

An 11-month-old African-American boy is brought to the pediatrician for a well-child visit. He was born full-term and is primarily breastfed. He eats homemade baby food but not store-bought food. The infant has been growing and gaining weight well. He has started cruising but is not yet walking. He is able to say "mama" and "dada." The boy has no chronic medical issues and takes no medications. He has a cousin with achondroplasia. On examination, the anterior fontanel is wide open. Palpation shows pliable skull bones without step-offs. Bony prominences of the costochondral junctions are noted bilaterally. Genu varum is present. The remainder of his examination is normal. Which of the following is the most likely cause of this infant's findings?

- ☐ A. Abnormal thyroid function
- ☐ B. Congenital infection
- ☐ C. Nonaccidental trauma
- ☐ D. Normal variant
- ☐ E. Nutritional deficiency
- ☐ F. Skeletal dysplasia



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- ☐ A. Abnormal thyroid function [1%]
- ☐ B. Congenital infection [1%]
- ☐ C. Nonaccidental trauma [0%]
- ☐ D. Normal variant [17%]
- ☒ E. Nutritional deficiency [65%]
- ☐ F. Skeletal dysplasia [15%]

[Proceed to Next Item](#)

Explanation:

User Id: [REDACTED]

Vitamin D deficiency rickets	
Risk factors	<ul style="list-style-type: none"><li>• Increased skin pigmentation</li><li>• Exclusive breastfeeding</li><li>• Inadequate sun exposure</li><li>• Maternal vitamin D deficiency</li></ul>
Clinical manifestations	<ul style="list-style-type: none"><li>• Craniotabes ("ping-pong ball" skull)</li><li>• Delayed fontanel closure</li><li>• Enlarged<ul style="list-style-type: none"><li>• Skull (frontal bossing)</li><li>• Costochondral joints ("rachitic rosary")</li><li>• Long-bone joints (wrist widening)</li></ul></li></ul>



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X-ray features	<ul style="list-style-type: none"> <li>• Osteopenia</li> <li>• Metaphyseal cupping &amp; fraying</li> <li>• Epiphyseal widening</li> </ul>
Serum laboratory findings	<ul style="list-style-type: none"> <li>• Calcium: Normal to ↓</li> <li>• Phosphorous: Normal to ↓</li> <li>• Alkaline phosphatase: ↑↑</li> <li>• Parathyroid hormone: ↑</li> <li>• 25-OH vitamin D: ↓</li> </ul>

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This infant's examination findings of costochondral joint hypertrophy ("**rachitic rosary**"), genu varum (**femoral and tibial bowing**), large anterior fontanel, and **craniotabes** are consistent with rickets. Nutritional rickets is caused by deficiency of vitamin D or calcium, with vitamin D deficiency being more common. The primary source of vitamin D is sunlight. People with **dark skin pigmentation** are at increased risk of vitamin D



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Clinical manifestations typically appear after several months of deficiency. X-ray findings include **genu varum** (blue arrows) in weight-bearing children, **enlargement of the costochondral joints**, and **metaphyseal cupping and fraying**. Treatment consists of vitamin D repletion with 1000-2000 IU daily.

**(Choice A)** Congenital hypothyroidism can present with a large anterior fontanel, lethargy, feeding difficulties, and macroglossia; it does not cause bony abnormalities.

**(Choice B)** Congenital syphilis can present with bony abnormalities such as frontal bossing, anterior bowing of the shins ("saber shins"), saddle-nose deformity, and notched, gap teeth (Hutchinson teeth). None are seen in this patient.

**(Choice C)** Nonaccidental trauma or child abuse should be suspected in children with injuries inconsistent with their developmental stage or when the history is not consistent with the injury. Characteristic radiographic injuries include "bucket-handle" fractures, also known as **classic metaphyseal lesions**, and **rib fractures**.

**(Choice D)** Although anterior fontanels vary in size, costochondral joint enlargement is always pathologic and should prompt evaluation for rickets.

**(Choice F)** Skeletal dysplasias such as achondroplasia often present with bony abnormalities, including frontal bossing, limb shortening, and other dysmorphologies; none are present in this patient.

**Educational objective:**

Exclusive breastfeeding, increased skin pigmentation, and lack of sun exposure are risk



Infant on formula should receive vitamin D supplementation of 400 IU daily to prevent rickets.

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#### Educational objective:

Exclusive breastfeeding, increased skin pigmentation, and lack of sun exposure are risk factors for vitamin D deficiency rickets. Examination findings include craniotabes, rachitic rosary, and genu varum. X-ray findings include cupping and fraying of the metaphyses of the long bones.

