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ATLANTOAXIAL INSTABILITY IN DOWN'S SYNDROME : A CASE REPORT

DOWN SENDROMUNDA ATLANTOAKSİYAL İNSTABİLİTE : OLGU SUNUMU

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SUMMARY

Down Syndrome (DS) is the most common chromosomal disorder in humans and presents abnormalities in several organ systems. Mental retardation, endocrinologic and cardiologic abnormalities are well recognized by pediatricians, but various cervical spine abnormalities are less appreciated. Widening of the atlanto-odontoid distance and atlantooccipital instability occur in up to 20% and 60% of DS patients. Most of the patients develop neurologic symptoms after acute trauma or anaesthesia. In this report 16 year old girl with DS who spontaneously had acute weakness and quadriparesis is presented.

ÖZET

Down Sendromu (DS) en sık rastlanılan kromozom anomalisi olup, çeşitli sistemlerde oluşan anomalilerle birliktedir. Mental retardasyon, endokrinolojik ve kardiyolojik anomaliler pediatristler tarafından iyi bilinmekte ve izlenmektedir, ancak servikal spin anomalilerine daha az sıklıkla rastlanılır. Atlanto-odontoid mesafenin genişlemesi ve atlantooksipital instabilite DS'lu hastaların %20-60'ında görülmektedir. Hastaların çoğunda nörolojik semptomlar akut travma veya anestezi sonrasında ortaya çıkar.Bu yazıda spontan olarak akut güçsüzlük ve kuadriparezi gelişen 16 yaşında DS'lu bir kız olgu sunulmuştur.

INTRODUCTION

Down syndrome (DS) is one of the most common chromosomal disorders. The children have mental retardation, skull, brain and cardiac abnormalities. These children are usually followed by genetic and cardiology departments. The association of DS and instability of the upper cervical spine is well known. Reported abnormalities involving cervical spine are occipito-atlantal instability, atlanto-axial instability, occipitalization of the atlas and os odontoideum.

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The number of patients with symptoms related to C1-2 instability or other anomalies of the cervical spine is much lower. Atlantoaxial instability(AAI) is defined as a distance greater than 4.5 mm between the odontoid process of the axis and the anterior arch of the atlas. Spinal cord compression may be a consequence of atlantoaxial instability. There is not a consensus about the usefulness of radiograph screening in predicting spinal cord injury in children with DS. A minority of children with DS with cervical instability have neurologic symptoms(1,2). In this report 16 years old girl with DS and AAI who had developed acute neurologic symptoms is presented.

CASE

16 year old girl with Down Syndrome referred to the hospital with a 3 months history of progressive clumsiness in her hands and walking difficulty. She had mild mental retardation and could read and write. She had been followed up by pediatric cardiology department because of mitrale valve prolapsus. Her family reported that she had fallen down from her fathers lap while coming to the intensive care unit because of a severe respiratory infection 4 months ago.

Her physical examination revealed mild spastic tetraparesis and brisk deep tendon reflexes. There was no pathological reflex. Muscle strength was 4/5 in all extremities. Magnetic resonance imaging of cervical spine revealed pressing of odontoid process to bulbus(Figure 1).

