

Anterior Horn Cell Disease - A Rare Case Report.

Nita R. Sutay¹, Devanand Chaudhari², Prachi Atmapoojya³, Md. Sabri Ahsan⁴, Yugesh Agrawal⁵

¹Professor and Head, Department Of Paediatrics, Grant Govt. Medical College and Sir JJ Hospital, Mumbai.

²Assistant Professor, Department Of Paediatrics, Grant Govt. Medical College and Sir JJ Hospital, Mumbai

^{3,4,5}Resident, Department Of Paediatrics, Grant Govt. Medical College and Sir JJ Hospital, Mumbai.

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ABSTRACT

Motor neuron diseases (MND) are heterogeneous group of neurologic disorders characterized by progressive degeneration of motor neurons. The broad classification of MNDs is done on the basis of whether upper or lower motor neurons or a combination of both are affected. Anterior horn cell disease is characterized predominantly by lower motor neuron affection. Since anterior horn cells of spinal cord contain motor neurons which primarily affect axial muscles, usually the first presentation of anterior horn cell disease is muscle weakness. Since the anterior horn cells are somatotopically organized in the spinal cord, diseases that affect anterior horn cells can cause highly selective affection of the muscles. Group of muscles, individual muscle or even portion of a muscle can be affected in anterior horn cell disease. Various disorders in which anterior horn cells of the spinal cord are affected include Amyotrophic lateral sclerosis, Spinal muscular atrophy, Poliomyelitis/Non Polio like illness, Progressive muscular atrophy, Werdnig- Hoffmann disease and Charcot-Marie-Tooth disease. These diseases have different etiopathogenesis but in all these conditions anterior horn cells of the spinal cord are affected in strikingly similar pattern. The diagnosis is usually confirmed by Imaging (Magnetic resonance imaging of spinal cord) in patients having signs and symptoms consistent with anterior horn cell disease. A 11 year old female was admitted with sudden onset weakness of both upper limbs. There was a history of tingling and numbness in hands since 1 month. On examination the child was having bilateral extensor weakness of upper limbs along with hyperesthesia over C3-C6 dermatome. On examination there was involvement of all groups of muscles of both upper limbs. Weakness was severe in extensor group of muscles. Deep tendon reflexes of upper limbs were depressed. There was no weakness in lower limbs. Sensory examination revealed hyperesthesia over shoulder, nape of neck, upper chest and whole of upper limb. The diagnosis of anterior horn cell disease was made on the basis of imaging findings. There was typical owl's eye appearance of cervical spine on MRI. Steroids and supportive treatment was given. Patient responded well to steroids and there was gradual decrease in weakness. With eradication of poliomyelitis from India anterior horn cell disease is one of the important differential diagnoses of acute flaccid paralysis. The first presentation is weakness of axial muscles. Diagnosis usually is done on the basis of typical clinical features in association with typical imaging findings.

Keywords: Anterior horn cell disease, Motor neuron disease, Muscle Weakness, Magnetic resonance imaging.

INTRODUCTION

Motor neuron disorders (MNDs) are pathologically heterogeneous group of neurologic disorders characterized by progressive degeneration of motor neurons. In this group of neurological disorders either upper motor neurons (UMN) or lower motor neurons (LMN) are predominantly affected. In some cases both UMN and LMN are simultaneously affected. These disorders are usually uncommon with an incidence of approximately 2 cases per 100,000 populations.^[1] The incidence of MNDs is reported to be increasing but it may be due to increased rate of diagnosis because of availability of superior imaging techniques, increased awareness of this entity in general population and treating physician. Part of the increased incidence may also be due to increase in aging population. As the incidence of degenerative diseases increases with advancement in age more people in their 60s and 70s means more people are prone for developing MNDs.^[2]

Name & Address of Corresponding Author

Dr. Prachi Atmapoojya
Department Of Paediatrics , Grant Govt. Medical College
and Sir JJ Hospital, Mumbai.
Maharashtra India.

