CERVICAL MYELOPATHY DUE TO ATLANTOAXIAL INSTABILITY ASSOCIATED WITH DOWN SYNDROME: A CASE REPORT

Min-Lan Tsai, Jia-Kan Chang

Abstract

Spinal cord compression secondary to atlantoaxial instability in Down syndrome is relatively uncommon in young children. We report a 4-year-old boy with Down syndrome and myelopathy caused by atlantoaxial instability (AAI). At presentation he had right limb hemiparesis, ataxia, and bilateral hyper-reflexia for 2 weeks. There was no obvious history of trauma except for frequent upper respiratory tract infection. Cervical spine x-ray showed increased distance between the atlas and the dental process, which fulfilled the criteria for atlantoaxial instability, and cervical MRI showed spinal cord compression and myelomalacia. His neurologic signs gradually improved after surgical treatment. This report emphasized the importance of early recognition and diagnostic approach for prevention of the potential cervical spinal cord injury secondary to AAI in very young children with Down syndrome.

Key words: Cervical myelopathy, Atlantoaxial instability, Down syndrome

INTRODUCTION

Down syndrome is one of the most common chromosome disorders. It is caused by trisomy of chromosome 21 and occurs in 1.5 of every 1000 live births. It is characterized by mental retardation and skull, brain, and cardiac abnormalities. Atlantoaxial instability (AAI), also known as atlanto-axial subluxation, is defined as a distance greater than 4.5 mm between the odontoid process of the axis and the anterior arch of the atlas.\(^1\) Atlantoaxial instability occurs in 10-30% of Down syndrome patients.\(^2-5\) The high incidence of AAI is thought to be due to abnormalities of the odontoid process of the axis, a loose transverse ligament of the atlas,\(^1,6\) and a hypoplastic C1 posterior arch causing the possibility of compression of the spinal cord.\(^7\) However, only 0.4-4.3% of Down syndrome patients with AAI develop cervical myelopathy.\(^4,8\) Morton et al.\(^9\) found an overall decrease in atlanto-dens distance over time and no new cases of AAI in a 5-year follow-up study. Other authors also reported the findings of no changes in atlanto-dens distance or clinical status during follow-up in different groups of Down syndrome patients.\(^4,10,11\) Pueschel et al.\(^12\) reported that the average onset age of myelopathy is 10.5 years. There have been a very limited number of case reports in Taiwan.\(^13,14\) Although there has been increased attention with regard to the diagnosis of AAI in patients with Down syndrome in recent years, the pitfall for the clinician is misdiagnosis of the central nervous system (CNS) problem.

Correspondence: Dr. Min-Lan Tsai
Department of Pediatrics, Cheng Hsin General Hospital; 45, Cheng-Hsin Street, Beito District, 112, Taipei, Taiwan
Phone: 886-2-2826-4400 ext. 3351; E-mail: minlan456@hotmail.com
when patients have focal signs such as hemiparesis and Babinski’s sign. We report a 4-year-old boy with Down syndrome who had cervical myelopathy caused by atlantoaxial instability.

**CASE REPORT**

A 4-year-old boy with Down syndrome was admitted to our hospital after he had right side limb weakness and limping gait for about 2 weeks. His forehead had multiple ecchymoses because of frequent falling down and unsteady gait which developed during the previous 3-4 days. He was diagnosed with Down syndrome at birth, but had never undergone cervical spine screening. He was mentally retarded and diagnosed with congenital hearing impairment and speech delay, and impaired gross and fine motor function. He did not have a diagnosis of congenital heart disease. Two months earlier he was transferred for the first time to a mental retardation school and underwent hydro-rehabilitation in our rehabilitation department. He did not participate in any remarkable aggressive exercises or sports. Based on recall by his caretaker and mother he had frequent upper respiratory tract infections since his infancy. He had just recovered from an upper respiratory infection 1 week earlier.

