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Case report

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Seckel syndrome: A rare cause of primordial dwarfism – A case report from Bangladesh

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Abstract

Seckel syndrome is an extremely rare genetic disorder characterized by intrauterine growth restriction, postnatal growth failure, microcephaly, proportional short stature and distinctive facial features. Skeletal abnormalities, dental abnormalities, and varying degrees of intellectual disability are commonly associated findings. We report the case of a 4-year-6-month-old girl born to non-consanguineous parents who presented with severe short stature and delayed developmental milestones. She had a history of intrauterine growth restriction and prematurity. Clinical examination revealed proportionate short stature, severe microcephaly, micrognathia, dental crowding, clinodactyly, and delayed bone age. Laboratory evaluation, including thyroid function tests and growth hormone stimulation testing, was unremarkable. Neuroimaging and echocardiography, as well as ultrasonography of the whole abdomen, showed no structural abnormalities. Based on clinical evaluation and radiological findings, our diagnosis is Seckel syndrome. Seckel syndrome should be suspected in children with severe proportionate growth failure, microcephaly, and developmental delay. Early assessment to identify associated systemic abnormalities and long-term multidisciplinary follow-up helps optimize growth, development, and quality of life. This case highlights the importance of careful clinical evaluation in diagnosing rare genetic growth disorders in resource-limited settings. *[J Assoc Clin Endocrinol Diabetol Bangladesh,* July 2026; 5 (2): e90172*]*

Keywords: Autosomal recessive; Clinodactyly; Microcephaly; Primordial dwarfism; Seckel syndrome.

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Introduction

Seckel syndrome (SS), first described by Seckel in 1960,[1] is an uncommon autosomal recessive disorder characterized by short stature, growth problems both before and after birth, distinct facial features such as micrognathia, dental anomalies, ear deformities, bird-like facial appearance, microcephaly, dislocation of the radial head, eleven pairs of ribs instead of normal 12, early fusion of cranial sutures, and intellectual disability.[2] Majewski et al. examined the literature and discovered that two-thirds of the documented cases did not fulfil all the diagnostic criteria established by Seckel.[3] Seckel Syndrome is caused primarily by a homozygous or a compound heterozygous mutation in a gene encoding ataxia-telangiectasia and RAD-3 related protein ATR, located on chromosome 3q22.1-q24.[4] This

gene is crucial for DNA damage repair. The prevalence of SS in Bangladesh remains unknown due to its rarity. To the best of our knowledge, no previously published case report from Bangladesh has been identified in indexed medical literature. Based on the most comprehensive published systematic review, at least 253 cases of SS had been reported worldwide as of May 2021.[5] As of 2026, the exact worldwide total is not officially known because new case reports continue to be published, and there is no central registry.

In addition to the above-mentioned anomalies, abnormalities have been found in the cardiovascular, endocrine, haematopoietic, and nervous systems.[6-8] Intellectual disability is not always as severe as might be expected in view of the very small brain. Here we describe a rare case of Seckel syndrome from Bangladesh, emphasizing its clinical presentation, diagnostic challenges, multidisciplinary management, and the importance of genetic confirmation and long-term follow-up.

Case report

A 4-year-6-month-old girl was admitted to the Department of Endocrinology & Metabolism, of Chittagong Medical College and Hospital, with concerns regarding poor growth compared to children of her age. She was the first child of non-consanguineous parents. According to her mother's statement, she had been small since birth. Antenatal history revealed intrauterine growth restriction detected at 8 months of gestation, followed by premature rupture of membranes. She was delivered preterm by cesarean section due to fetal distress associated with severe oligohydramnios. Her birth weight was 1900 g. The neonatal period was complicated by early-onset neonatal sepsis requiring hospitalization for 20 days. Developmental milestones

Table-I: Physical examination findings of the patient

Parameter	Findings	Interpretation / Reference
General Status	Alert and cooperative	Clinically stable **[Figure-1a]**
Height	81 cm	<-3 SD; severe stunting) [**Figure-1b**]
Weight	9.5 kg	<-3 SD; severely underweight **[Figure-1c**]
Head circumference	42 cm	<-3 SD; severe microcephaly [**Figure-1d]**
Arm Span	73 cm	Measured length
Upper segment (US)	41 cm	Used for ratio calculation
Lower segment (LS)	40 cm	Used for ratio calculation
US: LS Ratio	1.025	Proportionate short stature
Dental Exam	Crowding of teeth	Malocclusion [**Figure-2a and 2b**]
Extremities	Clinodactyly	Curve 5th finger [**Figure-2c and 2d**]
Craniofacial	Micrognathia	Mandibular hypoplasia [**Figure-2e and 2f**]
Intellectual Disability	Moderate*	