In anterior horn cell disease there are degenerative changes in the affected motor neurons namely anterior horn cells of the spinal cord.^[3] In some cases mixed features of upper and lower motor neurons affection may be seen. The exact cause of this disease is not known however many genetic and environmental factors are thought to be important in etiopathogenesis. An example is alterations in the survival motor neuron gene (SMN) which is responsible for spinal muscular atrophy.^[4] This gene is responsible for proper functioning and stabilization of the neuronal population. Mutations or absence of SMN gene causes continued apoptosis

of neuronal cell causing spinal muscular atrophy (A form of motor neuron disease). Since anterior horn cells of spinal cord contain motor neurons which primarily related to motor function of axial muscles, the usual first presentation of anterior horn cell disease is muscle weakness.^[5] The muscles are affected in heterogeneous fashion depending upon the severity and extent of neuronal involvement.

We present here a case of 11 year old female who presented with sudden onset of weakness in both upper limbs. There was also sensory involvement in the form of tingling and numbness since 1 month. EMG/NCV already done 2 days before (outside report) showed motor polyneuropathy predominantly involving left upper limb. Magnetic resonance imaging of cervical spine showed typical features of anterior horn cell disease.

CASE REPORT

An 11-year female came with history of tingling sensation of both upper limb extending to nape of neck and upper chest since 1 month. This was followed by weakness of both Upper limbs (Rt>Lt), manifesting in the form of not being able to raise

hands overhead and inability to hold pen while writing. There was no h/o trauma, fever, difficulty in walking or bladder involvement. On examination power was reduced and deep tendon reflexes were depressed in both upper limbs. Sensory examination revealed presence of hyperesthesia over shoulder, nape of neck, upper chest and whole of upper limbs corresponding to C4 to C6 dermatomes. Lateral spinothalamic pathway sensations (Pain and temperature) were intact. There was no s/o cerebellar or meningeal involvement. Rest of the systemic examination was normal. A complete blood count showed mild lymphocytosis. CSF examination was normal and CPK-MB was also within normal limits. Neuroimaging in the form of Computed tomography and Magnetic resonance imaging was done. CT brain showed Small linear hypodense areas seen in cord extending from lower border of C4 to upper border of C6, s/o syrinx and MRI spine showed features suggestive of anterior horn cell disease in the form of altered signal intensity in cervical cord in the region of anterior horn cells extending from C4 to C6 vertebral bodies (owls eye appearance) [Figure 1].

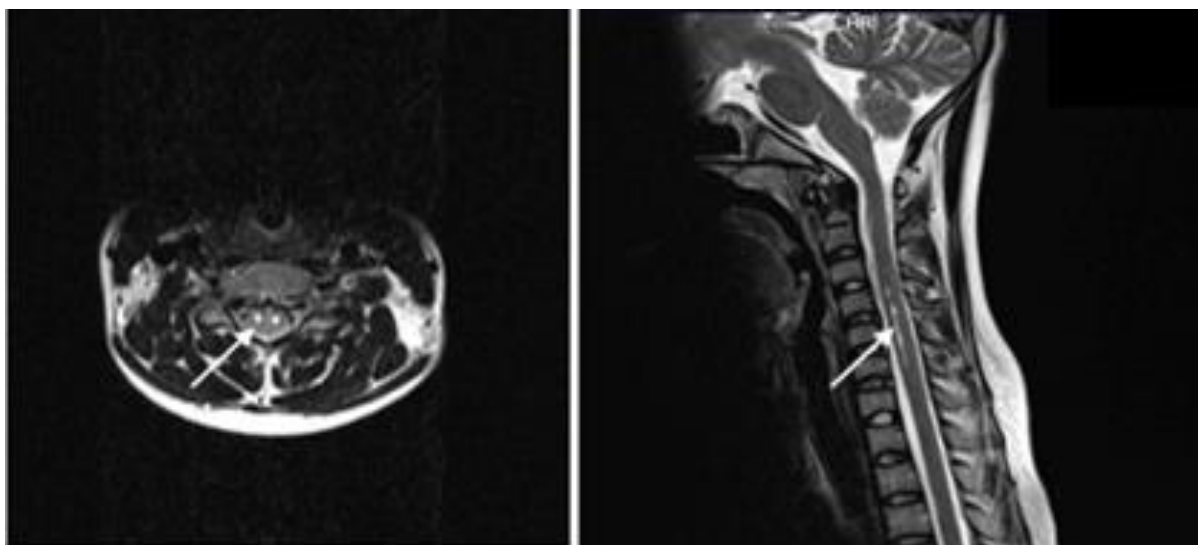


Figure 1: Axial and coronal section of MRI spine. In Axial T1W images there was presence of typical owl eye appearance (Left) T2 FLAIR sagittal images showing demyelination with enhancement (Right)

