura• Leukemia	↓Platelet count

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The acute onset of joint effusion following minor trauma is concerning for a hemarthrosis and should raise concern for a **bleeding disorder**. This child's examination finding of hemarthrosis and multiple ecchymoses and history of easy bruising are all concerning for a bleeding disorder.

The most common bleeding disorders in children include the hemophilias and von Willebrand disease. **Hemophilia** and other coagulation disorders commonly present with **hemarthroses**, soft-tissue hematomas, and intramuscular hematomas. In contrast, platelet aggregation disorders such as von Willebrand present predominantly with **easy mucosal bleeding**, ecchymoses, or petechiae. Initial screening studies should include coagulation studies as well as a complete blood count to evaluate for thrombocytopenia and anemia.

(Choices B and E) The bruising due to nonaccidental trauma overlaps with that of bleeding disorders. However, **nonaccidental trauma** should be suspected when no history is offered, when the history changes significantly with retelling, or when the injury

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(Choices B and E) The bruising due to nonaccidental trauma overlaps with that of bleeding disorders. However, **nonaccidental trauma** should be suspected when no history is offered, when the history changes significantly with retelling, or when the injury is inconsistent with the patient's history or developmental stage. Concerning types of injury include rib fractures, skull fractures, metaphyseal corner fractures, patterned bruising, genital injuries, or fractures in varying stages of healing.

(Choice C) Liver function tests are obtained in the workup of patients with abnormal coagulation studies as advanced liver disease can cause coagulopathy. They are not used for screening for bleeding disorders. Liver function tests may also be obtained in children with suspected nonaccidental trauma to screen for intra-abdominal injury.

(Choice D) Reassurance and supportive management are appropriate for children presenting with minor contusions or sprains. They are not appropriate for a child with a hemarthrosis, which requires further evaluation.

(Choice F) A type 1 collagen assay is used to diagnose osteogenesis imperfecta, or "brittle bone disease." Common findings include numerous fractures in multiple stages of healing, presence of blue sclerae, and short stature. The lack of any history of fractures makes osteogenesis imperfecta unlikely.

Educational objective:

Hemarthrosis after minor trauma in a young child is suspicious for a bleeding disorder. Screening should include coagulation studies and a complete blood count. Nonaccidental trauma should be suspected in children with injuries inconsistent with the history or developmental stage.

