C1-C2 Luxation in Patient with Down Syndrome. Case Report

Luxação C1-C2 em Paciente Portador de Síndrome de Down. Relato de Caso

Maurus Marques de Almeida Holanda1, Rayan Haquim Pinheiro Santos2, Gustavo de Moura Peixoto2, Normando Guedes Pereira Neto2, Luiz Ricardo Santiago Melo3

1Neurosurgeon, Associate Professor of Neurology at Universidade Federal da Paraíba (UFPB), João Pessoa, PB, Brazil
2Graduation Students of Medicine at Universidade Federal da Paraíba (UFPB), João Pessoa, PB, Brazil
3Neurosurgeon at Unimed Hospital, João Pessoa, PB, Brazil.

ABSTRACT

Introduction: Down syndrome is caused by trisomy of chromosome 21, and it is the most common chromosomal abnormality in humans. This genetic condition is characterized by multiple skeletal abnormalities, including atlantoaxial instability that occurs in 9-31% of patients presenting clinical consequences in 1-3% of them. This study aims to report a case of atlantoaxial instability with clinical repercussions of quadriaparesia. Case Report: A case of a 10-year-old boy, with Down syndrome, who could not get up for three months is reported. Clinical examination revealed quadriaparesis. The lateral radiograph of the cervical spine showed a atlanto-odontoid distance of 6mm, confirming atlantoaxial instability.

Key words: Down syndrome; Atlantoaxial instability; Atlanto-occipital instability; Craniovertebral junction

RESUMO

A Síndrome de Down é provocada pela trissomia do cromossomo 21, sendo a anomalia cromossômica mais comum em humanos. Esta condição genética caracteriza-se por múltiplas anomalias esqueléticas, incluindo a instabilidade atlantoaxial que ocorre em 9-31% dos pacientes, apresentando consequências clínicas em 1-3% dos mesmos. Este estudo tem por objetivo relatar um caso de instabilidade atlantoaxial com repercussões clínicas de tetraparesia. Relato de caso: Menino de 10 anos, portador de Síndrome de Down sem conseguir levantar-se por três meses. O exame clínico revelou tetraparesia com confirmação de instabilidade atlantoaxial através de radiografia lateral de coluna cervical evidenciando distância atlanto-odontóide de 6mm.

Palavras-chave: Síndrome de Down; Instabilidade atlantoaxial; Instabilidade atlanto-occipital; Articulação craniovertebral

INTRODUCTION

Down syndrome (DS), caused by human trisomy 21, is the most frequent live-born human chromosomal disorder, occurring in one in 700–800 newborns10,31. This syndrome was first described by Langdon Down in 1866, and characterized by multiple changes: cardiovascular, gastrointestinal, muscular hypotonia, abnormalities in the central nervous system (difficulty in learning, memory, susceptibility to epilepsy), disorders of ocular motility, susceptibility to leukemia, skeletal abnormalities, among others10, 14, 20, 33.

Among the skeletal abnormalities, skull and craniocervical junction have an important role, due to the risk of acute spinal cord injuries and even death. These changes were first described in 1961 by Spitzer et al. reporting nine cases of atlanto-occipital dislocations32,35. In addition to displacement between atlas and occiput, atlantoaxial instability has been described in 9-31% of cases, with clinical consequences in 1-3% of them4,27,35.

This study aims to report a case of a patient with Down syndrome and craniocervical junction abnormalities with clinical consequences.

CASE REPORT

A 10-year-old boy with Down syndrome was admitted to our service with severe quadriaparesis persisting for 3 months. He was the first child of a 40-year-old mother and was spontaneously delivered after a normal full-term pregnancy. He had a typical
mongoloid face and the intelligence of a 7 month-old infant. He had mental and motor developmental retardation, but he was able to stand with some assistance when he was 18 months old. However, he developed quadriparesis and became unable to stand by 10 year old.

He was admitted under a diagnosis of atlantoaxial instability (AAI). Neurological examination demonstrated severe spastic quadriparesis (Fig. 1) and bilateral Babinski’s sign. Plain cervical X-ray films disclosed AAI, which increased by neck flexion. The atlanto-odontoid distance (AOD) was 6 mm. The magnetic resonance imaging (MRI) showed compression of cervical spinal cord at C2 level (Fig. 2). Craniocervical fixation was performed with rigid occipital instrumentation up to C6 fusion, due to thinning of C1 and C2 posterior elements, fixed with C3 to C6 screws (Fig. 3). The patient developed bradycardia during surgery, precluding the use of rib autograft and opting for allograft hydroxyapatite. The patient underwent cervical collar immobilization for 6 months, evolving with motor deficit improvement (Fig. 4).
Children with Down syndrome have multiple skeletal abnormalities, from the appendicular skeleton (such as short femur and humerus and pelvic changes) to the axial skeleton, with differences in the number of ribs and abnormalities in the skull and spine. Spitzer et al., in 1961, described several facial skeleton and skull abnormalities, such as brachycephaly, lower location of the cribiform plate, greater foramina number and persistent metopic suture, changes in the nasal bone, the maxilla, mandible, among others.

