

## IMAGES IN CLINICAL PRACTICE

### SECKEL'S SYNDROME- A RARE ENTITY

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#### KEYWORDS

Seckel's syndrome, rare autosomal recessive disorder, genetic heterogenicity

#### ARTICLE HISTORY

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An 8-year-old girl child, first by birth order, born of non-consanguineous marriage, presented to pediatric OPD with complaints of not gaining height and weight. There was no history suggestive of any chronic infection. She was a full term, normal delivered baby with a low birth weight of 2.1 kg. There was no history of any intrauterine infections or exposure to drugs/radiation in the antenatal period. She was developmentally normal for her age. Other sibling was normal. On examination, she had marked growth failure with microcephaly. Height (107 cm against an expected of 120 cm), weight (15 kg against an expected of 22 kg) and head circumference (48 cm against an expected of 53 cm) were below 3<sup>rd</sup> centile as per WHO growth chart. Systemic examination was normal.

The child had a characteristic hypoplastic facies with a prominent nose, small jaw(micrognathia) that was recessed back(retrognathia) and low-set ears (Figure

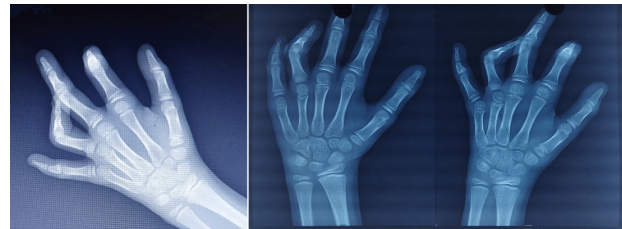
**Figure 1.** Facial Dysmorphism.



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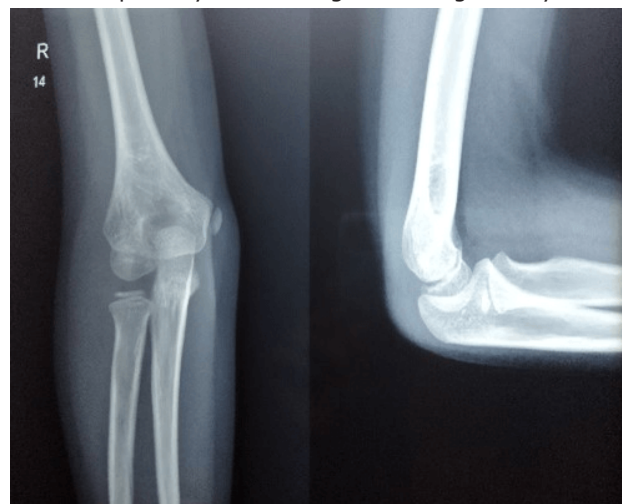
1). Bilateral 4th fingers were permanently fixed in **Figure 2.** Xray - AP and Oblique view both hands.



**Figure 3.** Clinodactyly of Fingers.



**Figure 4.** Elbow radiograph showing ossification of internal epicondyle indicating a bone-age of 5 years.



a bent position (clinodactyly) (Figure 3). Hypodontia (missing teeth), caries teeth and enamel hypoplasia were present. Skeletal survey revealed a bone age of 5 years (Figure 4) with absent epiphyses of middle phalanges (Figure 2).

#### *What is the diagnosis?*

Seckel's syndrome is a rare autosomal recessive disorder with craniofacial, oro-dental and skeletal defects, which causes marked growth retardation in children.<sup>2</sup> All these defects were present in this case. In spite of microcephaly, our child had moderate intelligence. Measures were taken to improve the nutrition and general health status of the child. Complete oral care, hygiene and orthopantomogram was advised and child is under regular follow-up.

Seckel syndrome is a rare condition of primordial dwarfism characterized by intrauterine and postnatal growth retardation, low birth weight, short stature, microcephaly, varying degrees of mental deficiency, typical facial appearance with beak-like protrusion of the mid face (bird-headed appearance), craniofacial dysmorphism, narrow face with malformed ears, micrognathia, receding forehead, large nose, prominent eyes, skeletal defects including clinodactyly, radial bone dislocation, significant dental alteration, caries tooth, defective hypoplastic enamel and other

physical deformities.<sup>1</sup> Abnormalities in cardiovascular, hematopoietic, endocrine, gastrointestinal, central nervous system are also seen in Seckel syndrome.<sup>4</sup> Approximately, 25% of patients have aplastic anemia or malignancies.<sup>3</sup>

It is a broad genetic heterogeneity syndrome comprising of 8 types-ATR mutation, RBBP8 mutation, 14q21-q22, CENPJ mutation, CEP152 mutation, CEP63 mutation, NIN mutation, ATRIP mutation.<sup>4</sup>

#### **Compliance with ethical standards**

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Conflict of Interest: None

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