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Brain Perfusion Single-Photon Emission Computed Tomography Findings in a Patient with Aicardi Syndrome

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Abstract: Aicardi syndrome is a rare X-linked dominant genetic disorder characterized by infantile spasms, agenesis or hypogenesis of the corpus callosum, and chorioretinal lacunae. There remain many unresolved questions regarding the clinical features of this disorder. Recent advancements in neuroimaging techniques have allowed us to obtain information about brain development and perfusion. Thus, we present herein the brain single-photon emission computed tomography (SPECT) findings in a 15-month-old female with Aicardi syndrome, which revealed that perfusion had shifted from the posterior to anterior regions. Furthermore, the right temporal lobe was not visualized, while significant hypoperfusion was determined on the occipital regions and left posterior parietal area. SPECT may be a valuable tool for the assessment of the pathologic functional state in patients with Aicardi syndrome.

Key Words: Aicardi syndrome, brain scintigraphy, SPECT, mental retardation, neuronal migration disorder, child

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Aicardi Sendromlu Bir Hastada Bilgisayarlı Tek-Foton Emisyon Tomografisi Beyin Perfüzyon Bulguları

Özet: Aicardi sendromu infantil spazmlar, korus kallozum agenezi veya hipogenezi ve korioretinal lakünlerle karakterize X'e bağıli dominant geçişli nadir bir genetik bozukluktur. Bu bozukluğun klinik özellikleri ile ilgili birçok çözülmemiş soru bulunmaktadır. Nöro-görüntüleme tekniklerindeki son ilerlemeler beyin gelişimi ve perfüzyonu hakkında bilgi sahibi olmamıza imkan sağlamıştır. Burada, Aicardi sendromlu 15 aylık bir kız bebekte, perfüzyonun posterioridan anterior bölgelere kaydığını gösteren beyin bilgisayarlı tek-foton emisyon tomografisi bulguları sunuldu. Ayrıca, oksipital bölgeler ve sol posterior parietal alanda belirgin hipoperfüzyon saptanmakla birlikte sağ temporal lob izlenemedi. Bilgisayarlı tek-foton emisyon tomografisi Aicardi sendromlu hastalarda patolojik fonksiyonel durumun değerlendirmesi için değerli bir vasta olabilir.

Anahtar Sözcükler: Aicardi sendromu, beyin sintigrafisi, SPECT, mental gerilik, nöronal migrasyon bozukluğu, çocuk

Introduction

Aicardi syndrome (AS) was initially described in 1965 by a French neurologist, Jean Aicardi, and was characterized by the classic triad of infantile spasms, total or partial agenesis of the corpus callosum, and chorioretinal lacunae (1). This condition has been recognized as an X-linked dominant disorder encountered almost exclusively in females, since early embryonic lethality in homozygous males is suspected. Except for one familial instance known to involve two sisters, no affected siblings have been documented, suggesting that the mutation seems to be *de novo* and sporadic rather than hereditary (2-4). The actual frequency of AS is unknown. In series of infantile spasms, 1% to 4% of cases may be due to AS (5). Furthermore, multiple cranial, ocular, and skeletal malformations may be present in the syndrome (6-8).

Many questions regarding the clinical features of this disorder remain unresolved. Recent advancements in neuroimaging techniques, such as magnetic resonance imaging (MRI) and single-photon emission computed tomography (SPECT), have allowed us to obtain information about those defects associated with brain development and perfusion. Thus, SPECT with technetium-99m-hexamethylpropyleneamine oxime (Tc-

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99m HMPAO) was used to assess the cerebral functional state in this anomaly. To the best of our knowledge, this case is a unique patient with AS whose cerebral abnormalities could be shown by brain scintigraphy.

Case Report

A 15-month-old female was referred with developmental delay, medically intractable epileptic seizures, and diminished vision. She was the second child of a non-consanguineous couple and was born at term by vaginal delivery after a healthy pregnancy. The medical history of the patient was normal until the third month of age when infantile spasms started, which were initially responsive to the phenobarbital medication.

Physical examination revealed microcephaly, with a head circumference of 40 cm (less than -3 S.D.). She had infantile spasms medically intractable to antiepileptic medications. Neurological examination revealed a generalized hypotonia and motor-mental retardation. Ophthalmologic examination revealed bilateral diminished sense to light. On fundoscopic examination, bilateral multiple chorioretinal lacunae were seen. The physical examination of other systems and the serological tests for TORCH group infections were normal.