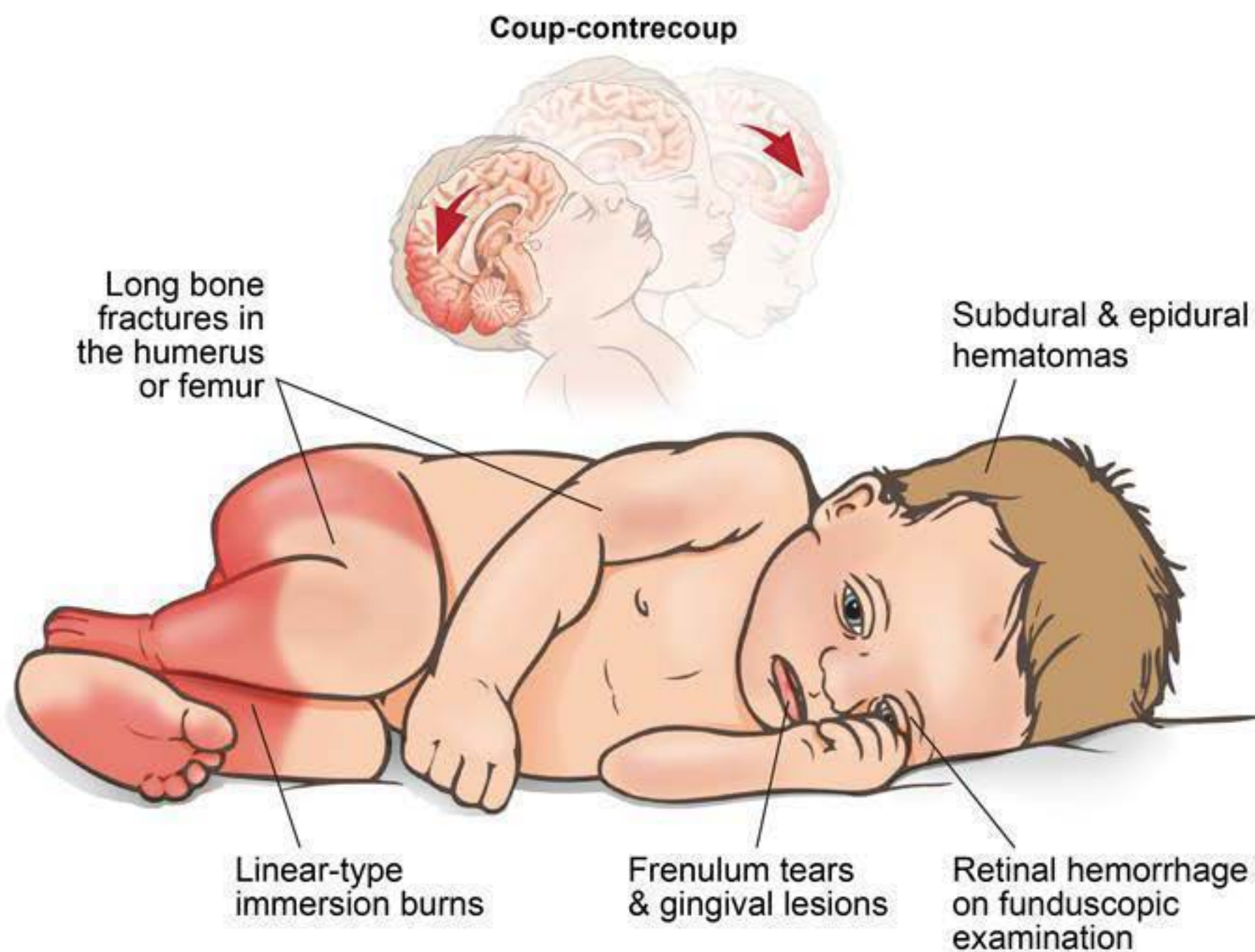
References:

1. **Easy bruisability.**
2. **Evaluating for suspected child abuse: conditions that predispose to bleeding.**