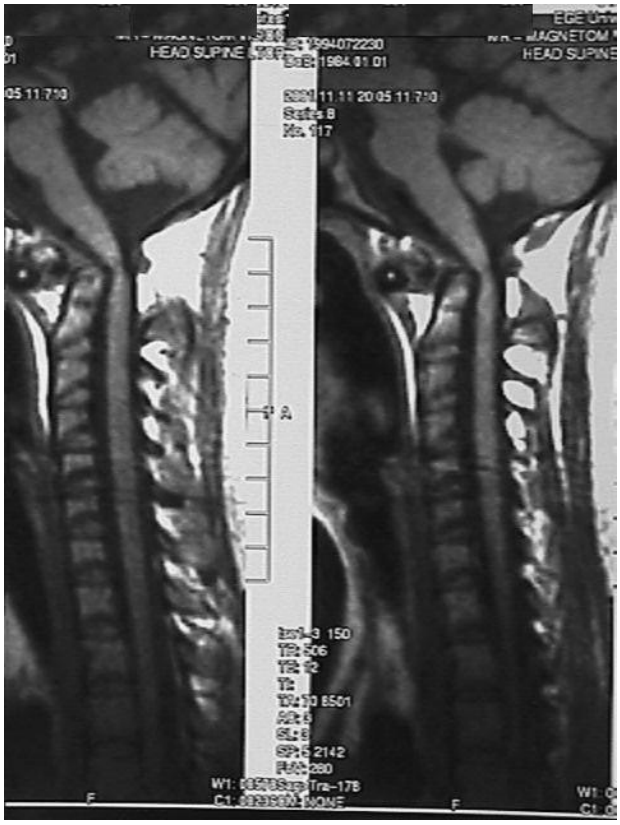

Figure 1. T1 weighted images showed significant pressing of odontoid process to bulbus in sagittal plans.

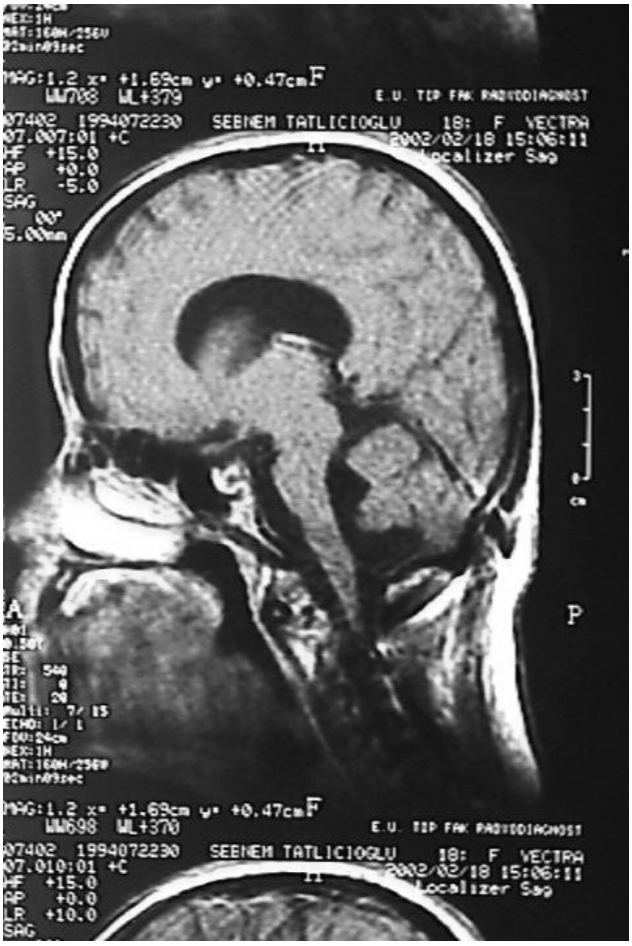

Figure 2. T1 weighted sagittal plan showed the disappearance of pressing after the resection of odontoid process.

C1-2 dislocation was determined with her clinic and radiological findings. Odontoid process was resected (Figure 2). After one week C1-2 stabilization by wiring and iliac graft was performed. Cervical axis was improved after occipitocervical instrument(Figure 3). Her quadriparesis significantly regressed and radiological findings improved after the operation Down syndrome can manifest abnormalities in several organ systems. Widening of the anterior atlanto-odontioid distance and atlantooccipital instability may occur up to 60% of the cases but they rarely become neurologically symptomatic.