EMG/NCV was normal. CSF culture for enterovirus and stool culture which was done as a part of AFP reporting was also normal (No growth). On the basis of history, clinical features and imaging findings a final diagnosis of Anterior horn cells disease with bilateral brachial plexus weakness was made. Intravenous steroids (dexamethasone) and physiotherapy to affected muscle was started in addition to supportive treatment. Child improved symptomatically but slight extensor weakness of left wrist was remained. Child was discharged with advice to follow up after 2 weeks and to continue physiotherapy.

DISCUSSION

MNDs are clinically heterogeneous group of disorders with a wide spectrum of affection depending upon the site and severity of involvement. Various disorders included in MNDs are Amyotrophic lateral sclerosis (ALS), Primary lateral sclerosis (PLS), Hereditary spastic paraparesis (HSP), Progressive bulbar palsy (PBP), Spinal muscular atrophy (SMA), X-linked spinobulbar muscular atrophy (SBMA; Kennedy disease), Post-polio syndrome (PPS) and anterior horn cell disease. The incidence of MNDs is variable but reported to be in the range of 2 per 100, 000 population.^[6] Sex

predilection also depends upon the type of disorder since in some cases there is male predilection but such a predilection is not seen in familial cases. Etiopathogenesis is not clear but thought to be multifactorial.^[6] The manifestations are triggered due to complex interactions between genetic and environmental factors. Increasing age predisposes individuals for development of MNDs. Of various theories proposed for development of MNDs the key theory consist of glutamate induced excitotoxicity. This excitotoxicity is thought to be responsible for disruption of intracellular calcium homeostasis, resulting in motor neuron death.^[7] Another theory having current supportive data suggests mitochondrial dysfunction causing oxidative stress to cause abnormal neurodegeneration via calcium-mediated motor neuron injury.^[8] Environmental factors responsible for MNDs are thought to be various metals like inorganic mercury. Many studies have suggested role of metals in development of motor neuron disease. A study conducted by Roger Pamphlet showed that heavy metals in neurons were found in significantly more MND patients than in controls. They further showed that the majority of MND patients had heavy metals in both locus ceruleus and spinal motor neurons.^[9]

Anterior horn cell disease is a type of MND primarily affecting axons of the anterior horn of spinal cord. Since the axons of the ventral portion of the spinal cord are concerned with motor function of the skeletal muscle first symptom of this disease usually is weakness of the corresponding muscles. The disease affecting anterior horn cells of the spinal cord may result in highly selective weakness of the group of muscles.

The diagnosis is based upon clinical features and characteristic imaging findings. Nerve conduction velocity is usually normal as the pathological process involves axons and myelin is mostly unaffected. EMG shows characteristics of neuropathic disorders showing muscle denervation. Neuroimaging is important part of diagnostic workup of the patients and may show characteristic Owl's eye appearance in the spinal cord. This owl's eye appearance (an abnormal intra medullary T2 hyper intensity in the area of anterior horn cells of spinal cord, as two small white dots, one in each half of cord on axial MRI images in the background of normal gray spinal cord) is not exclusive to anterior horn cell disease and may also be seen in Hopkins syndrome, radiation myelopathy and resolved cord contusion. But in combination with typical clinical features this particular appearance of spinal cord on axial sections is highly suggestive of anterior horn cell disease.^[10]

Treatment usually consists of steroids and supportive management. No definitive treatment is available for these devastating disorders. Recent treatment modalities which are being investigated are stem cell transplantation and gene therapy although many questions remain about the utility of these recent therapies.

CONCLUSION

With eradication of poliomyelitis from India the cases of non-polio acute flaccid paralysis are being increasingly identified and reported. Anterior horn cell disease is one such cause of acute flaccid paralysis. It should always be kept in mind while dealing with pediatric patients presenting with acute onset of muscle weakness. Characteristic imaging features on MRI usually confirms the diagnosis.

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