Management of cervical myelopathy due to ossification of posterior longitudinal ligament in a patient with Alström syndrome

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Abstract

Introduction Alström syndrome (AS) is a rare autosomal recessive genetic disorder with multisystemic involvement characterised by early blindness, hearing loss, obesity, insulin resistance, diabetes mellitus, dilated cardiomyopathy, and progressive hepatic and renal dysfunction. The clinical features, time of onset and severity can vary greatly among different patients. Many of the phenotypes are often not present in infancy but develop throughout childhood and adolescence. Recessively inherited mutations in *ALMS1* gene are considered to be responsible for the

causation of AS. Musculoskeletal manifestations including scoliosis and kyphosis have been previously described.

Case report Here, we present a patient with AS who presented with cervical myelopathy due to extensive flowing ossification of the anterior and posterior longitudinal ligaments of the cervical spine resulting in cervical spinal cord compression. The presence of an auto-fused spine in an acceptable sagittal alignment, in the background of a constellation of medical comorbidities, which necessitated a less morbid surgical approach, favored a posterior cervical laminectomy decompression in this patient. Postoperatively, the patient showed significant neurological recovery with improved function. Follow-up MRI showed substantial enlargement of the spinal canal with improved space available for the spinal cord. The rarity of the syndrome, cervical myelopathy due to ossified posterior longitudinal ligament as a disease phenotype and the treatment considerations for performing a posterior cervical decompression have been discussed in this Grand Rounds' case presentation.

Keywords Alström syndrome · Cervical · Myelopathy · Laminectomy

Case presentation

A 47-year-old gentleman with a diagnosis of Alström syndrome (AS) presented to us with progressive worsening of function in his hands and difficulty in walking. He started developing difficulty in using his hands for fine motor functions about a year ago before he first presented to us. This was associated with neck pain with radicular symptoms down his upper limbs. He also had gait disturbances with progressive imbalance while walking. Some of

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these gait-related symptoms have initially been attributed to his sensorineural deafness and arthritic hip joints, which are features of the AS.

The patient had been diagnosed with retinitis pigmentosa during his childhood when he developed progressive reduction in visual acuity. Subsequent to progressive retinal dystrophy, he soon became blind. He then had developed sensorineural hearing loss, insulin resistance and hyperinsulinemia and type 2 diabetes mellitus during his adolescence. Due to the extreme rarity of the syndrome and lack of awareness among the treating physicians, the syndrome was not diagnosed till his early 40s. When he was 40 years of age, he had developed most of the clinical features of AS, including a fatty liver, recurrent chest infections, renal dysfunction, hypertension and polyarthritis, which are part of the major and minor criteria described for diagnosing AS [1]. Based on this, a diagnosis of AS had been made and currently he is the one of the oldest living patients with AS in United Kingdom. Fortunately he had not developed dilated cardiomyopathy, which is considered as one of the cardinal features of this condition. At the time of presentation, he was actively involved in national level chess competitions, but had been experiencing increasing difficulty with sensation of his fingertips necessary for discerning the chess figures and reading braille.

The patient was short-statured and stocky built with a body mass index of 26. His neck movements were restricted. The motor power in all the myotomes was normal except for weakness in the hand grip. He had patchy non-dermatomal sensory loss below the C5 dermatome. The deep tendon reflexes in the upper and lower limbs were brisk and the plantar response was bilaterally extensor. Hand myelopathic signs including Hoffman's sign, finger escape sign and grip release signs could be elicited in both hands. His gait could not be conclusively classified as myelopathic due to the presence of blindness, deafness and arthritic joints.

Diagnostic imaging section

Based on the clinical presentation, a provisional diagnosis of cervical compressive myelopathy was made and further evaluation with radiographs, computerised tomography (CT) scan and magnetic resonance imaging (MRI) was performed (Figs. 1, 2, 3).

Description about the condition (condition, epidemiology, diagnosis, pathology, differential diagnosis)

Alström syndrome is a rare autosomal recessive disorder and was first described in 1959, by Carl Henry Alström

[2, 3]. It is characterised by multiorgan systemic involvement affecting the vision, hearing, endocrinal, hepatic, renal, gastrointestinal, respiratory, cardiac and musculoskeletal systems [4, 5]. Only fewer than 500 cases of AS have been reported in the literature thus far [1]. Due to the multisystem organ involvement, the life span of affected individuals is usually reduced.

Sensorineural hearing loss and congenital retinal dystrophy are two cardinal features of AS [6]. By the age of 16, at least 90 % of affected individuals are blind [1]. Approximately 80 % of affected individuals will develop bilateral sensorineural hearing loss [4]. This usually occurs at a later age in childhood. These early changes in neurosensory capabilities can have tremendous impact on the social development of the child. Patients with AS often have certain characteristic facial features such as round face, deep-set eyes, thick ears, dental anomalies, hyperostosis frontalis interna, and premature frontal balding [1]. Childhood obesity is present in over 95 % of individuals with AS [4]. AS has several features similar to the common metabolic syndrome such as truncal obesity, insulin resistance, type 2 diabetes and hypertriglyceridemia. Type 2 diabetes is diagnosed in over 80 % of individuals above the age of 16 [1, 4].