Additionally, Spitzer et al. were the first to describe craniovertebral junction abnormalities in Down syndrome, reporting atlanto-occipital dislocation in 9 of 29 patients. Tishler et al. later reported other abnormalities of the upper cervical spine in children with this syndrome, describing atlanto-odontoid distance greater than 4 mm in 13 of 18 patients studied; this demonstrates the occurrence of atlantoaxial instability in these cases.

Uno et al. reported that the increased occipitoaxial mobility occurs only in patients with atlantoaxial instability, which was later described by Taggard et al. with the simultaneous presence of atlantoaxial and atlanto-occipital instability in 11 cases. Atlanto-occipital joint hypermobility is observed in up to 63% of individuals with Down syndrome, and despite not being related to neurological risk, concomitant changes as bone malformations and atlantoaxial instability can exacerbate this condition. Rotational dislocations, though very rare, can also occur in these patients.

Atlantoaxial joint is the most mobile region of the entire spinal column, and therefore more likely to suffer instabilities. Atlantoaxial instability is the increased mobility in the joint between the atlas and axis, and it can also be called atlantoaxial subluxation. Many authors have described the presence of AAI in patients with Down syndrome with rates ranging from 9-31%. Other authors have reported that up to 40% of Down syndrome patients have AAI.

Due to this prevalence, AAI in patients with Down has been researched. Some authors have reported the importance of radiographic evaluations to avoid activities that can be life-threatening for patients with AAI. Furthermore, the difficulty in communicating with these people may cause no perception of these symptoms. Other authors report that this approach has high cost and the radiographs have low reproductive aspects. Szpak et al. add that there is no consensus in literature about the need for X-ray use for following-up Down patients in order to verify possible atlanto-occipital junction luxation, but its accomplishment, especially when these patients undergo medical procedures at high risk, is suggested.

Despite the high prevalence of AAI in this group of patients, only 1-3% of them are symptomatic. These symptoms include nuchal pain, poor cervical mobility, fatigue, difficulty walking, abnormal gait, occasional loss of consciousness, clonus, abnormal reflexes, sensory changes, which may progress to paraplegia, hemiplegia, quadriplegia or death.

AAI cause is still widely discussed. Many authors report that the main factor that predisposes the imbalance in atlantoaxial junction is the alar ligament laxity. Uno et al. reported 61.2% of patients with generalized ligamentous laxity and 22.5% of AAI among 80 patients with Down syndrome, but no statistically significant difference between generalized ligamentous laxity and AAI was found.

Nevertheless, the precipitation of symptoms by upper respiratory tract infections has been described due to the fact that the nasopharynx and the craniocervical junction possess the same lymphatic drainage. Therefore, infection can ascend causing ligament laxity by local inflammation. Other causal factors are atlas and axis malformations, such as hypoplasia of the arches, dysplasia and odontoid with incomplete fusion (the odontoideum), being the last ones present in 6% of patients with Down syndrome.

The diagnosis of atlantoaxial instability is determined by measuring the atlanto-odontoid distance, i.e. the distance obtained between the odontoid process and the posterior border of the anterior arch of the atlas, by means of lateral cervical spine radiographs. These radiographs should be performed with technical standards which are more accurate than those used for the general population, so as not to diagnose atlantoaxial instability in patients without this condition.

The normal ranges of AOD differ between adults and children. Values greater than 2.5 to 3mm are considered suggestive of AAI for adults. For children, several limits have been used: 4mm, 4.5mm, 5mm and 6mm. Matos distributed his patients into three groups according to AOD:
group I (AOD less than 4.5 mm), group II (AOD between 4.5 and 6 mm) and group III (AOD greater than 6 mm).

Braakhekke et al.\textsuperscript{4} and Pueschel et al.\textsuperscript{27} reported the importance of computed tomography (CT) in the measurement of sagittal diameter behind the odontoid (using as limit 14 and 16 mm, respectively), in addition to detecting other abnormalities (basilar impression, atlas and axis malformations). Browd et al.\textsuperscript{5,6} described the use of CT to evaluate the atlanto-occipital junction, but this approach should not be performed in all patients due to its high cost and irradiation. MRI is used to evaluate spinal canal as well as the presence of spinal cord compression\textsuperscript{36}. Angiogram or CT angiography is important for pre-surgical evaluation so as to seek anatomical variations in the vertebral arteries\textsuperscript{8}.