Media Exhibit

signs & symptoms in abusive head trauma

Injury patterns in nonaccidental trauma

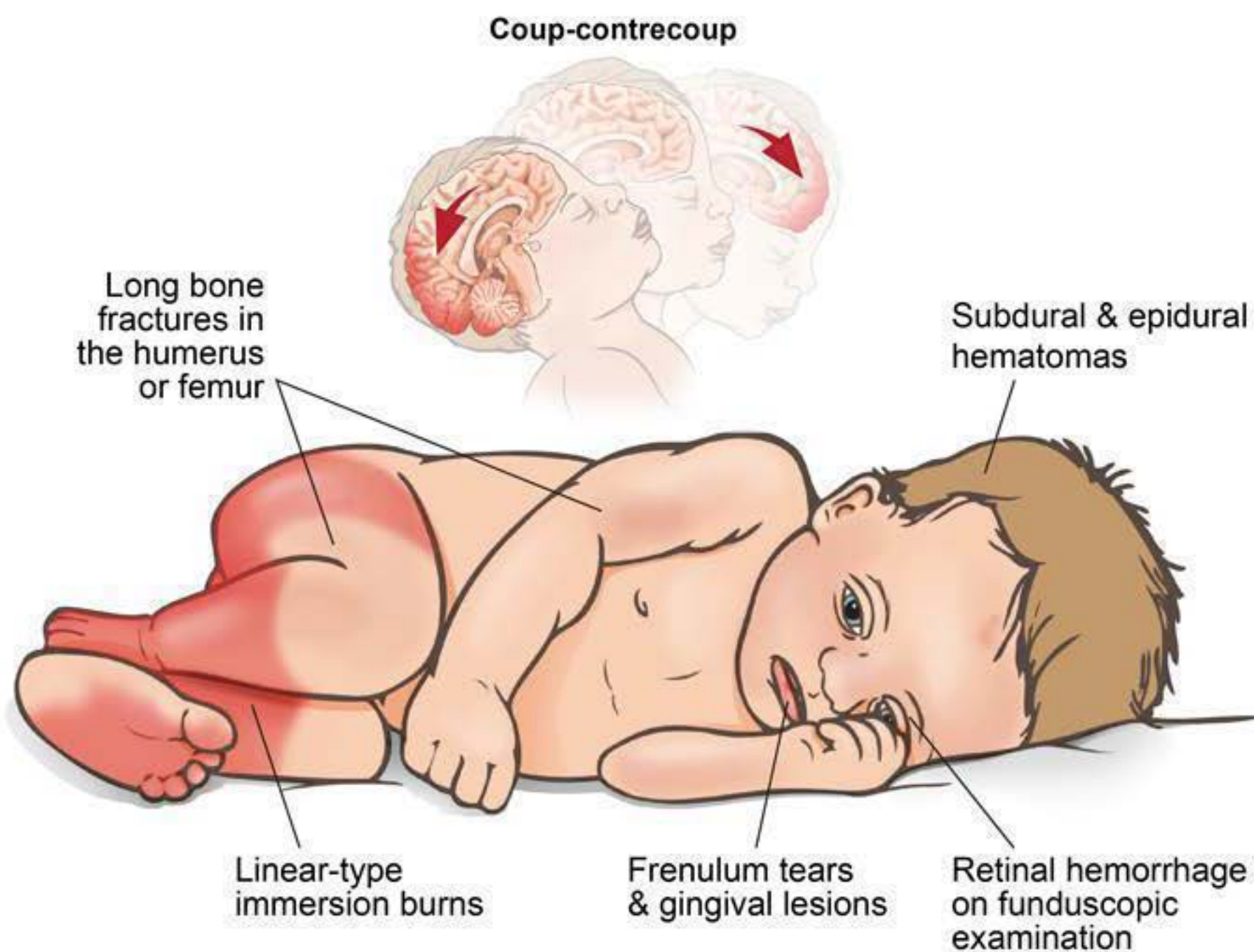


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Media Exhibit

signs & symptoms in abusive head trauma

Injury patterns in nonaccidental trauma



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An 18-month-old boy is brought to the emergency department due to blood in his stool, which the parents noticed when changing his diaper. The infant has had no previous bleeding and has been eating and drinking normally. He has a history of recurrent otitis media, frequent herpes labialis, and 2 episodes of pneumonia. Vital signs are normal. On examination, the patient is well developed, well nourished, and has a fair complexion. He has eczema on his cheeks, trunk, and extremities. Scattered petechiae are also visible on his lower extremities. The remainder of the physical examination is unremarkable. Laboratory studies show a platelet count of $24,000/\text{mm}^3$ and a leukocyte count of $9,000/\text{mm}^3$. Peripheral smear confirms the low platelet count and that the platelets are small. Genetic testing confirms the diagnosis. Which of the following processes is most likely affected by this patient's gene mutation?

- ☐ A. Antibody class switching [17%]
- ☒ B. Cytoskeleton regulation [38%]
- ☐ C. DNA repair [20%]
- ☐ D. Hydrogen peroxide production [5%]
- ☐ E. Maturation of T cells [19%]

Proceed to Next Item

Explanation:

User Id: [REDACTED]

Wiskott-Aldrich syndrome	
Etiology	<ul style="list-style-type: none">• X-linked recessive defect in WAS protein gene• Impaired cytoskeleton changes in leukocytes, platelets
Clinical features	<ul style="list-style-type: none">• Eczema• Microthrombocytopenia (small platelets, low platelet count)• Recurrent infections
Treatment	<ul style="list-style-type: none">• Stem cell transplant