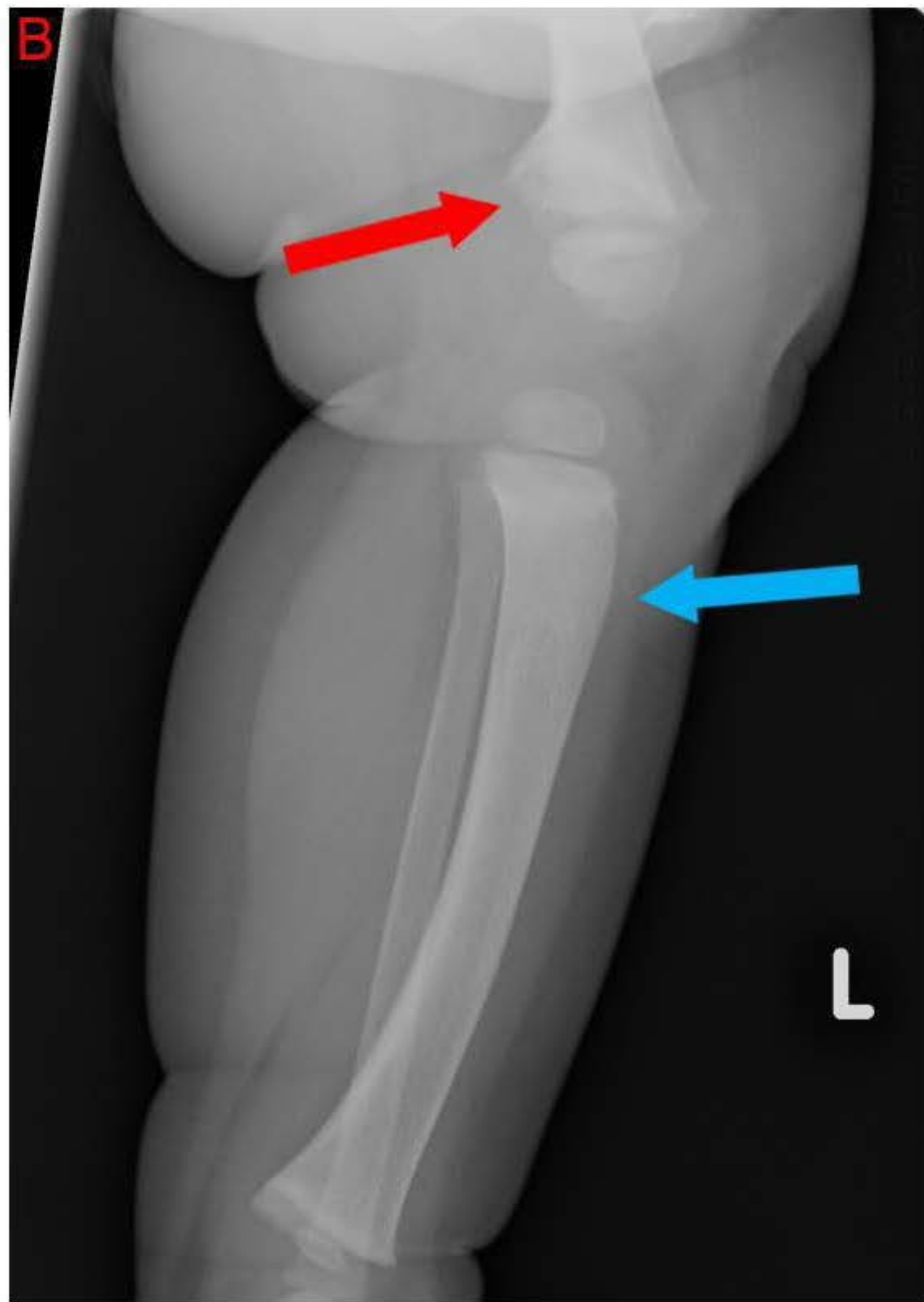
#### References:

1. **Resurrection of vitamin D deficiency and rickets.**
2. **Fractures in infants and toddlers with rickets.**
3. **Prevention of rickets and vitamin D deficiency in infants, children, and adolescents.**
4. **Vitamin D deficiency in children and its management: review of current knowledge and recommendations.**



