

Cranial MRI findings in a case with Seckel syndrome

Seckel sendromlu bir olguda kraniyal MRG bulguları

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SUMMARY

Seckel's syndrome (SS) is a rare, heterogeneous form of primordial dwarfism. The presence of mental retardation and neurologic signs is one of the peculiar features of this syndrome. We herein present the cranial MR findings in a child patient with Seckel's syndrome.

Keywords: Brain, Dwarfism, Magnetic resonance imaging, Seckel syndrome

ÖZET

Seckel sendromu (SS) nadir, heterojen bir primordiyal cücelik türüdür. Zeka geriliği ve nörolojik belirtilerin varlığı bu sendromun kendine has özelliklerinden biridir. Biz burada Seckel sendromlu bir çocuk hastada kraniyal MR bulgularını sunuyoruz.

Anahtar sözcükler: Beyin, Cücelik, Manyetik rezonans görüntüleme, Seckel sendromu

INTRODUCTION

Seckel syndrome (SS) is a rare cause of primordial autosomal recessive dwarfism. It was first defined by Seckel in 1960. This rare condition is characterized by low birth weight, short but proportional stature, microcephaly, mental retardation, and facial anomalies¹. Hematological, renal, cardiac, and central nervous system anomalies may also accompany the clinical picture^{1,2}. In this report, we discussed the co-existing magnetic resonance imaging (MRI) findings in the light of the available literature data in a rare case of SS.

CASE REPORT

A 14-year-old male patient with SS referred for a preliminary diagnosis of mental retardation was examined with cranial radiography and cranial MRI. The patient's prenatal and postnatal history were not revealing. There was a second-degree consanguineous marriage between the parents. The patient had a height of 137 cm and a head circumference of 51 cm. The bulges of the cranial sutures could be palpated. Additionally, he had a receding forehead, prominent eyes with a normal interocular distance, a prominent, beak-like nose, retrognathia, and low-set ears. He had a highly-arched, intact palate. He had mental retardation. He had deformed teeth and a maloccluded chin. The examination of other systems was normal. He

also had normal laboratory tests and a normal chromosome analysis.

There was no solid organ pathology on abdominal ultrasonography. Direct films were obtained to define skeletal system findings. Cranial roentgenograms revealed microcephalia, beak-like nose, retrognathia, frontal bone hypoplasia, and prominent convolutional signs. A cranial MRI to detect potential concurrent central nervous system

(CNS) abnormalities showed a prominent, beak-like nose and a hypoplastic mandible. Posterior fossa structures were normal. Corpus callosum was hypoplastic. The posterior horns of the lateral ventricles were minimally dilated but the outer subarachnoid spaces and white matter appeared normal. There was a lissencephalic appearance in both cerebral hemispheres, particularly in the frontotemporal regions (**Figure 1a-c**).

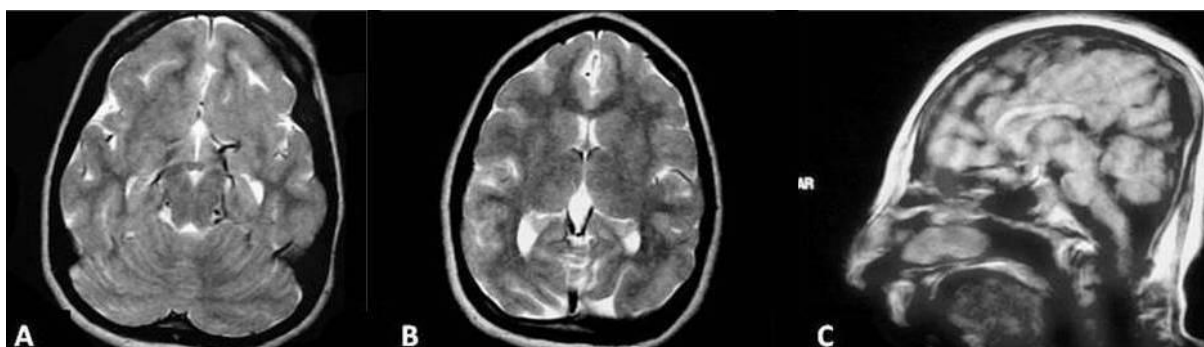


Figure 1. (A, B) Axial T2 weighted MR sections show lissencephaly in both frontotemporal regions and mild dilatation of the posterior horns of the lateral ventricle. (C) The sagittal T2 weighted MRI section shows a prominent beak-like nose, mandibular hypoplasia, and corpus callosum hypoplasia.

DISCUSSION

SS is a rare disorder with autosomal recessive genetic transmission that is characterized by low birth weight, short stature, microcephaly, mental retardation, and atypical face appearance (beak-like nose, small forehead, micrognathia). There is no gender predilection and the incidence is 1/10,000. More than 60 SS cases have been described in the literature. In SS several anomalies related to central nervous system (CNS) have been described¹⁻⁷. Arnold et al³ reported 3 cases with severe hydrocephalus. Howanietz et al⁴ detected enlarged cerebral ventricles in a 9-year-old patient with SS. Cherian⁵ detected marked dilatation in the right lateral ventricle in a patient with SS. Shanske et al¹ demonstrated corpus callosum agenesis, cerebellar vermis agenesis, cerebral cortical dysplasia, and medial dorsal cyst with computed tomography in a patient with SS. Krishna et al⁶ detected a small cerebrum, a relatively large cerebellum, and a midline arachnoid cyst in a patient with SS. Sugio et al⁷ reported hypoplastic cerebrum and cerebellum in a 2-month-old male infant. The similarities in these publications suggest a common element of neuronal migration anomaly. Furthermore, Vincenzo et al⁸ described multiple intracranial aneurysms in a 17-year-old girl that presented

with intracranial hemorrhage. Can et al⁹ reported the presence of lissencephaly and corpus callosum hypoplasia on cranial MRI in a 1-month-old newborn, as in our case.

In conclusion, we are of the opinion that in malformative syndromes such as SS, cranial MRI should be definitely performed to determine the correlations between the widespreadness and types of accompanying lesions, and to detect potential CNS anomalies.

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