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Missed diagnosied case with syrngomylia and the overlap presention with muscular disease in a child; Case report

Rana Alshiekh[1], Alqassmi Amal[2]
[1] Pediatric Neurology Associated Consultant, King Saud Medical, Riyadh, Saudi Arabia
[2] Pediatric Neurology Consultant, Head Section of Neurology, King Saud Medical, Saudi Arabia

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Abstract
Syringomyelia can have similarity in presentation with certain neuromuscular disorders. Delaying in diagnosis can lead to progressive pressure on the spinal cord, result in severe irreversible neurologic deficits. In our case 11 year old presented with progressive lower limb weakness over 5 years course, was referred to tertiary hospital as a muscle related cause of lower limb weakness for genetic study miss diagnosied as Muscular dystrophy, Spinal MRI done showed cervical syringomyelia treated accordingly.
Conclusion: The clinical presentation of syringomyelia may overlap with clinical presentation of several neuromuscular diseases. When left unmanaged may cause irreversible spinal cord injury. Establishing the diagnosis with magnetic resonance imaging is important in both reaching diagnosis and directing further surgical management.

Keywords: missed diagnosied case, syrngomylia, muscular disease

Introduction
Lower limb weakness is most common causes of visits to pediatric neurology and is often caused by variety of neuromuscular diseases such as Spinal muscular atrophy, Peripheral neuropathy, chronic inflammatory polyneuropathy, Myasthenia gravis, Duchene muscular dystrophy, and limb-girdle muscular dystrophies, Inflammatory myopathy (Polymyositis and dermatomyositis), Metabolic myopathy. Occasionally, this can be induced by other conditions such as tumors, fracture, syringomyelia and other types of compression or damage to the cervical nerve roots.
Syringomyelia is a rare condition where a cyst-like structure is found in the spinal cord, usually trauma-induced, and is commonly associated with Arnold-Chiari malformation [1].
Syringomyelia is occurred secondary to disturbance of normal cerebrospinal fluid flow inside the spinal cord caused by either acquired abnormality or congenital anomaly. Clinically it can range from asymptomatic to sever neurological impairment with irreversible spinal cord injury based on the location and extend of the underlying syrinx. Given the nature of the clinical course may mimic or misdiagnosed as other neuromuscular disorders [2].
The aim of this study is to emphasis that simple MRI image can identify Syringomyelia and Arnold-Chiari malformation that can be a cause of irreversible neurological deficient if missed and left untreated. It is also important to enhance the awareness of clinicians regarding the important of early detection and appropriate intervention.

Case presentation
11-year-old girl with unremarkable prenatal and post-natal history, she was in her usual state of heath till 5 years ago when she started to complain of progressive proximal muscle weakness of lower limb associated with pain mainly in the lower thigh just above knee increased with long distance walking and relieved partially by sitting. Her activity was limited during the day due to pain, as the time pass she requires assistance even when going to bathroom. Her upper extremity was spared.
Her family sought medical advice multiple time in general pediatrics at their local area since age of 5 year and did lower limb X-ray to rule out fractures, their impression was low vit D and was received multiple course of vit D replace therapy without any improvement. As her pain worsen her family brought her to ER at king Saud medical city.
She had positive history of urinary hesitancy and constipation. She has history of frequent headache responding to pain killer. No sensory changes, no loss of sphincter control. Doing well at school .No history of back pain No history of double vision, facial asymmetry nor difficulty in swallowing. No history of trauma, no weight loss nor loss of appetite, no history of recent infection. Positive family history of brother has scoliosis underwent corrective surgery at KFSH and the other brother has abnormal gait as well did not seek medical advice previously, her maternal aunt had cardiac disease following with cardiology.

On Examination
She looks well, alert, oriented to time, place and person. Cranial nerve intact. Normal tone throughout. No muscle atrophy, no muscle fasciculations. Power as measured using the Medical Research Council Scale, were 5/5 in right upper limb. 4/5 in all range of motion in her left shoulder due to pain, 5/5 in left elbow and wrist. She had corresponding weakness in her lower extremities 4- /5 in all range of motion of both hips bilateral (adduction, abduction flexion and extension), 5/5 in knee extension and flexion bilateral, 5/5 in ankle dorsiflexion and plantar flexion eversion and inversion bilateral.