Dilated cardiomyopathy (DCM) can occur at any age, but most typically occurs during infancy. It can affect about 60 % of individuals [4]. Dilated cardiomyopathy is the most common underlying cause of death in the infantile period in AS. In addition to cardiac problems, patients can have a variety of respiratory problems, including chronic asthma, sinusitis/bronchitis, alveolar hypoventilation and recurrent pulmonary infections [4]. Approximately 80 % of patients affected with AS may have hepatic involvement, ranging from mild elevation in liver enzymes to fatty liver to cirrhosis with portal hypertension [7]. Renal insufficiency occurs in about 50 % of AS-affected individuals with hypertension being present in approximately 30 % of individuals.

Ossification of posterior longitudinal ligament (OPLL) is a condition characterised by varying degrees of ossification of the posterior longitudinal ligament and is most commonly seen in the cervical spine. It is common among the Asian population with a reported prevalence of 1.9–4.3 % among the Japanese population whereas its prevalence in the Caucasian population has been reported as only 0.12 % [8, 9]. Ossification of posterior longitudinal ligament as a disease phenotype of AS has not been previously reported. Recent studies have shown an increased prevalence of OPLL in patients with various metabolic disorders, including hypoparathyroidism, acromegaly, and diabetes [10]. Considering that patients with AS also have several features typical of a metabolic syndrome (diabetes, obesity, hypertriglyceridemia), the development of OPLL could be

Fig. 1 a, b Antero-posterior and lateral radiographs of the cervical spine show degenerative changes and an extensive flowing ossification of both the anterior and the posterior longitudinal ligament. The

disc spaces have been completely obliterated. The normal lordotic alignment of the cervical spine is reduced. The lordotic angle between C2 and C7 measured 17°

Fig. 2 a–c Due to the presence of extensive ossification, a CT scan of the cervical spine was performed for further evaluation. The sagittal CT scan images reveal that the cervical spine is completely fused from C1 to T1 with complete ossification of both the longitudinal

ligaments. A continuous type of ossified posterior longitudinal ligament is identified with maximum thickness extending from C3 to C7. The axial images at C4 and C5 levels indicate a mushroom type of OPLL

Fig. 3 a–c Magnetic resonance imaging of the cervical spine show extensive cervical spinal cord compression from C3 to C7 due to the OPLL with high signal intensity changes observed in the sagittal and axial T2-weighted images

the result of such metabolic abnormalities. The occurrence of OPLL could also be an integral part of the syndrome in those living more than 40 years. Presumably since this syndrome is associated with a shortened life span, the occurrence of OPLL in these patients at a later stage in their life has not been previously reported. The present patient also had features suggestive of a coexistent diffuse idiopathic skeletal hyperostosis (DISH). The coexistence of DISH in patients with OPLL has been previously reported ranging from 25 to 50 % [11, 12].

Rationale for treatment and evidence-based literature

The presence of multisystem organ damage in AS had several implications while planning the surgical management for this patient. Patients with AS have blindness and sensorineural deafness which can lead to potential communication problems at the time of anaesthetic work up and induction. They need adequate pre-operative counselling

about the possible sequence of events in the operating theatre to avoid anxiety. Furthermore, the patient had a short neck with limited neck extension which resulted in a difficult endotracheal intubation. The patient also had significant medical comorbidities including insulin-dependent diabetes mellitus, an enlarged fatty liver with hepatic dysfunction, renal dysfunction, hypertension and previous history of recurrent chest infections. Hence, optimising the patient's physiological status before surgery is of paramount importance and appropriate perioperative critical care support will be needed for successful outcome in these patients.

Surgical decompression of the spinal cord in OPLL can be performed by either the anterior or the posterior approach and both the approaches have their own merits and disadvantages [13]. In this patient, the cervical cord compression was treated by a cervical laminectomy from C3 to C6 vertebra. Several patient factors, both clinical and radiological, were instrumental in guiding us to select this posterior approach. In the background of various medical

comorbidities, an extensive cervical corpectomy (at least four vertebral levels) would be a major undertaking for this patient. Since these patients are also prone for postoperative chest infections, long surgeries would significantly affect the perioperative care and increase the risk of morbidity and mortality. Though good results have been reported for 1- and 2-level cervical corpectomies for OPLL, corpectomies of 3 levels or more are fraught with complications including graft fracture, graft pistoning, graft dislodgement, instrumentation failure, and pseudarthrosis [14, 15]. Anterior surgery in OPLL is also associated with risks of dural injury leading to wound complications and extended hospital stay. Repair of an anterior durotomy in the cervical spine can be quite challenging, particularly in the presence of calcified dura [16]. The engorged epidural venous plexus may bleed significantly, making a dural repair even more technically challenging. The fear of dural tear can also lead the surgeon to perform an inadequate decompression with resultant continued neurological deterioration.