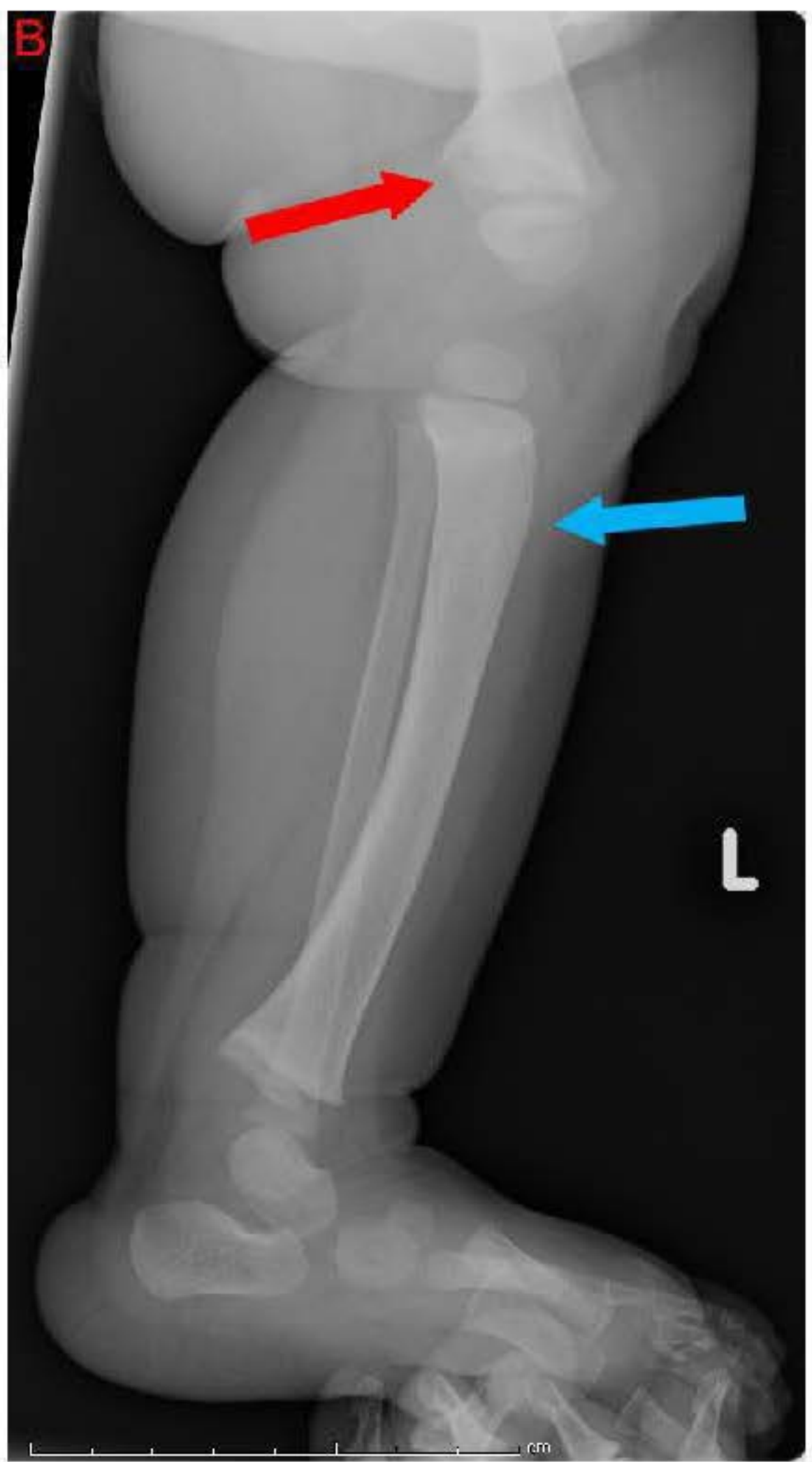
Media Exhibit

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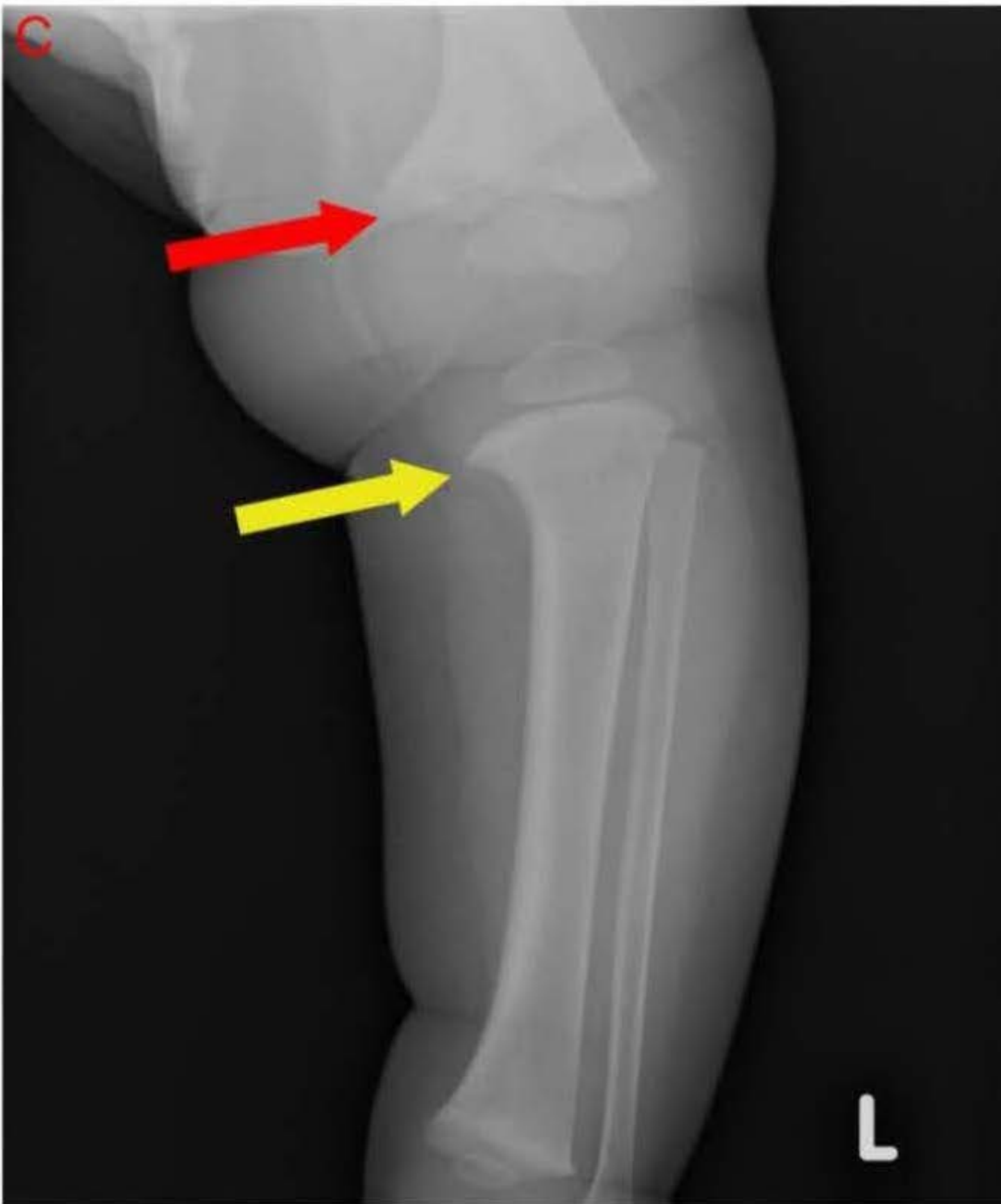
