Monomelic MND

Introduction

Monomelic amyotrophy (MMA) is characterized by progressive degeneration and loss of motor neurons, the nerve cells in the brain and spinal cord that are responsible for controlling voluntary muscles (skeletal muscles).

It is characterized by weakness and wasting *in a single limb, usually an arm and hand rather than a foot and leg.*

There is no pain associated with MMA.

MMA mostly affects muscles innervated by C7-T1 spinal segments are most severely affected with relative ***sparing of brachioradialis****.*

While some physicians contend that mild sensory loss may be associated with this disease, many experts suggest that such symptoms actually indicate a cause other than MMA.

MMA occurs in males between the ages of 15 and 25. Onset and progression are slow. MMA is seen most frequently in Asia, particularly in Japan and India.

In 1984 Mandavilli Gourie-Devi (et al) introduced the term "monomelic amyotrophy"

It is much less common in North America. In most cases, the cause is unknown, although there have been a few published reports linking MMA to traumatic or radiation injury. There are also familial forms of MMA.

The symptoms of MMA usually progress slowly for one to two years before reaching a plateau, and then remain stable for many years. Disability is generally slight. Rarely, the weakness progresses to the opposite limb. There is also a slowly progressive variant of MMA known as O'Sullivan-McLeod syndrome, which only affects the small muscles of the hand and forearm and has a slowly progressive course.

Clinical presentation and examination of MMA

History

- Young ,commonly male patient presenting
- Wasting of muscles
- weakness
- Fasciculations in one of either sided upper limb
- Very rarely both upper limbs may be affected –'BAD' –Bibrachial amyotropy
- Lower limb is spared commonly.
- Activities of daily living may or may not be affected in spite of the wasting or weakness.
- No sensory complains –however if any , DD should be considered (listed later)
- No autonomic disturbances.
- Follow up usually show no deterioration or if any very slow.
- Any history of Poliomyelitis in childhood

Clinical examination:

- Wasting in upper limb ulnar distribution muscles –common.
- Other muscles may also be affected in upper limb.
- Thorough clinical examination to see for facial muscles(to R/O Facioscapulo humeral dystrophy
- Abscence of sensory signs
- Combination of UMN & LMN(eg absent jerk with Hoffman positive.

Investigations

Diagnosis is made mainly by physical examination and medical history.

- Electromyography (EMG), a special recording technique that detects electrical activity in muscles, shows a loss of the nerve supply, or denervation, in the affected limb
- MRI and CT scans may show muscle atrophy.
- MRI of cervical spine to r/o Cervical melopathy.
- People believed to have MMA should be followed by a neuromuscular disease specialist for a number of months to make certain that no signs of other motor neuron diseases develop.
- Blood for riboflavin level

Course

Benign monomelic amyotrophy (BMA) is **a rare condition in which neurogenic amyotrophy is restricted either to the upper or to the lower limb**. BMA is usually sporadic, it has an insidious onset and slow progression followed by stabilization in 2-4 years. It is found mostly in young adults.They just nee follow ups ad no treatment.

Variant

Madras motor neuron disease

Madras motor neuron disease (MMND) is **characterized by weakness and atrophy of limbs, multiple lower cranial nerve palsies and sensorineural hearing loss** .
Epidemiology. Less than 200 cases have been reported to date, predominantly from Southern India. Hence the name.

There is a considerable overlap in the phenotype of MMND with Brown–Vialetto–Van Laere syndrome (BVVL) Boltshauser syndrome, Nathalie syndrome and Fazio–Londe syndrome.

Recently a number of BVVL cases and families have been described with mutations in two riboflavin transporter genes SLC52A2 and SLC52A3 (solute carrier family 52, riboflavin transporter, member 2 and 3 respectively).

Differential diagnosis

1] Hirayama disease

Hirayama disease, also known as monomelic amyotrophy (MMA), is a rare cervical myelopathy that manifests itself as a self-limited, asymmetrical, slowly progressive atrophic weakness of the forearms and hands predominantly in young males.

The forward displacement of the posterior dura of the lower cervical dural canal during neck flexion has been postulated to lead to lower cervical cord atrophy with asymmetric flattening.

It mainly develops in the late teens and early twenties with a male preponderance. The typical clinical features include insidious onset and slow progression of unilateral or bilateral muscular atrophy with weakness of the forearms and hands.