*by bedside IQ test

Table-II: Laboratory and imaging findings of the patient

Investigation	Result	Interpretation / Reference
Hemoglobin	10.8 g/dL	Below normal range
Total WBC Count	10.58×109/L	Within normal range
Platelet Count	275×109/L	Within normal range
Serum TSH	1.32 µIU/mL	Within normal range
Free T4 (FT4)	1.05 ng/mL	Within normal range
Growth hormone (GH) (1-hr after tab Levodopa)	8.8 ng/mL	No deficiency
Chest X-ray	No Abnormality	-
Radiological bone age	2 years	Delayed skeletal maturation (**Figure-3**)
Abdominal USG	All organs are of normal size according to age. Both ovaries are present.	-
Echocardiography	Normal morphology; LVEF 66%; No shunts.	-
MRI of the brain	Normal brain parenchyma, intact corpus allosum.	**Figure-4**

were significantly delayed. Neck holding was achieved at 9 months, independent walking at 24 months, and speech development improved after therapy sessions at a child development center. There was no family history of similar conditions or other congenital anomalies. Physical examination revealed severe short stature, microcephaly and characteristic craniofacial dysmorphism suggestive of primordial dwarfism [**Table-I**, **Figure 1 and 2**]. Systemic examination, including ocular examination, was unremarkable. Laboratory and imaging findings are shown in **Table-II**, **Figure-3 and 4**. Routine laboratory investigation, thyroid function and growth hormone stimulation testing were unremarkable, while bone age was significantly

