

A 7-year-old boy is brought to the emergency department for a suspected fracture. His mother says that he was running around the house and fell on the carpet an hour ago. The boy has been crying and complaining of thigh pain since the fall. He did not hit his head or lose consciousness. His past medical history is notable for mild hearing loss and multiple fractures after seemingly minor injuries. On physical examination, he has decreased muscle tone throughout. The right thigh is markedly tender to palpation with obvious deformity. Multiple bruises in various stages of healing are present on his extremities. His eye examination is **shown**. Which of the following is most likely associated with this patient's underlying condition?

- ☐ A. Aortic root dilatation
- ☐ B. Intellectual disability
- ☐ C. Opalescent teeth
- ☐ D. Retinal hemorrhages
- ☐ E. Subdural hematoma
- ☐ F. Vitamin D deficiency

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- ☐ A. Aortic root dilatation [15%]
- ☐ B. Intellectual disability [3%]
- ☒ C. **Opalescent teeth** [73%]
- ☐ D. Retinal hemorrhages [3%]
- ☐ E. Subdural hematoma [2%]
- ☐ F. Vitamin D deficiency [3%]

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

This patient has osteogenesis imperfecta (OI), a connective tissue disorder most commonly inherited from an **autosomal dominant** mutation of *COL1A1*. The disorder has a varying spectrum of severity, from mild (type I), moderate (types III-IX), to fatal perinatal (type II) disease. Patients with all types of OI have **osteopenia**, and the diagnosis should be suspected in any patient with **blue sclerae**. Other manifestations depend on the severity of the disorder and can include **recurrent fractures**, easy bruisability, hypotonia, and **hearing loss**. Many patients with OI also have **dentinogenesis imperfecta**, an opalescent blue-gray to yellow-brown discoloration caused by discolored dentin shining through the translucent and weak enamel. Both primary and permanent teeth are affected.

(Choice A) Cardiac anomalies such as aortic root dilatation are not usually seen in OI. Aortic root dilatation can be seen in other connective tissue disorders such as Marfan syndrome.

(Choice B) Patients with OI typically have normal intelligence.

(Choices D and E) Although fractures and bruises in various stages of healing should

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This patient has osteogenesis imperfecta (OI), a connective tissue disorder most commonly inherited from an **autosomal dominant** mutation of *COL1A1*. The disorder has a varying spectrum of severity, from mild (type I), moderate (types III-IX), to fatal perinatal (type II) disease. Patients with all types of OI have **osteopenia**, and the diagnosis should be suspected in any patient with **blue sclerae**. Other manifestations depend on the severity of the disorder and can include **recurrent fractures**, easy bruisability, hypotonia, and **hearing loss**. Many patients with OI also have **dentinogenesis imperfecta**, an opalescent blue-gray to yellow-brown discoloration caused by discolored dentin shining through the translucent and weak enamel. Both primary and permanent teeth are affected.

(Choice A) Cardiac anomalies such as aortic root dilatation are not usually seen in OI. Aortic root dilatation can be seen in other connective tissue disorders such as Marfan syndrome.

(Choice B) Patients with OI typically have normal intelligence.

(Choices D and E) Although fractures and bruises in various stages of healing should raise suspicion for physical abuse, this diagnosis is less likely as the blue sclerae suggest an underlying collagen disorder. In addition, inflicted bruises typically occur in central areas (eg, back, buttocks, neck, cheeks); accidental bruises of childhood tend to occur on extremities. However, if abuse is suspected or if the child has altered mental status or neurologic deficits, evaluation for retinal hemorrhages and subdural hematomas should be pursued.

(Choice F) Vitamin D deficiency can lead to rickets and pathologic fractures. However, scleral abnormalities typically do not occur, making this associated problem less likely.

Educational objective:

Osteogenesis imperfecta is associated with blue sclerae, hearing loss, recurrent fractures, and opalescent teeth. Patients with osteogenesis imperfecta have normal intelligence.

References:

1. **Osteogenesis imperfecta.**
2. **Osteogenesis imperfecta: diagnosis and treatment.**
3. **Pediatric dental management of a patient with osteogenesis imperfecta and dentinogenesis imperfecta.**

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era Blue sclera



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era Blue sclera



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Osteogenesis imperfecta