In order to avoid the effect of seizure on the brain perfusion, scintigraphic study was done during a seizure-free period. In the brain SPECT study, at 30 minutes after good venous access was established, 5 mCi (185 MBq) dose of Tc-99m HMPAO (Brain-SPECT, Medi-Radiopharma Ltd, Budapest, Hungary) was injected. Sedation of the patient was done with chloral hydrate (50 mg/kg/dose) approximately 15 minutes before the scanning. We obtained images at 1 h. A single-head gamma camera (Toshiba GCA 602A) equipped with a low-energy, high resolution, parallel-hole collimator was used. The patient's head was fixed with a Velcro restraint and held motionless. The projection data were obtained in a 64x64 format for 60 angles in a 360° arc with 25 s per angle. The raw data were reconstructed and x2 magnification was applied to images after reconstruction for a better visual evaluation. Transverse, coronal and sagittal slices were evaluated visually regarding hypo- and hyperperfusion of the cerebral cortex. Taking account of the whole brain, it was observed that perfusion had shifted from the posterior to the anterior regions. The right temporal lobe was not visualized, while significant

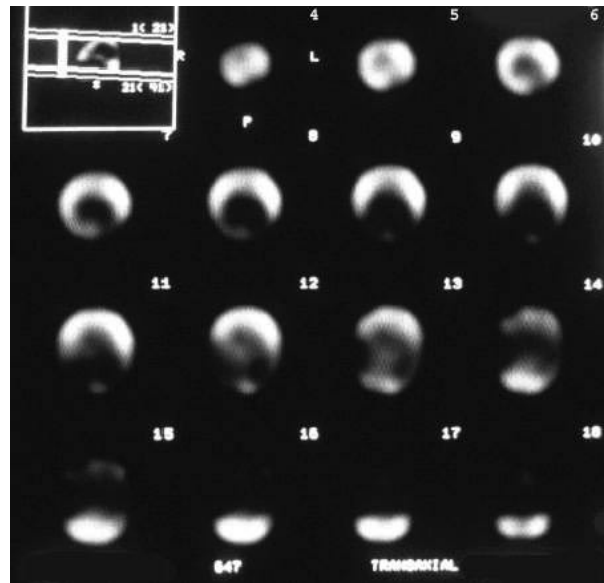
hypoperfusion was determined on the occipital regions and left posterior parietal area. In addition, ventricular areas were seen as slightly larger than normal (Figure 1 A,B,C).

Electroencephalography (EEG) showed hypsarrhythmia with asymmetrical bursts. Cranial MRI revealed the partial agenesis of the corpus callosum and dilatation of the lateral and third ventricles.