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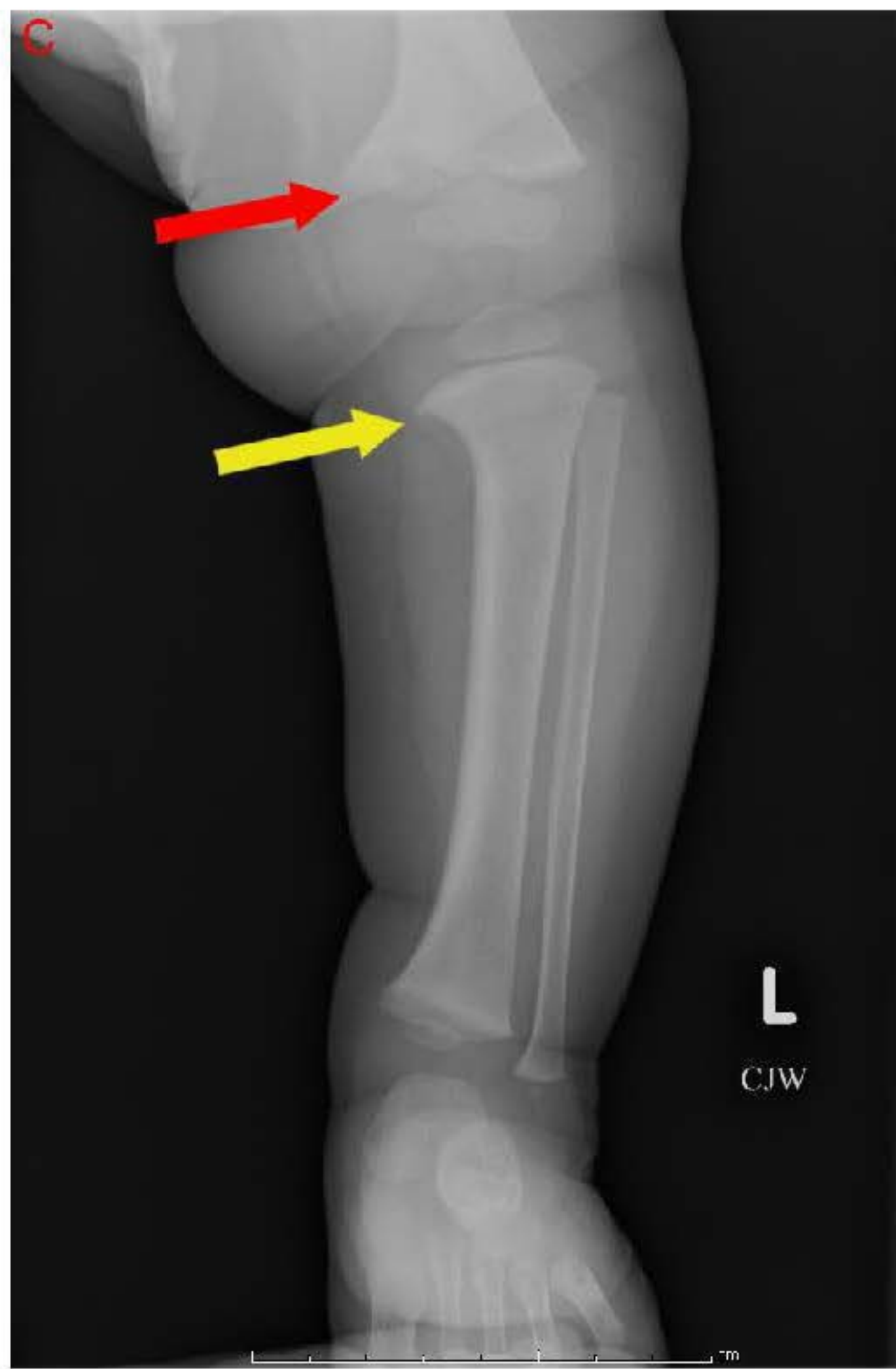
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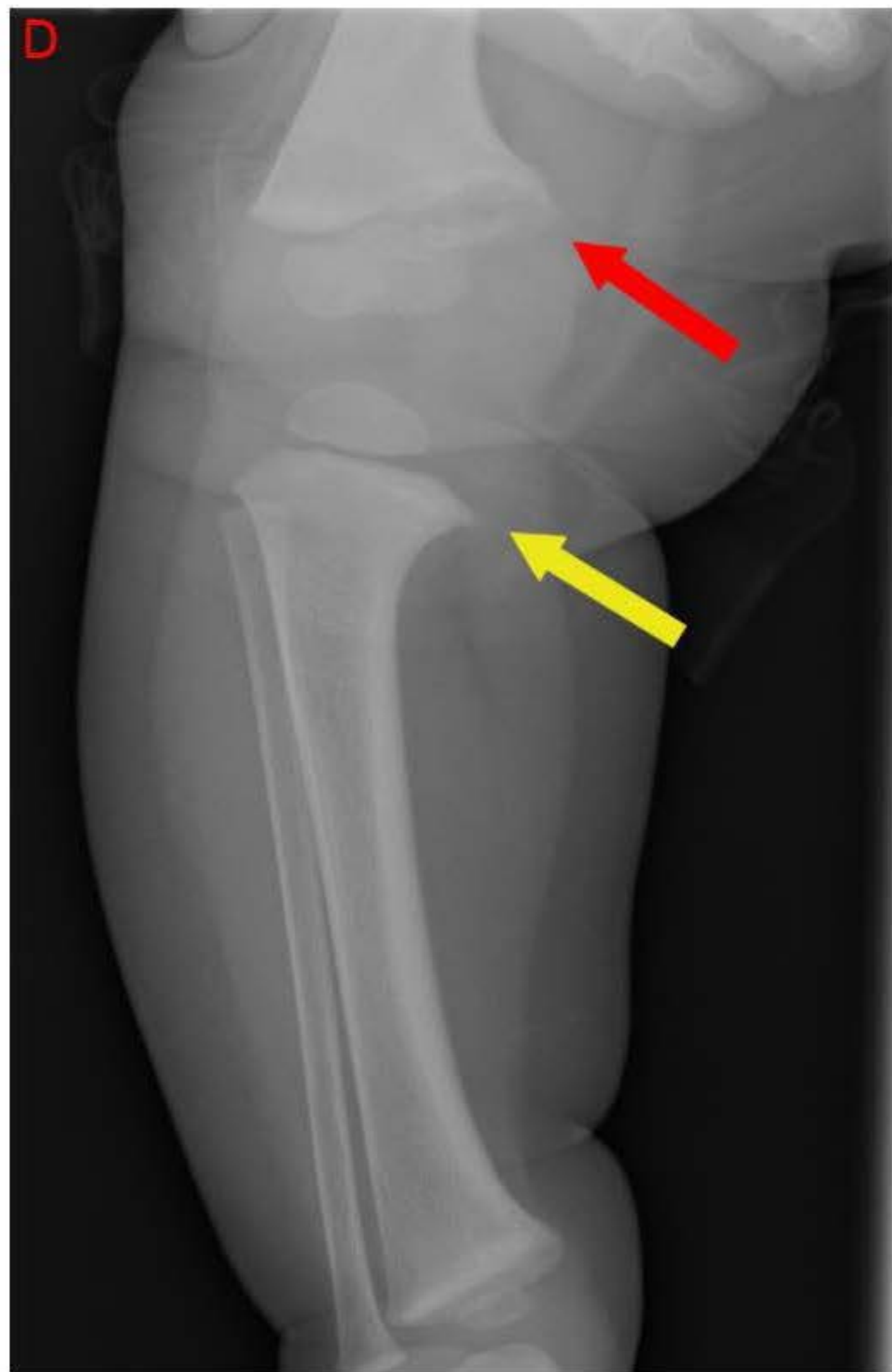
