

## Seckel Syndrome: In a Two and a Half Months Old Male Presenting at Tertiary Care Hospital in Karachi.

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### Abstract

Seckel syndrome is a rare disease, with autosomal recessive mode of inheritance. Clinically manifests as intrauterine and postnatal growth retardation along with microcephaly, receded forehead, micrognathia, hypoplastic ears and mental retardation. Here we present a case of two and a half months old boy with four days history of loose stools and fever with facial dysmorphism and growth retardation of prenatal onset. This case is notable as detailed physical examination in patients leads to early diagnosis of the disease.

**Keywords:** Seckel syndrome, bird beak appearance, bird beak dwarfism,

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### Introduction

Seckel was the first to describe Seckel syndrome in 1960<sup>1</sup>. A rare autosomal recessive disease, which has no sex predilection. An incidence of 1:10,000 live born children has been reported<sup>2</sup>. Rudolf Virchow named "bird-headed dwarf" to proportionate dwarfism with low birth weight, mental retardation, beak like nose, and micrognathia<sup>3</sup>.

The craniofacial features of Seckel syndrome differentiates it from other syndromes of growth deficiency with microcephaly, such as Dubowitz syndrome, fetal alcohol syndrome, trisomy<sup>18</sup> syndrome, DeLange syndrome, and Fanconi syndrome<sup>4</sup>.

Craniofacial appearance of Seckel syndrome include "bird-headed appearance" with receding

forehead, large eyes, pointed nose, narrow face, receding lower jaw, and micrognathia. Dental abnormalities were also reported which includes oligodontia, hypoplastic enamel, delayed eruption of teeth and demineralization of dentin<sup>5</sup>. Cleft palate has been reported in Seckel syndrome cases<sup>6</sup>.

Majority of the cases of severe intrauterine growth retardation and microcephaly are detectable by serial ultrasound scan from 16 to 20 weeks of pregnancy<sup>7</sup>.

We present this rare case with characteristic features supported by relevant literature search and possible etiology with a differential diagnosis.

### Case report

We report a case of a two and a half months old male who presented to emergency department of Abbasi Shaheed Hospital on 18-December-2015, with chief complaint of loose stools with fever for 4 days. Stools were watery, which gradually increased accompanied with fever due to which his oral intake was decreased.

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His past medical history revealed that he was admitted twice due to similar complaints in public sector hospitals of Karachi from 1st to 4th of December- 2015 and then from 11th to 14th of December-2015. No history of blood transfusion at anytime.

Birth history revealed that the mother had history of loose stools in the 4th month of gestation due to which she was admitted in hospital for two days. Antenatal scans showed IUGR. Patient was born at 36 weeks via emergency lower segment c-section due to premature rupture of membranes for 1 day and bleeding per vagina for few hours. He was admitted in Neonatal Intensive Care Unit (NICU) for twelve days due to sepsis. Weight at birth was 1.8 kg. with no history of delayed cry or cyanosis. Vaccination was up to date. BCG scar was present. Child was exclusively breastfed for one month then lactose free formula feed was started after admission in public sector hospital due to acidic reaction of stool on detailed stool report. Patient is the first issue of non consanguinous marriage. Paternal aunt had given birth to a daughter with similar features. Socioeconomic status was low.

On general physical examination patient was ill looking and febrile with microcephaly, receded forehead, bird beak nose, low set ears with hypoplastic ear lobule and micrognathia Fig 1,2,3. Child displayed signs of severe dehydration. Anthropometric measurements included weight 2.1 kg, length 43cm, and head circumference 29cm. His heart rate was 153/min, respiratory rate 48/min and temperature 101°F. Oxygen saturation was 98% on room air. Complete blood count showed microcytic, hypochromic anaemia. Haemoglobin was 7.1g/dl. Stool detailed report revealed acidic reaction. Ultrasound abdomen and of kidney, ureters and bladder was normal. Echo was also normal. Based on history and examination child provisional diagnosis of Seckel syndrome with acute gastroenteritis was made.

Treatment prescribed was injection cefotaxime (100mg/kg/day) and injection amikacin (15mg/kg/

dose x BD). This combination of drugs provided maximum synergistic effect and was also suitable for this age group considering maturity of liver. Child showed improvement on follow up visit scheduled one week after discharge. He had gained 0.5 kg weight as well. Parents were counselled regarding chances of recurrence of the disease in next pregnancies.

## Discussion

Mann and Russell reported in 1959 and this disease was thoroughly studied by Seckel in 1960<sup>8</sup>. There is a recently set criteria for diagnosis of Seckel syndrome. There are number of abnormalities which includes growth deficiency; birth weight at term is 1543gms (1000-2005gm), mean postnatal growth deficiency is  $-7.1 \text{ SD} \pm 2.08$  and delayed bone age<sup>9</sup>. In the present case, IUGR was detected on anenatal scans, delivered at 36 weeks via emergency C-section. Birth weight was 1.8kg (less than 5th centile) at examination, length was 43cm (less than 5th centile) and head circumference was 29cm (less than 5th centile). He was the only child of unrelated parents. Height of mother was 143cm and height of father was 151cm, both weighing 54kg.

Cental nervous system abnormalities include mental retardation with low IQ. Severe intrauterine growth retardation, postnatal dwarfism, microcephaly, receded forehead, narrow bird like face with beak like pointed nose, micrognathia, mental retardation and short stature<sup>12</sup>. In the present case, characteristic craniofacial abnormalities were observed. Our patient's anthropometric measurements plotted on CDC growth charts were all below 5th centile. Patient also had low set ears with slanted palpebral fissures.

Other features of Seckel syndrome included premature closure of cranial sutures secondary to diminished brain growth, slanted palpebral fissures, dysplastic ears, clinodactyly of the fifth fingers, cryptorchidism, clitoridomegaly, hirsutism, agenesis of corpus callosum, pachygyria, retarded bone age, frequent hip dysplasia, dislocation of the head of ra-



**Fig 1,2,3.** Baby with Seckel syndrome showing: microcephaly, receded forehead, bird beak nose, slanted palpebral fissures, low set ears with hypoplastic ear lobule, micrognathia and retractile testes.

dius<sup>13</sup>. The patient had, slanted palpebral fissures, Iretractile testes.

The autosomal recessive inheritance pattern can be due to chromosomal instability or chromosomal breakage. Seckel syndrome is caused by heterogeneity mapped on 2q33.3-34<sup>14</sup>, and the other mapped on 18p11.31-q11.2<sup>15</sup>. In the present case chromosomal mapping was not done. Abnormalities in haematopoietic system can also be found, and have been reported in 15% of the cases<sup>15</sup>. The complete blood count of the patient also showed microcytic, hypochromic anaemia.

History of intrauterine growth retardation, low birth weight, general physical examination revealing microcephaly, receded forehead, bird beak appearance, low set ears, micrognathia, paternal aunt's daughter with similar malformations and complete blood count are all in favour of Seckel syndrome. Therefore the diagnosis of Seckel syndrome was conferred.

## Conclusion

Seckel syndrome is a rare disorder, therefore patients with craniofacial malformation, intrauterine growth retardation and short stature should be examined thoroughly to ensure an early diagnosis.

## Conflict of interest

Authors have no conflict of interests and no grant/ funding from any organization.

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