Deep Tendon Reflexes: was brisk in right triceps, and bilateral knee as well as ankle.
There is tenderness over medial thigh bilateral more in the left side.

Gait: Antalgic gait Normal sensory exam, normal back exam and Cerebellum exam.

Examination of other systems revealed no abnormality Complete blood count and biochemical investigations, including thyroid function, ESR, CRP and CK were normal.

Neuroimaging MRI Brain and spin with contrast: Chiari I malformation with syringomyelia (long segment over cervical spine, starting at the lower border of C2 vertebra and ended at the upper border of T8 vertebra with associated cord expansion as well there is cystic component noted at its upper portion at the level of C3 and C4,) and surrounding interstitial edema. Lower lumbar dural ectasia. CT cervical spine without contrast: The vertebral bodies and discs are of normal height. The facet joints have a normal appearance. Straightening of the cervical spine. The craniocervical junction is unremarkable, with normal foramen magnum. The spinal canal is widely patent with normal appearing cord. Fig (1) The patient subsequently underwent endonasal approach for C1 anterior laminectomy with odontoiectomy and clival drilling, 3 days later she underwent second procedure posterior approach for occiptocervical fusion, C1 laminectomy, C2and C3 pas screws with aid of neurophysiology and navigation.

The patient subsequently underwent suboccipital craniectomy with C1 laminectomy and C3 through C5 laminectomy to enable craniocervical decompression of the syrinx.

Postoperatively, she was fully engaged in comprehensive rehabilitation program, with steady improvements in independence with ambulation at the community level, and independence in activities of daily living

Follow-up MRI revealed a reduction in syrinx size, see Figure 2. (Will be up loaded)

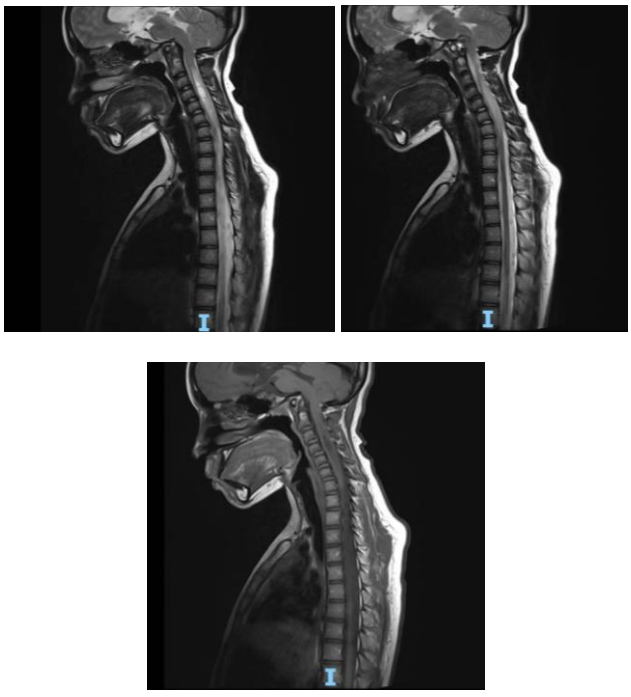

Fig 1: MRI T2 and T1 sagital. MRI Brain and spin with contrast: Chiari I malformation with syringomyelia (long segment over cervical spine, starting at the lower border of C2 vertebra and ended at the upper border of T8 vertebra with associated cord expansion as well there is cystic component noted at its upper portion at the level of C3 and C4,) and surrounding interstitial edema. Lower lumbar dural ectasia

