

Interesting Images

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Interesting Images

Genu Recurvatum and Rocker Bottom Foot Co-Occurrence in a Unique Clinical Image of Edwards Syndrome

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Abstract: Edward syndrome, clinically known as trisomy 18 syndrome, is an autosomal chromosomal disorder characterized by an extra copy of chromosome 18. It belongs to the category of autosomal trisomy syndromes and is the second most common, with trisomy 21 being the only more frequent occurrence. In the context of this research, we present a case study focusing on a 3-month-old female infant admitted to the emergency department due to a complex array of symptoms. The primary clinical manifestations included a persistent cough, cold, and fever lasting for eight consecutive days. Comprehensive clinical evaluations, encompassing radiological and hematological investigations, confirmed the diagnosis of bronchiolitis along with viral bronchopneumonia. Chest X-rays revealed bilateral patchy opacities and diffuse haziness, primarily concentrated in the right hemithorax. Beyond respiratory issues, the infant displayed a range of distinctive physical attributes, including low-set ears, micrognathia, cranial anomalies, flattened nasal bridge, widely spaced nipples, overlapping fingers, abdominal distention, rocker-bottom feet, and recurring limb deformities known as recurvatum. Attempts to correct the limb deformity with a hip spica cast over an extended month-long period proved unsuccessful. Chronic villus sampling (CVS) ultimately confirmed the presence of Edward Syndrome (Trisomy 18), linked to the extra chromosome 18, leading to diverse physiological and developmental abnormalities. This case underscores the critical importance of timely and precise diagnosis, highlighting the challenges involved in addressing complex medical conditions marked by multifaceted clinical phenotypes.

Keywords: Edward Syndrome; x-ray; genu recurvatum; rocker bottom foot; camptodactyly



Figure 1. An X-ray examination of a 3-month-old female subject reveals several noteworthy observations.

- In the anterior-posterior view, there are discernible patchy opacities evident in both lungs, accompanied by a diffuse haziness within the right hemithorax (encircled in red).
- The examination also discloses bowel dilation with the presence of gas. The distinctive bubbly gas pattern observed in the right lower quadrant signifies the presence of an intramural gas (encircled in blue) [1].



Figure 2. Shows micrognathia (encircled in blue) and a distended abdomen (encircled in purple) [2].

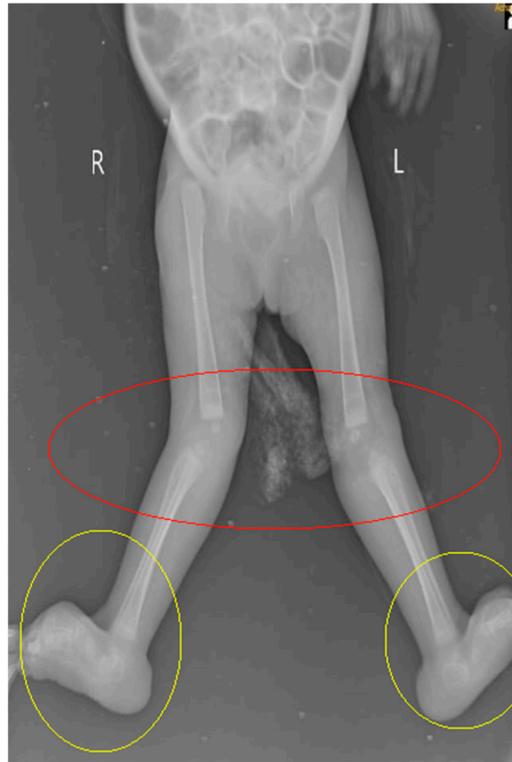


Figure 3. month-old female illustrates the presence of Genu recurvatum (encircled in red), in conjunction with bilateral rocker bottom foot deformity, (encircled in yellow).



Figure 4. Depicts the occurrence of Genu recurvatum (encircled in red), concomitant with bilateral rocker bottom foot deformity with the prominent calcaneus, (encircled in blue) [3].



Figure 5. Demonstrates the distinctive clenched hand posture featuring camptodactyly (encircled in yellow) [2].

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