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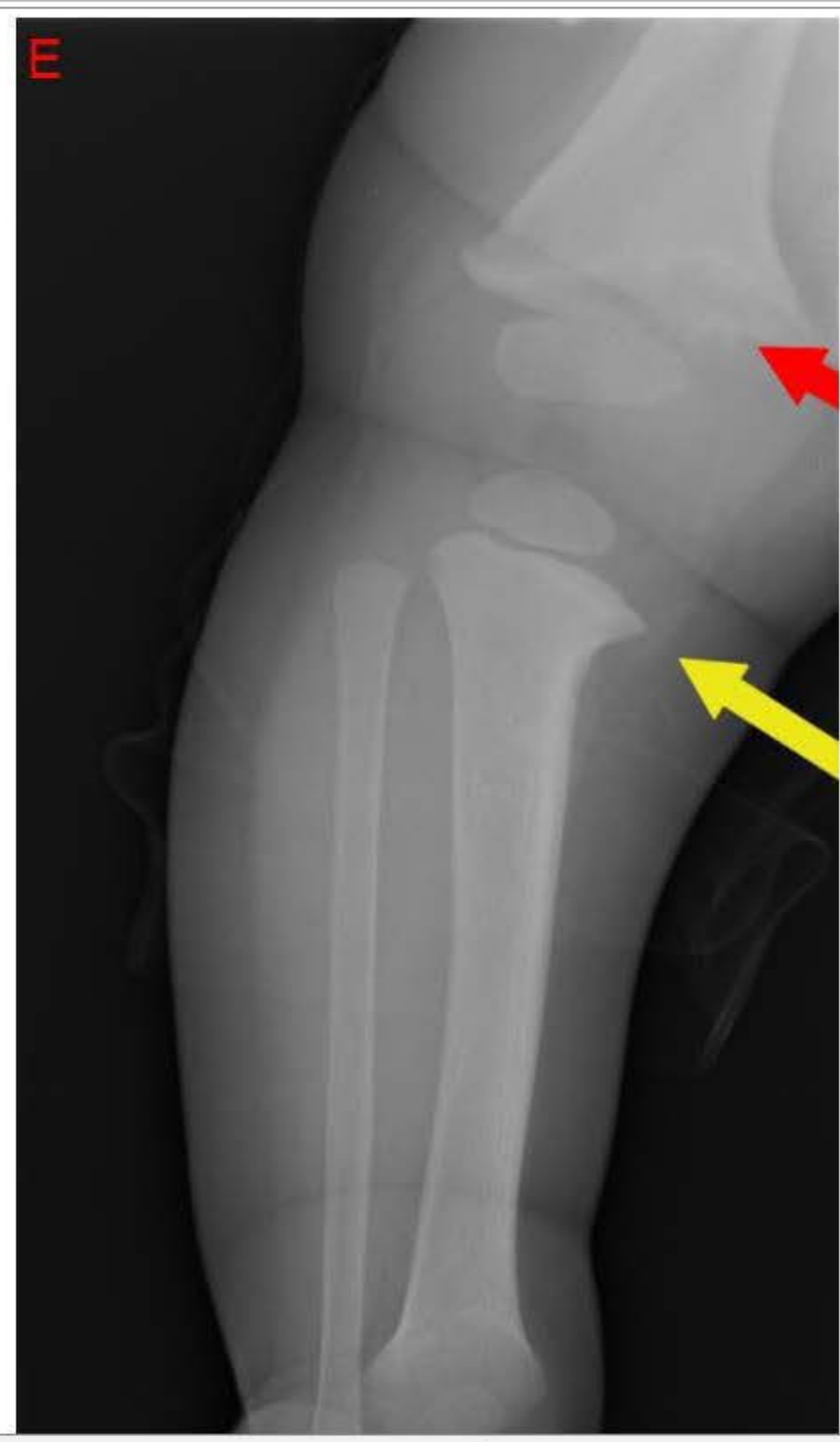
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