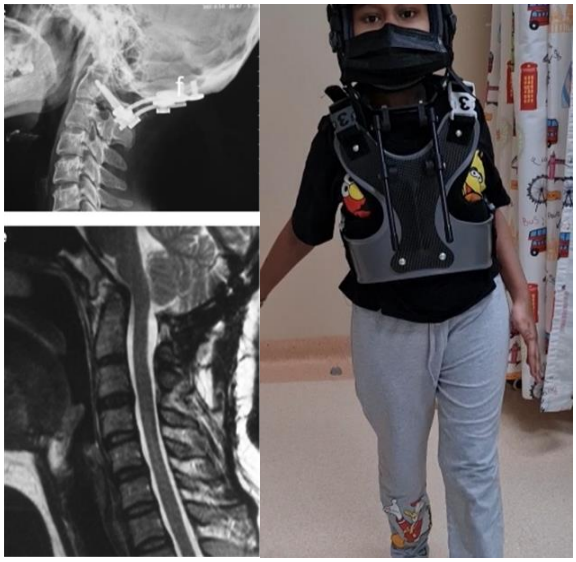

Fig 2: (a) CT scan showe fixiation of cervical bone, (b) Mri brain after correction of syryngomylia © patient photo after fixiation.

Discussion

Syringomyelia is terminology referred to any fluid-filled cyst (syrinx) within spinal cord or dilatation of central canal. Over time, the cyst can enlarge, leading to irreversible damaging to spinal cord and causing pain, weakness and stiffness, among other symptoms [3].

Epidemiological data on syringomyelia is limited, No clear data about epidemiology in Saudi Arabia. However, Estimated prevalence is about 8.4 cases per 100,000 people being males are predominant. The disease mostly occurred in the third or fourth decade of life, with a mean age of onset of 30 years. Syringomyelia development in childhood or late adulthoods rare [4, 5].

The pathophysiology of syringomyelia is still debated, and numerous reviews on the subject are available [6, 7].

In the context of absence of an identifiable cause the syrinx is called Idiopathic Syringomyelia, while if there is an identifiable cause called secondary.

The secondary syringomyelia include: post infectious, post inflammatory, Post-subarachnoid hemorrhage, post-operative or traumatic.

Any cord compression by spinal cord tumors such as hemangioblastoma, herniated disc and spondylosis. Clinical presentation of syringomyelia vary depending upon the location and size of syrinx. The course of syringomyelia is unpredictable may fluctuate between period of stability and progression. Most of the patients with syringomyelia presents with sensory symptoms such as pain and temperature insensitivity, and less common symptoms is muscle weakness and wasting, in most cases it is an incidental finding [8, 9]. The progression of the disease is gradual and slow over years [10]. Sudden onset of symptoms in previously asymptomatic patient may occurred secondary to bouts of coughing or sudden head movement which may be explained by increase in tonsillar descent [11, 12].

The incidence of syringomyelia increased recently in the presence of advanced neuroimaging that made it easy to recognized by routine brain and spine MRI for evaluation of neurological manifestation. The early recognition and early management may contribute in preventing irreversible

neurological damage [9].

Our patient presented with five years course of slowly progressive lower limb weakness visited neurology clinic multiple time with provisional diagnosis of neuromuscular disease Limb-Gridle muscular dystrophy and referred to physical therapy which led to delay of the diagnosis and further progressive of her symptoms affecting her daily activity, later was referred to our hospital for genetic study and further counseling. Further work-up with simple imaging techniques easily confirmed the etiology of her symptoms, fortunately she underwent urgent surgery to relieve the pressure on spinal cord with significant improvement of her symptoms. The pathogenesis of syringomyelia is not fully understood multiple theories was proposed. Given the vague manifestation of the disease makes it challenging for early recognition. Moreover, once diagnosis is established the management strategy and surgical approach is challenging. Hence, increasing awareness about the variability of the presenting symptoms and the nature of the diseases among the clinician in different specialty is crucial as may lead to sever debilitation in misdiagnosed cases.

Conclusions

Syringomyelia presentation can mimic variety of neuromuscular diseases. Early diagnosis with MRI and providing appropriate treatment is important to prevent serious irreversible neurological disability resulting from spinal cord compression.

Consent

Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

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