Surgical treatment in patients with Down syndrome is challenging. These patients have immunologic and collagen abnormalities which predispose them to various surgical complications such as infection, healing problems, instability and bone graft resorption\textsuperscript{8,22,35}. The approach adopted by many authors is related to the fulfillment of surgical treatment in symptomatic patients and careful observation of asymptomatic ones; prophylactic arthrodesis has not been adopted due to symptomatic patients and careful observation of asymptomatic authors is related to the fulfillment of surgical treatment in region and surgical technique accuracy has been described\textsuperscript{8}. Furthermore, surgeries have been carried out in asymptomatic patients with AOD greater than 10 mm\textsuperscript{22}.

Several techniques are used to stabilize atlantoaxial junction, from semi-rigid forms, using grafts and wires / cables, to rigid approach using screws and rods\textsuperscript{1,25}. Although many authors use patient age as a limiting factor (10 years for rigid instrumentation and 36 months for sublaminar wiring), a noteworthy importance to factors such as anatomic size of region and surgical technique accuracy has been described\textsuperscript{8}.

Sublaminar wiring techniques were first described in 1939 by Gallie, consisting of use of grafts between the posterior elements of C1 and C2\textsuperscript{1,25,40}. Despite having lower stability and fusion rates, these techniques have lower risks of neurovascular injury as well as the possibility of use in patients whose dorsal elements of atlantoaxial junction do not allow the use of rigid instrumentation techniques\textsuperscript{1,8,25}.

The most used rigid instrumentation techniques are C1–2 transarticular screw fixation (described by Magerl et al.)\textsuperscript{18}, C1 lateral mass screw and C2 pedicle screw (Harms’ technique)\textsuperscript{13}, C1 lateral mass screw and C2 pars interarticularis screw, C1 lateral mass screw and C2 translaminar screw (described by Wright)\textsuperscript{40}. It is not possible to perform the Magerl’s technique in patients with severe thoracic kyphosis, anatomical variations of vertebral artery and congenital anomalies of the atlas and skull base\textsuperscript{1,11,25}. Harms’ technique has the advantage of being performed in patients with abnormalities in the posterior elements of C2, while Wright’s technique has lower rates of vascular injury, since in the last one the fixation occurs in the lamina of C2\textsuperscript{13,25,40}.

Occipitocervical fusion is indicated in patients who have occipitocervical instability (congenital anomalies, trauma, cancer, rheumatoid arthritis) and in cases that atlantoaxial fusion failed or was contraindicated\textsuperscript{11,25}. Occipitocervical fusion techniques can be carried out only with the use of grafting or through rigid instrumentation (screws, rods and plates), and the level of fusion being dependent on the patient’s clinical and radiological features\textsuperscript{1,25}. In this case we opted for fixation up to C5 level because of the impossibility of instrumentation in the posterior elements of C1 and C2.

Occipitocervical fusion with rigid instrumentation has success rates higher than the ones using only grafts, however such technique must be carried out carefully, since screw placement above the superior nuchal line can lead to subdural and extradural haemorrhage, dural laceration, transverse sinus injury, cerebral spinal fluid leak, among others\textsuperscript{11,15}.

The rib graft offers advantages over the iliac bone as higher regeneration rates, tension forces, bone morphogenetic protein and malleability; in addition to lower morbidity and complications, with fusion rates described up to 98%\textsuperscript{1,25,35}. Autografts have higher tensile strength and lower risk of complications than allografts\textsuperscript{25}. It was not possible to perform autograft in this case due to intraoperative bradycardia, opting for the use of hydroxyapatite. Post-operative immobilization should be carried out for three months in patients who underwent atlantoaxial fusion and for six months in patients who underwent occipital fusion\textsuperscript{44}.

Although patients with Down syndrome frequently present skeletal changes, few of them are symptomatic. Nevertheless, it is important for the doctor to be able to deal with peculiarities that make the treatment challenging for these patients, being either anatomical or derived from the disease itself, in order to provide a better therapeutic behavior and prognosis.
REFERENCES


The authors shows a well documented, yet not so rare, case of C1-C2 subluxation in a patient with Down’s syndrome. Some points adressed at the review should be reinforced: 1. There must be a high level of suspicion since the patients frequently have difficulties in describing their symptoms; 2. Children up to 10 years old often have pedicles or laminas too thin to be screwed; therefore, although only a short fusion (C1-C2 or even occiput-C2) is necessary, extension to the lower cervical spine could be necessary; 3. Grafting is an issue in this situation, and ribs are usually a good option. In the case, due to intraoperative problems, hidroxiapatyte was the option in an emergency situation. Would it be sufficient to accomplish a fusion in long-term follow up? It is impossible to know at this momment. Nevertheless, the authors should be congratulated for the way they conducted the case.

Jerônimo B. Milano