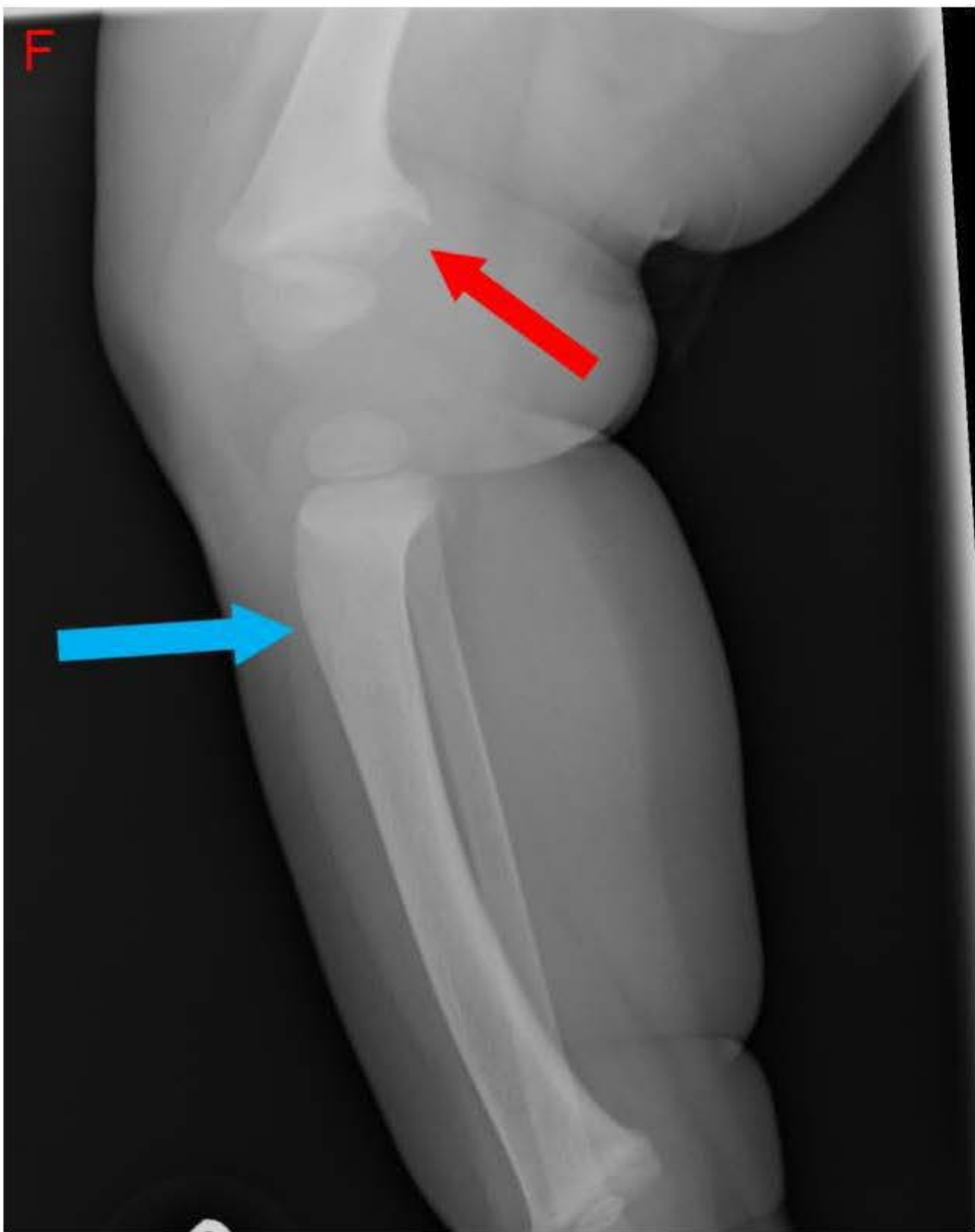
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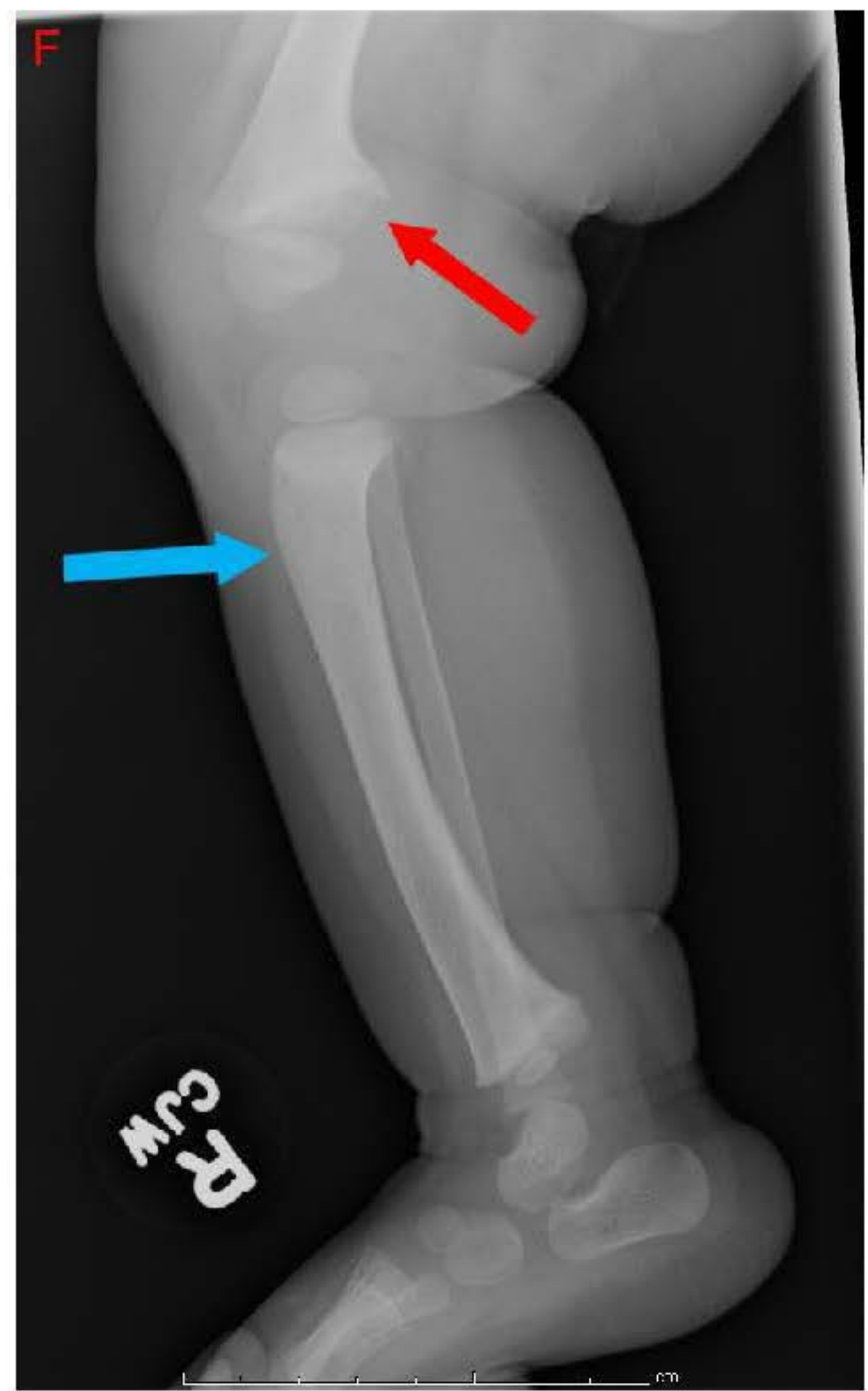
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