

Moyamoya Syndrome in a Patient with Down Syndrome Presenting with Chorea

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An 11-year-old girl with Down syndrome is reported with moyamoya syndrome; she presented with chorea and mental regression, but had no hemiplegia or convulsions. Magnetic resonance imaging and magnetic resonance angiography were valuable for diagnosis of moyamoya syndrome. It is suggested that moyamoya syndrome be considered as a possible cause of involuntary movements in Down syndrome patients.

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Introduction

Moyamoya disease is a rare cerebrovascular occlusive disease characterized by the angiographic appearance of an abnormal vascular network at the base of the brain, known as moyamoya vessels. There have been increasing reports regarding the association of moyamoya and Down syndromes [1-5]. This association has been assumed to be caused by a systemic vascular disorder in patients with Down syndrome [6]. The main clinical manifestations in patients with Down syndrome-associated moyamoya syndrome, which were reported previously [1-5], include hemiplegia, muscle weakness, and convulsions. We initially reported a child with Down syndrome-associated moyamoya syndrome, confirmed by magnetic resonance imaging (MRI) and magnetic resonance angiography (MRA), who presented with choreic movements.

Case Report

An 11-year-old female with Down syndrome (21 trisomy, 47XX, +21) was referred to Asahi General Hospital for evaluation of chorea and mental deterioration. She uttered her first word at about 20 months and walked unassisted at 22 months. No cardiac malformations were present. Her developmental quotient (DQ) score at the age of 6 years was 76. At age 10, she presented with mental deterioration and fluctuating choreic movements, involving mainly both upper extremities.

On admission, she could only utter several words, although she had been able to converse before age 10 years. Her DQ score had decreased to below 50. Neurologic examination was characterized by distal choreic movements of all limbs, especially of both upper limbs. Cranial nerves, muscle strength, and pain sensation were normal. There were no signs of cerebellar dysfunction or pathologic reflexes. We performed extensive laboratory studies, including thyroid function, serum antistreptokinase, antistreptolysin O, lactate and pyruvate, and antinuclear, anti-DNA titers, all of which were within normal limits.

Cranial MRI revealed multiple, round signal void signs in the basal ganglia and thalamus, occlusive changes of the middle cerebral arteries, and infarction in the white matter adjacent to the right anterior horn (Figs 1A-1C), which were consistent with the findings in moyamoya syndrome on MRI. Further evaluation by MRA demonstrated collateral channels at the base of the brain and the disappearance of the anterior and middle cerebral arteries, which are typical of moyamoya syndrome (Figs 2A,2B). Cerebral angiography was not performed because of this unequivocal evidence of moyamoya syndrome.

Discussion

The clinical manifestations in childhood moyamoya disease or Down syndrome-associated moyamoya syndrome usually comprise transient cerebral ischemic episodes and infarctions (i.e., paralysis, headache, sensory disturbance, seizures) [5,7]. Conversely, involuntary movements caused by transient ischemia or infarction of striatal lesions, probably resulting from the regulatory release of the globus pallidus [8], have been rare clinical symptoms of childhood moyamoya syndrome. The frequency of involuntary movements in idiopathic childhood moyamoya patients has, at most, been reported to be 2 of 81 patients [7], but none of 19 Down syndrome-associated moyamoya patients [5].

We previously reported that the results of MRI indicated that younger children with moyamoya disease demonstrated a significant increase in cortical and subcortical infarction, and a decreased incidence of deep white matter infarction in the centrum semiovale and basal ganglia, in contrast to adult patients [9]. We added the hypothesis that the basal ganglia and centrum semiovale may be spared from infarction due to the excellent collateral circulation in early childhood moyamoya syndrome, which may account for the high frequency of motor palsy, seizures, and mental retardation, and low incidence of involuntary movements.