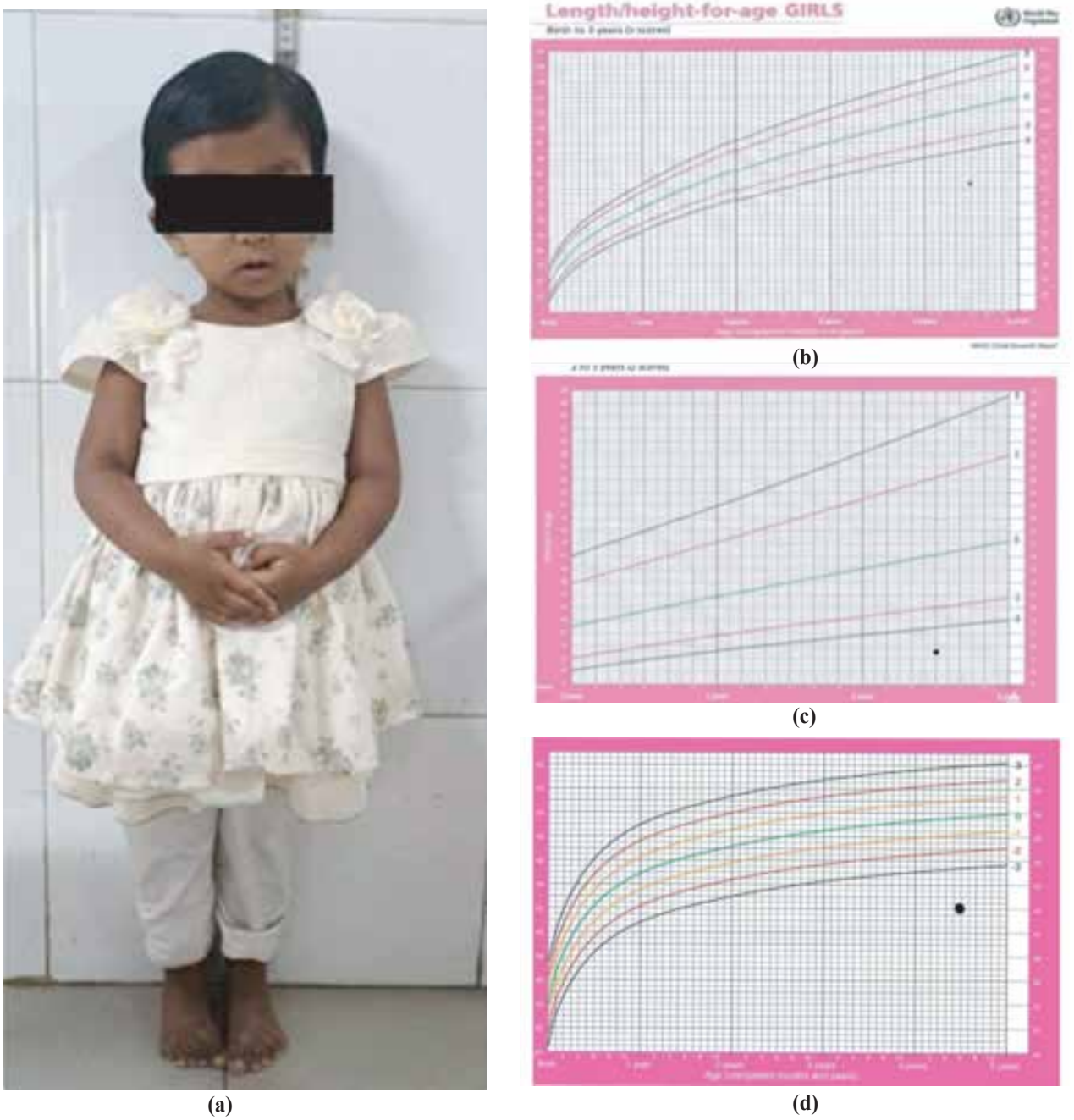

Figure-1: (a) Physical appearance of the patient, (b) height-for-age chart showing severe stunting, (c) weight-for-age chart showing severe underweight, (d) head circumference-for-age z-score chart showing severe microcephaly

Figure-2: (a) and (b) showing crowding of teeth; (c) and (d) showing clinodactyly - curved 5th finger; (e) and (f) showing micrognathia (mandibular hypoplasia)