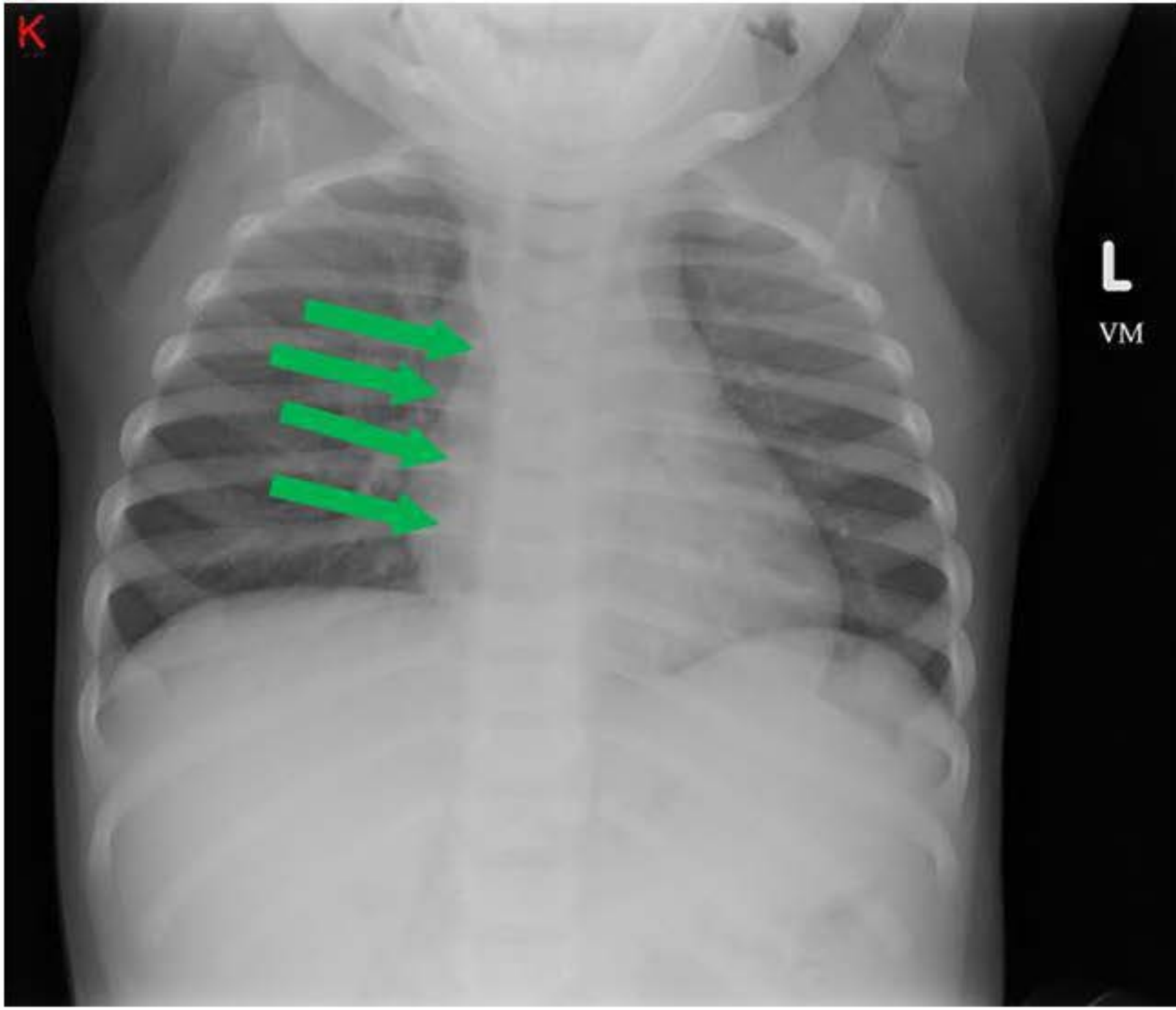
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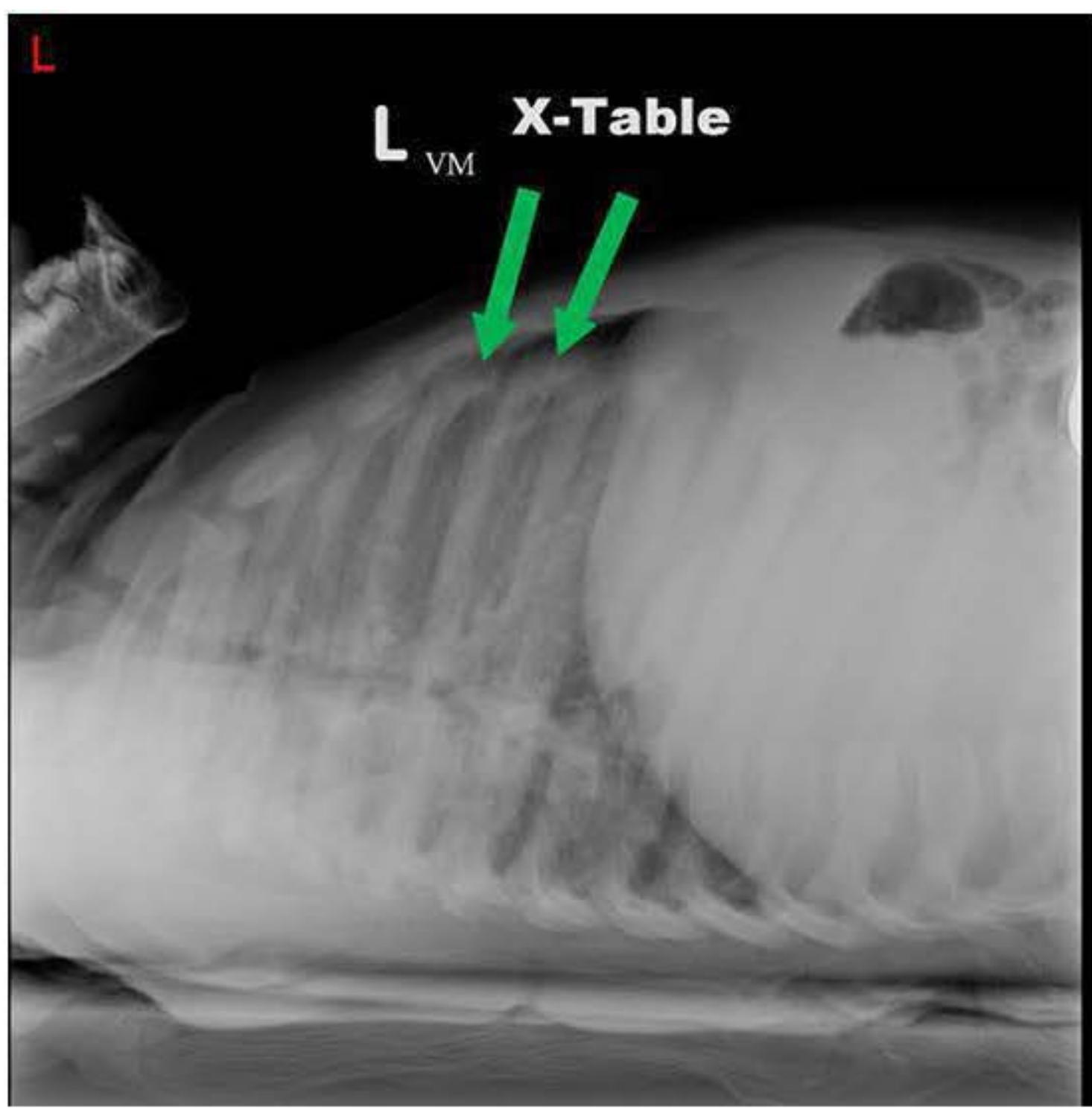
Rickets





