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Research Article

Psychomotor Retardation, Low Set Ears, Retrognathia, Facial Dysmorphism and Schizencephaly: A New Dysmorphic Syndrome

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Abstract

Background: Schizencephaly is a rare primary congenital brain defect of heterogeneous nature resulting from abnormal morphogenesis with a very early disruption of the grey matter migration during embryogenesis. Braga et al (2018) reviewed 156 articles including 734 patients with schizencephaly, and none of them had facial dysmorphism, low set ears or micrognathia
Patients and methods: A dysmorphic male infant who was referred to the neuropsychiatric consultation clinic of the Children Teaching hospital of Baghdad medical city was studied.
Results: Four month male infant presented with psychomotor retardation with no interaction with the mother and no recognition of her face. He had low set ears, retrognathia, and facial dysmorphism with narrow and upslanting palpebral fissures and thin upper lips. Family history was negative for a similar condition. Brain CT-scan showed open limb bilateral schizencephaly and karyotype showed normal finding.
Conclusion: A new dysmorphic syndrome associated with schizencephaly, facial dysmorphism, low set ears and micrognathia is reported.

Keywords: new dysmorphic syndrome; schizencephaly; facial dysmorphism; low set ears; micrognathia

Introduction

Schizencephaly is a rare primary congenital brain defect of heterogeneous nature resulting from abnormal morphogenesis with a very early disruption of the grey matter migration during embryogenesis. It is attributed to failure of formation of the cerebral mantle in the regions of the cerebral fissures that can be the result of genetic factors, intrauterine physical insult, such as infection, infarction, hemorrhage, and exposure to a toxin. The condition is characterized generally by bilateral clefts in the grey matter within the cerebral hemispheres usually symmetrical in the region of the central sulcus.The term schizencephaly is derived from two Greek words ; skhizein which means to split, and enkephalos which means brain. Clinical manifestations have variable severity and include developmental delays and retardation, microcephaly, focal or generalized motor abnormalities, and seizures. Diagnosis of schizencephaly is based on brain computed tomography (CT-scan) and/or magnetic resonance imaging. Unilateral sschizencephaly can be differentiated from porencephaly because the fluid-filled component is entirely lined by heteropic grey matter in schizencephaly, whereas a porencephalic cyst is lined mostly by white matter [1-7].

Patients and methods

A dysmorphic male infant who was referred to the neuropsychiatric consultation clinic of the Children Teaching hospital of Baghdad medical city was studied.

Results

H.G was first seen at the age of four month because of psychomotor retardation with no interaction with the mother and no recognition of her face. The boy was hypotonic and had low set ears, retrognathia, and facial dysmorphism with narrow and upslanting palpebral fissures and thin upper lips (Figure-1). Family history was negative for a similar condition.

Brain CT-scan showed large cystic lesion on both sides of the brain communicating with the lateral ventricles bilaterally with a layer of grey matter lining the openings with the ventricles and irregularities in the wall of the ventricles. The third and fourth ventricles were not dilated and the septum pellucidium was normal. The radiologist report suggested the diagnosis of open limb bilateral schizencephaly. Karyotype showed normal finding.

Discussion

Brain migration congenital malformations including schizencephaly, agyria, pachygyria, polymicrogyria, unilateral megalencephaly, and gray matter heterotopias are thought to be caused by insults to migrating neuroblasts during the third to fifth gestational months. They are associated with developmental delay and seizures, and abnormal motor skills.

Schizencephaly is an unusual condition of obscure etiology possibly resulting from an in utero insult leading to maldevelopment or a vascular impairment causing destruction of brain tissues. The cause has most often been described as vascular or idiopathic dysgenesis [8-11].

Figure-1: The boy had low set ears, retrognathia, and facial dysmorphism with narrow palpebral and upslanting palpebral fissures and thin upper lips

Braga et al (2018) reviewed156 articles including 734 patients with schizencephaly, and none of them had facial dysmorphism, low set ears or micrognathia [12].

Conclusion

A new dysmorphic syndrome associated with schizencephaly, facial dysmorphism, low set ears and micrognathia is reported.

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