His physical examination revealed a typical mongoloid face. He was unable to speak because of his hearing impairment and mental retardation. Muscle strength of his right arm and leg was grade 3-4/5 without obvious spasticity, and bilateral Babinski’s signs were positive. Deep tendon reflex showed hyper-reflexia over all four extremities and bilateral ankle clonus was positive. He was ataxic and only able to walk a few steps. His bladder control had not developed since birth. He was first brought to our emergency department and CT of the brain was performed immediately. No active brain lesion was found. He was then referred to our pediatric neurologic clinic and admitted for further evaluation.

Plain cervical x-ray films showed that the atlanto-dental interval was 6.2 mm in the neutral position (Fig. 1), which confirmed the diagnosis of atlantoaxial instability. Chest x-ray showed 12 ribs and no remarkable active pulmonary lesion. Magnetic resonance imaging of the cervical spine revealed a focal intracordal high signal intensity lesion (0.92 cm in length) on the T2 weighted image at the C1 level and myelomalacia was evident. Spinal cord compression at the C1-2 level was due to compression by the posterior arch of the atlas dislocated forward (Fig. 2). Widening of the C1-2 distance and poor development of the anterior arch of C1 was also suspected on the cervical MR image. Brain MRI showed dilatation of the bilateral lateral ventricles especially the bilateral temporal horns (not shown) and a large cyst without mass effect or enhancement over the retrocerebellar region (Fig. 3). An enlarged cistern magnum or an arachnoid

![Fig. 1. Lateral cervical spine x-ray demonstrates forward subluxation of the atlas and increased atlantoaxial distance (long arrow). Agenesis of the odontoid process of the axis (short arrow) was also shown and there was no fracture of the odontoid process, which was confirmed by surgical findings.](image-url)
Fig. 2. Magnetic resonance imaging of the cervical spine in A showed a focal intracordal high signal intensity lesion (0.92 cm in length) on a T-2 weighted image at the level of C1 (white arrow), and in B isodensity on a T-1 weighted image (white arrow). Myelomalacia at the level of C1 spinal cord was evident.

Fig. 3. Brain magnetic resonance image showed a large cyst (arrows) without mass effect or enhancement over the retrocerebellar region, as shown in T-1 weighted axial view in A and T-1 weighted mid-sagittal view after contrast medium in B. A large cistern magnum or arachnoid cyst was suspected.
cyst was suspected. Echocardiography revealed physiologically mild pulmonary and tricuspid regurgitation and no evidence of congenital heart disease. He was subsequently referred to the neurosurgical department of Taipei Veteran General Hospital for further surgery and rehabilitation. C1-2 stabilization and posterior fusion with autologous iliac bone and artificial bone grafting were performed. His condition gradually improved and he was discharged with halo-vest fixation for 3 months. His muscle power gradually improved and he was able to walk a relatively long distance compared to before the operation.

**DISCUSSION**

Atlantoaxial instability infrequently becomes neurologically symptomatic in very young children (less than 5 years of age) with Down syndrome. Sporadic case reports are of young children who usually presented with neurologic symptoms such as limb paralysis (spastic tetraplegia, hemiplegia, or quadriplegia), neck pain, torticollis, ataxia, hyper-reflexia, paresthesia, and bladder dysfunction. Taggard et al. found that the most common chief complaints were hyper-reflexia (67%), neck pain and torticollis (42%), quadriparesis (35%), ataxia (19%), minor trauma (11%), and frequent upper respiratory tract infection (14%). The youngest child reported in the literature was a neonate who presented with respiratory difficulty after patent ductus arteriosus ligation. Mortality has been documented after spinal cord compression.

Our patient was 4 years of age. He had progressive right limb hemiparesis and ataxia for 2 weeks, and improved gradually after surgery. Cervicomedullary compression is usually associated with ataxia and progressive weakness. Our patient developed ataxia followed by right hemiparesis, indicating the progression of spinal cord and possibly lower medullary compression. There was no major or minor trauma in our patient except a history of frequent upper respiratory tract infection. A functional anatomic explanation is that the upper jugular digastric lymphatic chain serves as the site of lymphatic drainage for both the craniovertebral junction and the nasopharynx and paranasal sinuses. Infection of the latter can result in retrograde change with hyperemia, synovial inflammation, joint effusion, and further ligamentous laxity in the craniovertebral junction. Patients with Down syndrome may have an increased risk of AAI during upper respiratory infection.