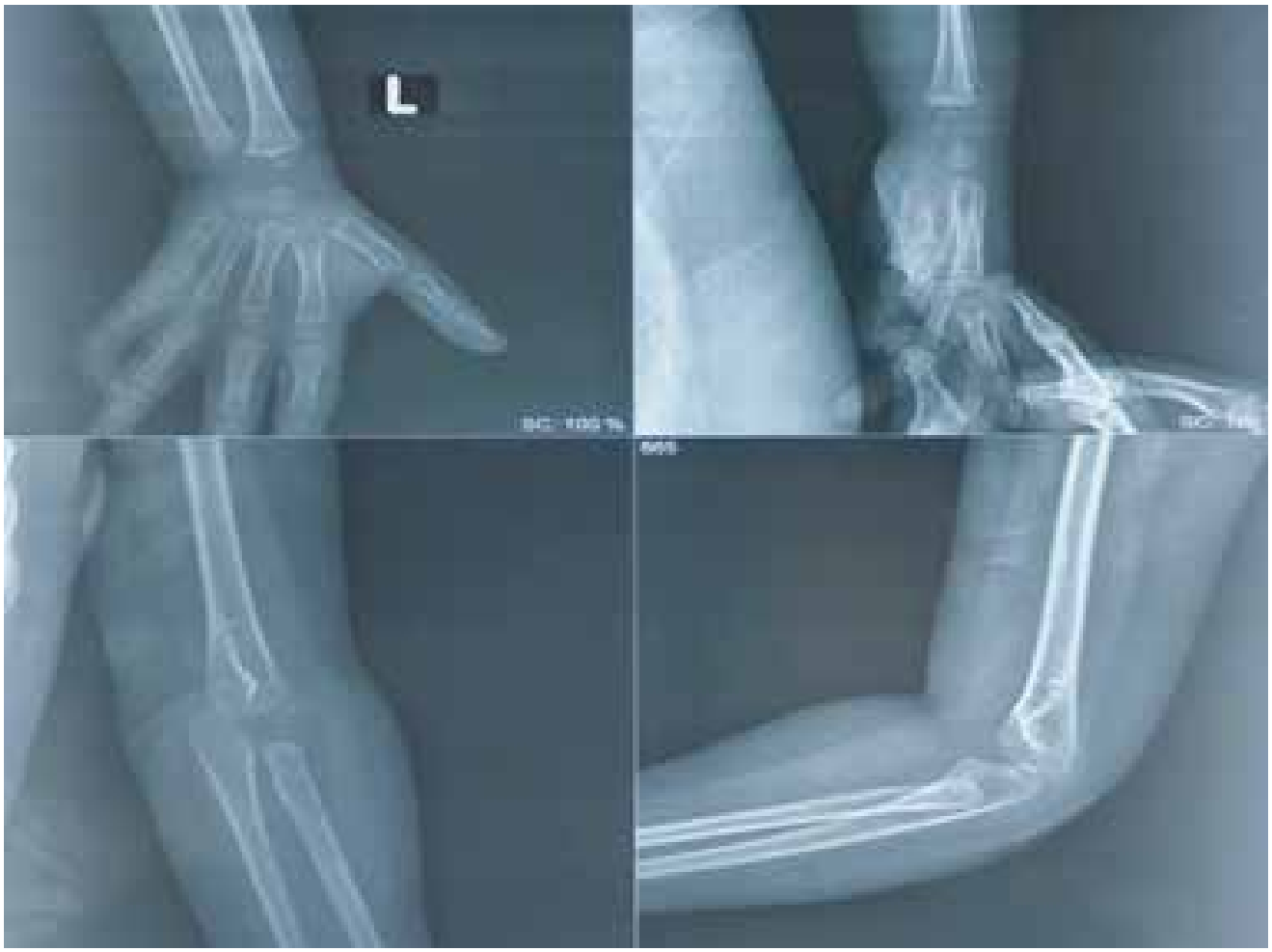

Figure-3: X-ray of left wrist joint and elbow showing skeletal maturation of 2 years at a chronological age of 4 years 6 months.

delayed. Based on clinical manifestation and exclusion of other diagnostic possibilities, a clinical diagnosis of Seckel syndrome was made. Genetic testing could not be performed due to financial constraints and limited availability. Therefore, molecular subtyping could not be determined.

Discussion

A wide range of clinical features can be present in Seckel syndrome. There is no universally accepted formal diagnostic criterion for SS. Diagnosis of SS is based on a combination of characteristic clinical findings, radiological features, and, when possible, molecular genetic confirmation. Essential clinical features include severe IUGR, postnatal proportionate short stature, severe microcephaly, characteristic craniofacial appearance (bird-like face, beaked nose, micrognathia, dental anomaly, receding forehead, large-appearing eyes, clinodactyly), osteodysblastic changes, developmental delay or intellectual disability. The present case demonstrated several classical features of SS, including severe proportionate short stature (<-3 SD), microcephaly (<-3 SD), delayed bone age, micrognathia, dental crowding, and clinodactyly. The history of IUGR and prematurity, birth weight 1900 g, was below the 50th percentile for 34 weeks of gestation, further supporting the diagnosis. Seckel's patient resembles that of a bird, therefore historically it was named 'Bird headed dwarfism'. Although the patient did not exhibit the classical 'bird-like' facial appearance or prominent beaked nose described in earlier literature, phenotypic heterogeneity is well documented in SS. About seven clinical features and radiological findings are similar to the characteristics of SS. Not all patients fulfil the complete classical diagnostic features, which may lead to underdiagnosis or misclassification. Bone age in our patient was significantly delayed (2 years at a chronological age of 4 years 6-months), which is

consistent with previous documented cases. Levodopa-induced growth hormone stimulation testing revealed a peak value of 8.8 ng/mL, which did not meet standard diagnostic criteria for growth hormone deficiency,[9] as most authorities recommended that the threshold be revised to 7 ng/mL, depending on the assay.[10] However, under endocrinological evaluation to exclude GH deficiency, this test was performed. Endocrine abnormalities have been described in some patients with SS, but short stature is primarily attributed to the underlying genetic defect rather than isolated hormonal deficiency.

Differential diagnoses of primordial dwarfism include Majewski Osteodysplastic Primordial Dwarfism (MOPD) type II, which is usually associated with more severe skeletal abnormalities and an increased risk of cerebrovascular complications.[11] Meier-Gorlin syndrome is characterized by small ears and the absence of hypoplastic patellae.[12] Silver-Russell syndrome - typically presents with disproportionate short stature and relative macrocephaly rather than microcephaly.[13] Cockayne syndrome - associated with photosensitivity, retinal degeneration, and progressive neurological decline.[14] Dubowitz syndrome features microcephaly, facial dysmorphism (often with eczema), and short stature.[15] Fanconi anemia is characterized by short stature, low birth weight, and developmental delays, café-au-lait spots (light brown patches) or hypopigmentation, hypothyroidism, increased risk of malignancy, hearing loss, and strabismus.[16] Bloom syndrome is associated with pre and postnatal proportional growth retardation, microcephaly, keel-shaped, malar hypoplasia, nasal prominence, and ear protuberance.[17] The absence of disproportionate skeletal dysplasia, major brain malformations, or significant hematological abnormalities supported the diagnosis of Seckel syndrome over other primordial dwarfism syndromes. Although genetic confirmation is considered the gold standard for diagnosis, especially through whole-exome sequencing, financial and technical constraints prevented molecular testing in this case. In many low-resource settings, clinical diagnosis remains essential.