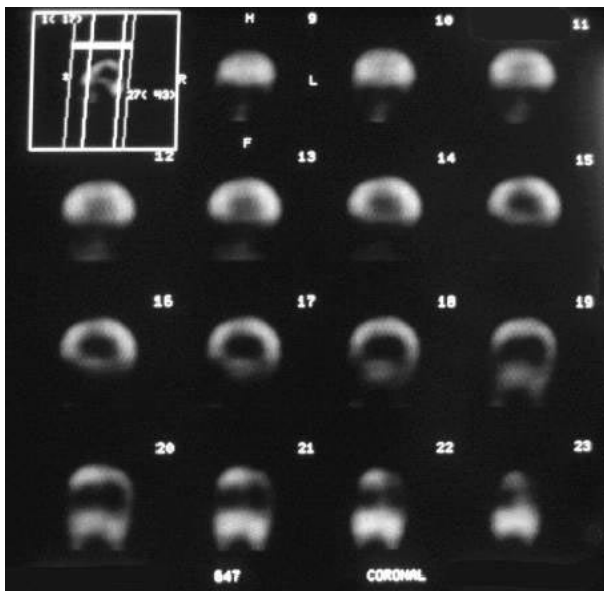
Discussion

Aicardi syndrome should be considered as a syndrome in which the clinical findings and prognosis are heterogeneous. Patients usually present with developmental delay, intractable epilepsy, and numerous other medical problems. Developmental delay in AS is generally profound, involving both motor and linguistic skills, although cases of higher functioning individuals have been reported. The spectrum of motor and mental disabilities is to some extent dependent on the severity of the underlying brain abnormalities (2,8). Variable neurological abnormalities are usually present; the most frequent is hemiparesis or hemiplegia, often on the side where the asymmetric spasms are predominant. Microcephaly of mild to moderate degree may develop but the head circumference is normal at birth (4,6), as in our patient. The epileptic seizures are often difficult to control. In the EEG studies, suppression-burst pattern is almost always asymmetrical and paroxysmal bursts may be unilateral or, when bilateral, may arise independently from hemispheres (3,4,6). Infantile spasms, hypotonia, microcephaly, motor and mental retardation, chorioretinal lacunae, hypogenesis of the corpus callosum, and typical EEG findings were the clinical findings of our case and were strongly suggestive of AS.

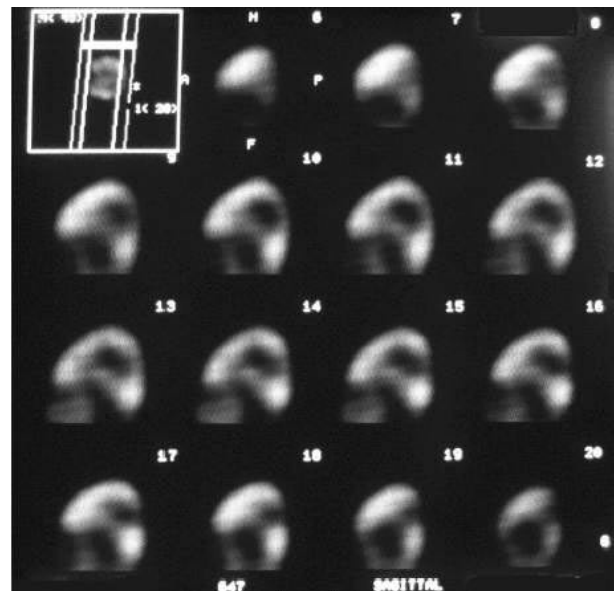
Neuroimaging has been essential in the delineation of the syndrome. Modern neuroimaging techniques have shown that a complex brain malformation was more characteristic than isolated agenesis of the corpus callosum. In a large majority of cases, MRI had shown complex central nervous system (CNS) malformations in addition to partial or total agenesis of the corpus callosum as initially described (3,6,7). Because of the developmental abnormalities of the corpus callosum, AS was considered as one of the few syndromes taking part in the neuronal migration disorders (9,10). Therefore, these complex CNS malformations are more important



(A)



(B)



(C)

Figure 1. Transaxial (A), coronal (B), and sagittal (C) brain perfusion SPECT slices of the patient showing the shifting of cerebral perfusion to the anterior regions, hypoperfusion in occipital and left parietal areas, and no visualization of the right temporal lobe.

for the definition of AS than the callosal agenesis itself, which is a non-specific finding encountered in various disorders or in isolation. In rare cases, only one or two features are present, most frequently cortical malformations and periventricular heterotopias. However, those may be absent or undetectable in a small

number of otherwise typical cases (4). Periventricular heterotopias are more common, as they may escape notice if the quality of MRI is not optimal and if they are not specifically searched. SPECT may be more sensitive in some instances than computed tomography (CT) and MRI (9,10). However, in one study, SPECT was pathologic in

six of eight patients with neuronal migration disorders, which was more sensitive than CT in the diagnosis of heterotopias (11,12). In contrast, Morioka et al. (13) reported the glucose metabolism and perfusion of periventricular nodular heterotopia to be almost identical to those of the normal cerebral cortex using [¹⁸F] fluoro-2-deoxy-D-glucose positron emission tomography (FDG-PET) and HMPAO-SPECT. In the literature, there is no reported case with AS evaluated by brain perfusion SPECT. In our case, the hypo-perfused areas seen on SPECT were not correlated with MRI findings. It can be

said that brain malformations in neuronal migration disorders show various regional cerebral blood flow abnormalities, and these are spread over a larger area than detected by CT or MRI. These perfusion abnormalities may cause the neurophysiological dysfunction in the various areas of the CNS.

We conclude that SPECT is a valuable examination method for the determination of abnormal brain areas and the assessment of the pathologic functional state in patients with CNS malformation, including AS.

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