Magnetic resonance imaging is the best tool to show the severity of spinal cord compression and exclude other combined CNS anomalies such as Arnold-Chiari malformation or syringomyelia. T-2 weighted MRI usually demonstrates high intensity in the spinal cord, whereas T-1 weighted MRI shows low intensity when myelopathy occurs. In severe cases, MRI can demonstrate well the deviation of angle of the clivus/odontoid process. Cervical MRI in our patient demonstrated a 0.92 cm in length intracordal lesion of myelomalacia (Fig. 2). Myelomalacia is a term that refers to an area of softening of the spinal cord that has been reabsorbed by the body leaving a “hole” or cavity, or spinal fluid-filled space. Myelomalacia can be caused by trauma from any etiology. It is thought that myelomalacia was due to spinal cord compression by AAI in our case. Interestingly, the brain MRI in our patient demonstrated a posterior retrocerebellar cyst without mass effect (Fig. 3), which is similar to the other case report of AAI and neurologic involvement. The posterior fossa cyst may have resulted from the poor development of the occipito-cervical region; the relationship with AAI is not clear. The brain MRI in our patient also displayed ventriculomegaly over the bilateral temporal horns.

Compared to the screening of cardiologic, endocrinologic, and hearing abnormalities, the screening of AAI in patients with Down syndrome may be sometimes overlooked. Our patient never underwent cervical spine screening before. The Committee on Sports of the American Academy of Pediatrics recommends cervical roentgenograms to rule out AAI in children with Down syndrome between ages 2 and 6. It recommends a limitation on sports and other activities that
produce stress in the head and neck region if the children have an atlantodental distance greater than 4.5 mm and the requirement of further evaluation. 25-27 Harley et al. 26 also suggested that the screening of the cervical spine should be done before surgery because of subsequent complications during anesthetic procedures and positioning. 21 If neurologic symptoms or signs appear, all strenuous activities should be restricted and operative stabilization of the cervical spine should be considered.

In conclusion, we report a case of cervical myelopathy with neurologic signs of right hemiparesis, ataxia, and hyper-reflexia caused by AAI in a patient with Down syndrome. We conclude that the diagnosis of AAI should be immediately considered in any child with Down syndrome who has developed deterioration in ambulatory skills, brisk reflexes, change in bowel or bladder function, or neck posturing, as many of these children are retarded and have little ability to vocalize their complaints. Cervical spine roentgenograms should be obtained, and we suggest that MRI is the best tool to diagnose injury to the spinal cord and related structures. Early recognition of neurologic signs and early management with fixation or surgical approach can reduce morbidity and disability of these children and guide parents in making decisions for their children with regard to safe participation in sports.

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REFERENCES


唐氏症合併頸部寰椎關節不穩定引起之頸部脊髓
病變：一病例報告

蔡明蘭, 張嘉侃

摘要

唐氏症合併頸部寰椎關節不穩定引起之頸部脊髓病變在年齡小的小孩並不
常見，我們報告一個四歲的唐氏症小男孩出現右側肢體無力，走路不稳，易跌
倒已有二星期，據照顧者陳述，除了有上呼吸道感染外，並無明顯外傷，亦無
做翻筋斗的動作。頭椎X光檢查發現寰椎關節距離有顯著增加，而造成半脫位
情形，磁共振檢查呈現脊髓被脱位的關節壓迫，而呈現局部脊髓軟化現象，
病人經開刀手術固定及裝置halo-vest 固定後，運動功能已經有顯著改善。對
於唐氏症的孩童，因其第一、二頸椎易於鬆脫及其韌帶鬆弛，建議應早期做頸
椎X光篩檢，而有神經學的臨床症狀時亦應早期診斷及處置，以減少唐氏症兒
的後遺症。

關鍵詞：唐氏症，頸部寰椎關節不穩定，脊髓病變

聯絡人：蔡明蘭醫師
112 台北市北投區振興街 45 號，振興醫院小兒科
電話：02-2826-4400 轉 3351；E-mail：minlan456@hotmail.com