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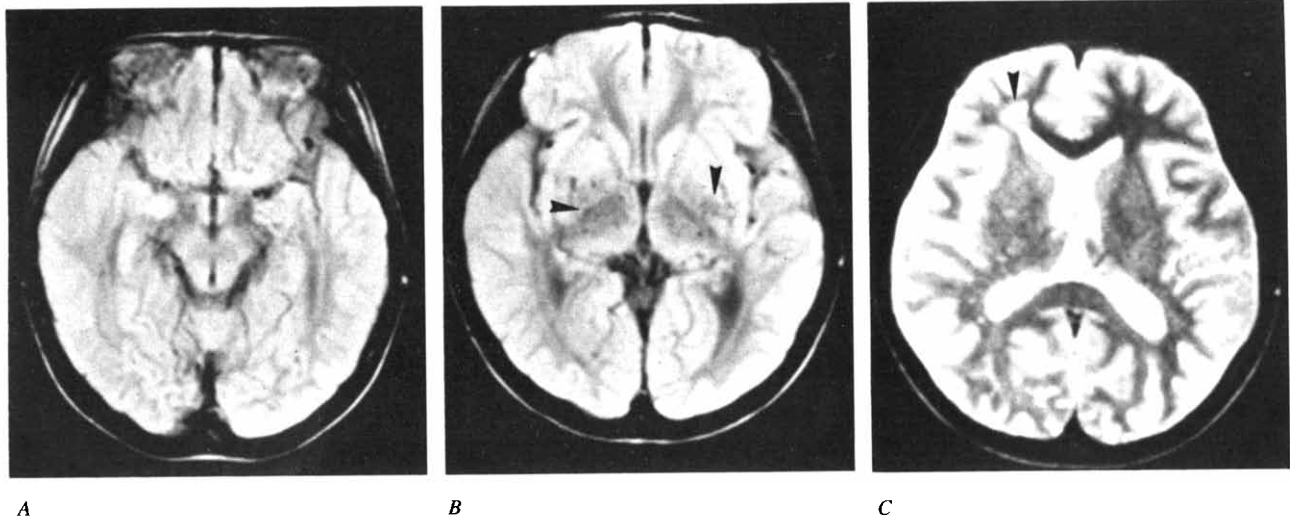


Figure 1. (A) Proton-density weighted MRI (TR: 2,000, TE: 30) documenting the disappearance of the signal void sign on bilateral MCA. (B) Proton-density weighted MRI of multiple round and tortuous signal void areas (moyamoya vessels) in the basal ganglia and thalamus (arrowheads). (C) T₂-weighted MRI (TR: 2,000, TE: 80) demonstrating white matter infarction adjacent to right anterior horn (arrowhead).

Our patient mainly presented with fluctuating choreic movements in addition to mental regression. Chorea can be observed in patients with Sydenham chorea, Huntington chorea, systemic lupus erythematosus, hyperthyroidism, and drug-induced movement disorders. Although it has been observed most commonly among these causes of chorea in childhood, the incidence of Sydenham chorea is now greatly diminished with the concomitant decrease of rheumatic fever. In spite of thorough examinations, we could not find any evidence that supported these disorders. From the results of MRI and MRA, we made a diagnosis of moyamoya syndrome.

The incidence of moyamoya syndrome could be far greater in children with Down syndrome. According to Fukushima et al., the estimated prevalence of moyamoya syndrome in Japanese children could be 1:15,000, but could be as frequent as one in several hundred in Down syndrome [10]. As to the pathogenic mechanism for the higher rate of this association, several hypotheses have been presented, including developmental vascular abnormality, vascular fragility, and vascular hyperpermeability [3,5]; however, no conclusive data have been reported.

Moyamoya syndrome should be considered in the differential diagnosis of acquired chorea in Down syndrome

patients, even though the complicated incidence of involuntary movements during childhood is low. Furthermore, we should examine Down syndrome patients much more closely for involuntary movements that may not have come to their attention.

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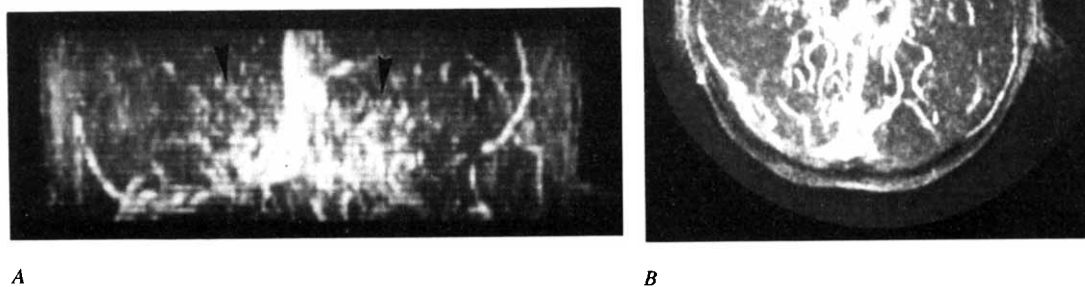


Figure 2. (A,B) MRA (TR: 40, TE: 9) demonstrating the disappearance of ACA and MCA and the appearance of moyamoya vessels (arrowheads).

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