Selection of either a posterior or a anterior approach should be based on a number of factors including patient age, comorbidities, severity of symptoms, previous surgery, type and extent of OPLL, degree of stenosis, surgeon's preference, and assessment of cervical sagittal alignment [17]. Apart from clinical factors, various radiographic criteria have also been proposed to help select the best approach for patients with cervical myelopathy. Gwinn et al. [18] proposed a simple straight-line method to measure the effective cervical lordosis in patients with cervical myelopathy. Here, a straight line is drawn from the dorsal-caudal aspect of the C-2 vertebral body to the dorsal-caudal aspect of the C-7 vertebral body. Effective lordosis is considered to be maintained if no ventral bone structure such as vertebral body, disc-osteophyte complexes, or hypertrophic calcifications project dorsal to this line. Otherwise, effective lordosis is considered to be lost. Fujiyoshi et al. [19] had proposed another method to plan the surgical approach for cervical OPLL. They introduced a new index called the K-line to help determine the effectiveness of a dorsal approach. This line was defined as a line on a lateral radiograph drawn between the midpoints of the spinal canal at C-2 and C-7. Based on this line, two groups of patients with OPLL were identified. In the K-line (+) group, the OPLL lies ventral to the K-line. In the K-line (−) group, the OPLL passes the line and lies dorsal to the line. In their series, 27 patients with OPLL underwent either laminoplasty or laminectomy and the relationship between the dorsal shift of the spinal cord and the K-line classification was made. They concluded that patients with cervical OPLL that extend dorsal to the K-line have a better chance for neurological improvement with a ventral approach. In the present patient, effective lordosis had been maintained

as by Gwinn's line and he fell in the K-line (+) group and hence it favoured a posterior approach. So based on several clinical factors such as patient morphology, presence of comorbidities, need for multilevel decompression, and radiographic features such as type of OPLL, ossified ALL, extensive nature of disease, radiographic measurements, cervical lordosis and the rigidity of spine, a plan for dorsal approach for cervical decompression was made.

The advantages of dorsal approaches include familiarity, ease of decompression of multiple levels, and the ability to extend the decompression either to the occiput or to the thoracic spine [17]. The overall neurological recovery rate after posterior procedures for OPLL has been reported to be approximately 60 % [20, 21]. The major disadvantage is the disruption of the posterior tension band, which can result in a high rate of postlaminectomy kyphosis especially in patients with preexisting cervical kyphosis [21]. However, this is not the case in the present patient whose cervical lordosis was 17°. We also presumed that the presence of extensive ossification along both the ventral and the dorsal aspects of the vertebral bodies of the cervical spine could also prevent a postlaminectomy kyphosis in this patient.

Procedure

The patient was positioned prone on padded bolsters. The head was fixed in a Mayfield frame and mild flexion was possible at the craniocervical junction. The mean arterial blood pressure was maintained at above 80 mmHg to prevent hypoxic injury to the spinal cord. Standard precautions to prevent injury to the eyes and the brachial plexus were undertaken. A standard midline posterior exposure from C3 to C6 was carried out up to the lamina-facet border taking care to preserve the attachments to C2 and C7. Once the lamina-facet junction was identified, two longitudinal trenches were created from C3 to C6 on both sides using a high speed cutting burr. Care was taken to avoid damage to the facet joints which can predispose to postlaminectomy kyphosis. The inner cortex was thinned using a diamond burr. The interlaminar space between C2–3 and C6–7 was cleared to identify the midline cleft in the ligamentum flavum. A 1-mm Kerrison's rongeur was used to remove the flavum up to the lateral furrows created. The laminectomy was completed by lifting the laminae en bloc from the caudal end and gently dissecting any adhesions between the ligamentum flavum and the dura. The laminae of C2 and C7 were undercut partially. Care was taken to fully decompress the C5 nerve root bilaterally and there was no C5 nerve root palsy postoperatively.

Fig. 4 a–c Postoperative sagittal and axial T2-weighted images show good posterior cervical spinal decompression through a C3–6 laminectomy. Adequate CSF flow is noted both anterior and posterior to the spinal cord

Outcome with documentation

The patient showed progressive neurological recovery with improved hand function and gait. In the last follow-up about 18 months after the surgery, he has improved from a pre-operative Nurick grade of 3–1 but the patient continued to use a wheelchair as primary choice mainly due to the persistent peripheral joint arthritis and blindness. A follow-up MRI scan of the cervical spine was performed which showed adequate cervical spinal cord decompression with improved space available for the cord, good flow of CSF anterior to the spinal cord and a maintained sagittal spinal alignment (Fig. 4).

Conflict of interest None.

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