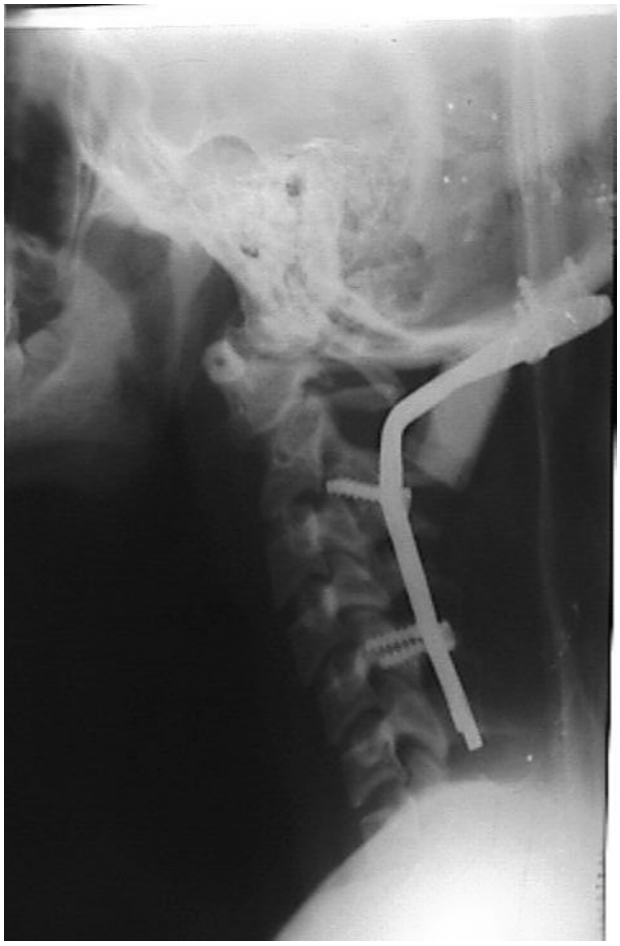

Figure 3. Lateral cervical graphy showed the improvement of the cervical axis after occipitocervical instrument

Atlantoaxial subluxation rarely becomes neurologically symptomatic in very young children. The youngest child in literature is a neonate. Her neurologic abnormalities revealed after patent ductus arteriosus ligation. She had trace movement, few spontaneous respirations and left lower extremity clonus(3). The authors emphasized the need for preoperative screening of all patients with DS. Another young child in literature is a 32 month old girl with DS who had tetraparesis due to AAI. The authors reported that her neurologic symptoms regressed and muscle strength was normal after operation(4). The incidence of instability increases with age. Our case was 16 years old. She was asymptomatic previously and neurologic impairment especially brisk reflexes developed after a minor trauma. Neurologic abnormalities improved completely after the operation.

Harley et al also recommended the screening before surgery, because subsequent complications during anesthetic induction, positioning and manipulation can occur. So the children with an atlantodental interval of greater than 4.5 mm should have further evaluation(5).

Cremers et al evaluated 400 children with DS. The atlantoaxial distance was more than 4 mm in 91 of them. They divided these children into 2 groups. One group was allowed to do habitual sports and exercise activities but the other was not. After a year there were no differences between the groups in scores of functional motor scale, the frequency of neurological signs and atlantoaxial distance. They concluded that there was no reason to stop children with DS from playing certain sports and screen before sports activities(6). But the screening has been widely recommended and enforced by some sporting organisations for several decades(7). Our case had no predispositioning factors such as sport activity or manipulation for anethesia.

Taggard et al studied the cranivertebral junction abnormalities in 36 DS patients. They found that the most common clinical complaints included neck pain and torticollis. Cervicomedullary compression was associated with ataxia and progressive weakness. Hyperreflexia was documented in 24 of the 36 cases. Thirteen patients suffered from varying degrees of quadriparesis(8). Our case did not have neck pain or ataxia. Weakness had developed progressively. Craniovertebral abnormalities can be very important sometimes. Progressive respiratory failure and death had been reported in previously asymptomatic 49 years old patient with DS patient(1).

Morton et al examined 90 children with radiography and repeated the observation 5 years later. No one developed AAI on repeat testing who had not had it earlier. Only one child who had previously had normal neck radiography developed acute symptomatic AAI after ear, nose and throat surgery(9). So radiography is recommended for screening programme. Our case did not have any cervical radiography for screening previously. We also think that screening patients with DS is useful for following up and restricting patients from dangerous activities.

As a result cardiologic, endocrinologic abnormalities are usually screened in DS patients, but screening of AAI can be sometimes underlooked. It is important in these children especially preoperatively and before their participation in sports. Patients with no neurologic abnormalities but with radiologic findings require further evaluation. At least they should be restricted from active sports and followed up periodically. Another important point is; our case represented with mild clumsiness and walking difficulty. Brisk deep tendon reflexes were the first evidence of AAI. So the physician must be aware of AAI in DS patients, sometimes only brisk reflexes can be a guide sign. Early recognition and management of these children can reduce the morbidity and guide parents toward safe participation in athletics. So the primary care physicians must be careful for screening the Down syndrome patients.

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