CASE REPORT

Cerebral infarction in incontinentia pigmenti: the first report of a case evaluated by single photon emission computed tomography

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A 2-month-old girl with incontinentia pigmenti presented with acute-onset right-handed focalized seizures and subsequent seizure generalization. Computed tomography, magnetic resonance imaging and single photon emission computed tomography results indicated that she had multiple cerebral infarctions. These findings suggest that incontinentia pigmenti should be included among the neurocutaneous syndromes associated with ischemic strokes in childhood. This is the first report of a case with incontinentia pigmenti associated with cerebral infarction evaluated by single photon emission computed tomography. \Box Cerebral infarction, incontinentia pigmenti, MRI, seizure, SPECT

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Incontinentia pigmenti (IP, Bloch-Sulzberger syndrome) is a rare neurocutaneous syndrome characterized by congenital skin lesions, dental anomalies, and scalp and ocular abnormalities (1). About 30–50% of IP patients show evidence of central nervous system (CNS) involvement, including mental retardation, seizures, microcephaly, and cerebellar ataxia (1). Recently, there have been several reports of cases of IP associated with stroke or ischemic lesions (2, 3). We report here the case of a IP patient who showed ischemic stroke evaluated by single photon emission computed tomography (SPECT).

Case report

The patient is a female who was born at full term by normal spontaneous delivery from a 30-year-old mother. The pregnancy was normal. At birth, skin lesions such as erythema and vesicles were found on the lower part of her abdomen and on her extremities, and hyperpigmented linear streaks were evident on her limbs. Up to 8.0% of the cells in her peripheral blood were eosinophilic leukocytes. Ophthalmological examination revealed dilatation of vessels and small haemorrhage. She was diagnosed as having IP, while her family medical history was unremarkable.

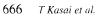
At 2 months of age, she experienced clonic seizures of the right hand, and it soon became generalized and subsequently became status epilepticus. Physical examination findings on admission were normal except for marbled pigmentation of the skin of the legs. Neurologic examination revealed disturbance of consciousness, neck stiffness, a prominent anterior fossa and an increased patellar reflex bilaterally. Haemogram findings and serum electrolyte levels were within normal limits. Lumbar puncture revealed elevation of the leukocyte count $(15/\text{mm}^3)$ and protein concentration (96 mg/dl), while erythrocytes and xanthochromia was not evident.

Electroencephalographs (EEGs) showed multifocal spikes and sharp waves. Computed tomography (CT) revealed a marked low-density area in the left hemisphere and diffuse brain oedema. The patient was treated with glycerol, steroid infusion, acyclovir and anti-convulsants, and her condition gradually improved. CT on the 14th hospital day revealed improvement of oedema, but showed low density in the left hemisphere. Magnetic resonance imaging (MRI) on the 16th hospital day revealed that a large part of the left hemisphere and a small part of the right parietal and occipital regions had abnormal low intensity on T1-weighted image and abnormal high intensity on T2-weighted image (Fig. 1a and b). SPECT performed at the same time revealed markedly decreased blood flow in the various regions (Fig. 2).

A follow-up MRI study performed 4 months later revealed marked atrophic changes of the left hemisphere. Magnetic resonance angiography (MRA) revealed no significant blood flow decrease of the main cerebral arteries, while SPECT revealed decreased cerebral blood flow with slight improvement. EEGs showed multifocal spikes and sharp waves, and she now has delayed motor development and mental retardation.

Discussion

IP is an inherited neurocutaneous disorder with uncertain



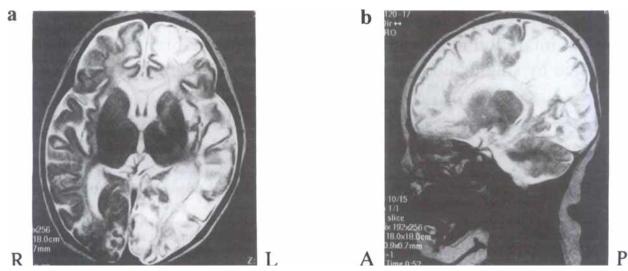


Fig. 1. T2-weighted magnetic resonance imaging (MRI) taken on the 16th hospital day shows abnormal high intensity in the left hemisphere, especially in the left frontal, parietal and occipital regions. (R: right, L: left, A: anterior, P: posterior.)

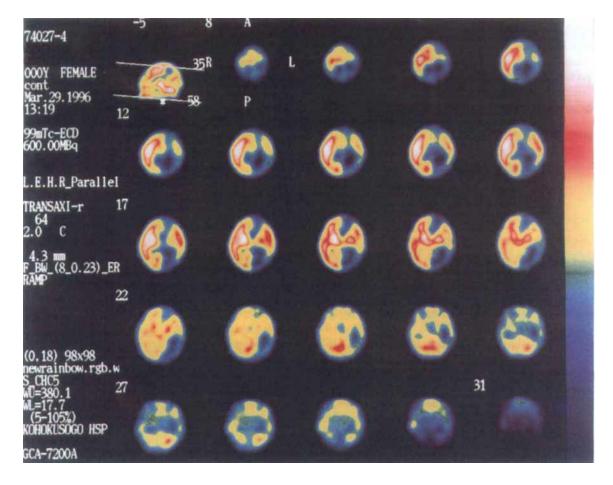


Fig. 2. Single photon emission computed tomography (SPECT) with 99mTc-ethyl-cysteinate-dimer performed on the 16th hospital day shows marked decrease in blood flow in a large part of the left hemisphere, and a small part of the right parietal and occipital regions.

transmission. It is associated with characteristic skin changes and various abnormalities of other organs (teeth, eyes and CNS) (1). Our patient had skin lesions characteristic of IP and a mild retinal disorder, and was diagnosed as having IP.

Recently, several cases of stroke in IP patients have been reported (2, 3). The abnormal CT, MRI, or MRA findings were consistent with blood flow insufficiency, such as infarction, at various periods from prenatal to adulthood (3, 4). In our patient's case, CT and MRI findings and the clinical course are consistent with a diagnosis of infarction in multiple arterial regions. Moreover, SPECT could directly detect decreased cerebral blood flow indicating loss of activity, so it should be used in evaluation of IP patient with neurological symptoms. In addition, Lee et al. reported the presence of lactate by magnetic resonance spectroscopy indicating chronic ischemia in one of five IP patients (3).

These findings indicate that IP should be included among the neurocutaneous syndromes associated with ischemic strokes, and that many of the CNS abnormalities reported in cases of IP could be the result of cerebral ischemic disorders occurring at various periods. In cases of IP, prospective evaluation of cerebral vasculopathy by SPECT or angiography (2, 3), and prophylactic treatment such as aspirin administration (5, 6), should be considered before severe neuropathy occurs.

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