Few cases have been reported from South Asia. In our case, the patient presented with severe short stature, severe microcephaly, micrognathia, clinodactyly, dental crowding, delayed bone age, moderate intellectual disability, no bird-like face, no pigmentary changes, and no history of consanguinity in parents and genome sequencing cannot be done at that time. A similar case from Pakistan describes a 10-year-old girl, suspected SS patient from consanguineous marriage, who presented with severe microcephaly, severe intellectual disability, short stature, absence of speech, pointed nose, narrow face and bilateral cataract in two siblings, where the whole genome sequence discovered a missense variant in exon 2, which is pathogenic to SS patients.[18] Another case from India, a 3-year-old female, presented with microcephaly, a triangular-shaped face, large eyes, café-au-lait spots, micrognathia, and crowding of mandibular teeth. A similar case from India, where a 10-year-old female presented with a history of severe IUGR, delayed developmental milestones, low IQ, microcephaly, and dental caries, was diagnosed as a case of SS.[19] However, the absence of cardiovascular, cerebrovascular, and haematological abnormalities in our case differentiates it from some reported variants associated with bone marrow involvement. This case emphasizes the importance of detailed anthropometric assessment and recognition of characteristic dysmorphic features in diagnosing rare genetic growth disorders, particularly in resource-limited countries. Recognition of rare genetic causes of short stature is crucial in endocrine practice to avoid unnecessary hormonal therapies and to ensure appropriate genetic counselling for affected families.

Molecular testing (preferably whole-exome sequencing [WES]) to identify pathogenic variants (e.g., ATR, CEP152, CENPJ, RBBP8) was advised, although it could not be done due to financial constraints. A dental consultation was conducted, and the dentist advised a pulpectomy and a filling. The patient was advised to visit the dental department on the due date. The patient was referred to a child development center for speech therapy. Evaluation for puberty & fertility was not performed at that time, as LH and FSH might be within the normal prepubertal range. The patient was advised to follow up at 13 years of age if normal pubertal changes do not occur. Regular monitoring of growth, development, blood counts, and endocrine function, ophthalmologic care as needed. Growth hormone therapy remains controversial, as evidence supporting its long-term benefit in genetically confirmed Seckel syndrome is limited.[20] Patient's parents were counselled about the need for physiotherapy, occupational therapy, and educational support in future. Genetic counselling (autosomal recessive; 25% recurrence risk), psychological support, early intervention, and multidisciplinary follow-up were provided.

Puberty is possible in SS patients and may be normal,

delayed, or precocious depending on the individual. Fertility remains unknown, as there are no well-documented published reports of successful reproduction in individuals with confirmed SS up to the current literature reviewed.

Data on life expectancy in Seckel syndrome are unavailable, but case reports in the scientific literature have documented survival beyond age 50.

The main limitation of this case is the lack of confirmatory genetic testing to determine the exact molecular basis of Seckel syndrome. Although the clinical, radiological, and hormonal characteristics provide strong evidence supporting the diagnosis, the absence of genotypic validation hampers our ability to identify the specific gene mutations underlying the condition. Also, to diagnose isolated GH deficiency or partial GH deficiency, at least 2 stimulated tests are required. Other than this, low IGF-1 is more reliable for finding GH deficiency, which could not be performed due to financial constraints. Formal IQ assessment was also not performed.

Conclusions

Seckel syndrome is a rare primordial dwarfism disorder that may present with variable clinical features. In this case, severe proportionate short stature, microcephaly, and characteristic craniofacial anomalies should prompt consideration of this diagnosis. In settings where genetic testing is unavailable, careful clinical evaluation remains critical. Early multidisciplinary intervention and family counselling are essential to optimise developmental outcomes and improve quality of life.

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Disclosure

The authors have no conflicts of interest to disclose.

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Data Availability

Any inquiries regarding supporting data availability of this study should be directed to the corresponding author and are available from the corresponding author on reasonable request.

Consent to Participate

Written informed consent was obtained from the patient's legal guardian for publication of this case report and accompanying clinical images.

Disclosure of generative AI and AI-assisted technologies

During the preparation of this work, we used the AI Tool ChatGPT 5.5 to assist with language editing, improving grammar, refining sentence structure, formatting the manuscript, word counting, comparing with clinical findings and for reference formatting. After using this tool, we thoroughly reviewed, edited, and modified the content as needed. We take full responsibility for the accuracy, originality, and integrity of the final work.

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