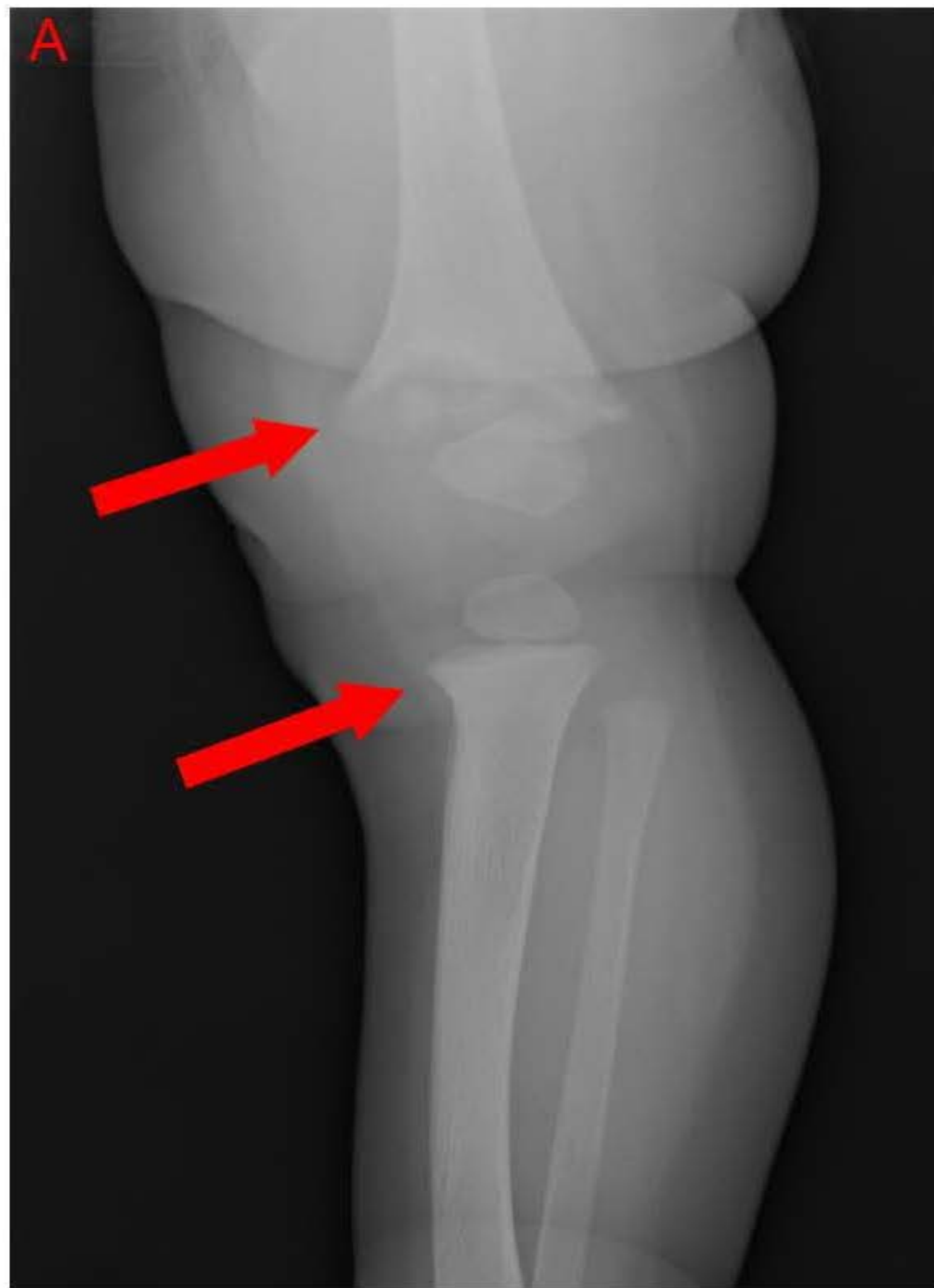
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metaphyseal lesions





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fractures secondary to non-accidental trauma

