

A 3-year-old boy is brought to the office by his 27-year-old white mother for the evaluation of recurrent bone fractures. His first fracture was that of the femur, and occurred when he was 6 months old. He had a fracture of the wrist 4 months ago. His mother also has a history of multiple fractures since childhood. She lost all her teeth at a very early age and is complaining of deafness. Her husband has a history of severe alcohol abuse. On examination, both mother and son have blue sclerae. What is the most likely involved disease process?

- ☐ A. Mutations in type 1 collagen
- ☐ B. Mutations in fibrillin1 gene
- ☐ C. Child abuse
- ☐ D. Vitamin-D deficiency
- ☐ E. Congenital syphilis

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- ☒ A. Mutations in type 1 collagen [93%]
- ☐ B. Mutations in fibrillin1 gene [5%]
- ☐ C. Child abuse [1%]
- ☐ D. Vitamin-D deficiency [1%]
- ☐ E. Congenital syphilis [1%]

[Proceed to Next Item](#)

Explanation:

User Id: 

The above child is most likely suffering from osteogenesis imperfecta, which is an inherited connective tissue disorder caused by a mutation in the genes coding for type I collagen. Since type I collagen is an important structural protein that is present in the skin, sclera, bone, tendon and ligament, patients with this disorder present with multiple recurrent fractures, blue sclera, hearing loss, joint laxity, short stature, and scoliosis.


(Choice B) Marfan syndrome is one of the most common connective tissue disorders, and is caused by mutation of the fibrillin-1 gene. Its mode of inheritance is autosomal dominant. The common features of this disorder include skeletal manifestations (e.g., arachnodactyly, hypermobility of joints), ectopia lentis, and aortic root dilatation.

(Choice C) Child abuse suspected in the presence of any of the following: retinal hemorrhages, burns, bruising, fractures or abrasions. This is a good differential diagnosis for the given case; however, the features of this patient (e.g., fractures, blue sclerae, family history of fractures and hearing loss) are very typical of osteogenesis imperfecta.

(Choice D) Children with rickets have slow growth and skeletal deformities. Blue sclerae and hearing loss are not the features of rickets.

(Choice E) Early manifestations of congenital syphilis include jaundice, rash,

- ☐ B. Mutations in fibrillin-1 gene [5%]
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- ☐ E. Congenital syphilis [1%]

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

The above child is most likely suffering from osteogenesis imperfecta, which is an inherited connective tissue disorder caused by a mutation in the genes coding for type I collagen. Since type I collagen is an important structural protein that is present in the skin, sclera, bone, tendon and ligament, patients with this disorder present with multiple recurrent fractures, blue sclera, hearing loss, joint laxity, short stature, and scoliosis.

(Choice B) Marfan syndrome is one of the most common connective tissue disorders, and is caused by mutation of the fibrillin-1 gene. Its mode of inheritance is autosomal dominant. The common features of this disorder include skeletal manifestations (e.g., arachnodactyly, hypermobility of joints), ectopia lentis, and aortic root dilatation.

(Choice C) Child abuse suspected in the presence of any of the following: retinal hemorrhages, burns, bruising, fractures or abrasions. This is a good differential diagnosis for the given case; however, the features of this patient (e.g., fractures, blue sclerae, family history of fractures and hearing loss) are very typical of osteogenesis imperfecta.

(Choice D) Children with rickets have slow growth and skeletal deformities. Blue sclerae and hearing loss are not the features of rickets.

(Choice E) Early manifestations of congenital syphilis include jaundice, rash, lymphadenopathy, rhinitis and hepatosplenomegaly. Late manifestations occur around two year of age; these include saber shins, keratitis, Hutchinson's teeth, saddle-nose deformity and deafness. The above child has typical features of osteogenesis imperfecta, not congenital syphilis.

Educational Objective:

Osteogenesis imperfecta is caused by mutations in type 1 collagen. Its typical features are blue sclera and recurrent fractures.

*Extremely high yield question for USMLE step-1 and step-2

Time Spent: 2 seconds

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