Sensory disturbance, autonomic involvement, and upper motor neuron (UMN) signs like hyperreflexia and hypertonia are rare.However minipolymyoclonus may be observed

The motor neuron disease (MND) is a very close differential diagnosis of HD, *but, unlike MND, the disease progresses initially and is followed by spontaneous arrest several years after the onset*.

This disease is more prevalent in Japan and other Asian countries, but cases have been reported from other parts of the world as well.

As "Hirayama *disease*" is considered a self-limited disease and **often stops progressing after 1–5 years of onset**, the mainstay of treatment consists of preventing neck flexion using a cervical collar to halt further progression.

2] Split hand disease(SHD)

The term split hand, first coined by Willbourn et al, refers to **a type of hand muscle atrophy in which the muscles of the lateral aspect of the hand** (first dorsal interosseus and thenar) are more affected than those of the medial aspect (hypothenar).

The physiological mechanisms underlying the split hand in ALS are incompletely understood but both cortical and spinal/peripheral mechanisms are probably involved. Motor potentials evoked by magnetic stimulation are significantly smaller when recorded from the thenar complex, compared with the hypothenar muscles, supporting a cortical mechanism.

A slow onset and a **lack of pain** or sensorial symptoms are arguments against a lesion of the spinal root or plexus brachialis. To an extent, these features can also be seen in normal aging (although technically, the apparent muscle wasting is sarcopenia rather than atrophy).

3]Ulnar neuropathy and SHD.

Ulnar nerve entrapment can cause **pain, numbness and tingling in the forearm and the fourth and fifth fingers**.

Symptoms of ulnar nerve neuropathy may include:

- Weakness or tenderness in the hand.
- Tingling in the palm and fourth and fifth fingers.
- Sensitivity to cold.
- Tenderness in the elbow joint.
- Cervical muscles are involved in SHD but not in pure ulnar neuropathy
- USG of elbow is an important tool to check the ulnar nerve entrpement

4] Kennedy's disease

Kennedy's disease is a **form of motor neuron disease**, and therefore may appear clinically similar to amyotrophic lateral sclerosis (ALS / Lou Gehrig's disease). Kennedy disease is named **after William R. Kennedy, MD**, who described this condition in an abstract in 1966 and a full report in 1968.

Lou Gehrig's disease – a form of ALS with predominant involvement of lower limbs.

Early symptoms include **tremor of the outstretched hands, muscle cramps with exertion**, and fasciculations (fleeting muscle twitches visible under the skin). Eventually, individuals develop limb weakness which usually begins in the pelvic or shoulder regions.

Currently there is no known cure for Kennedy's disease. Treatment is symptomatic and supportive. Physical therapy and rehabilitation to slow muscle weakness and atrophy may prove helpful. Currently there is no known cure for Kennedy's disease.

5] Parsonage Turner Syndrome

PTS is an idiopathic brachial plexopathy or neuralgic amyotrophy, a rare disorder consisting of a complex constellation of symptoms with abrupt onset of shoulder pain, usually unilaterally, followed by progressive neurologic deficits of motor weakness, dysesthesias, and numbness.

Although the etiology of the syndrome is unclear, it is reported in various clinical situations, including postoperatively, postinfectious, posttraumatic, and postvaccination(Vaccines, including certain ones for the flu and COVID-19, have been **tied to some cases of Parsonage-Turner syndrome**. A health agency in France says a small number of PTS cases have been reported after people got the Pfizer or Moderna vaccines to prevent COVID-19).

The identification of the syndrome in the postoperative patient remains a challenge as symptoms may easily be attributed to sequelae of surgical positioning, postoperative recovery, or postanesthetic block pain.

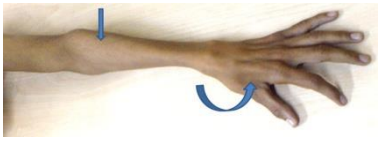
Treatment

- At present **there is no cure for MMA**.
- The impact on the affected individual ranges from minimal to significant depending on the extent of the weakness.
- Physical and occupational therapies include muscle strengthening exercises and training in hand coordination.

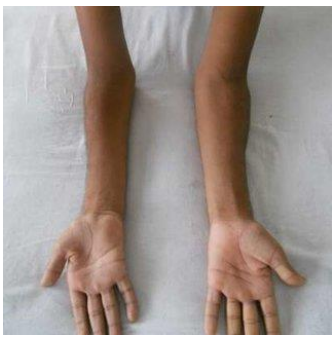
- Wearing of a cervical collar has been recommended in certain studies.
- As the disease is mainly stationary there is no US-FDA approval of riluzole in MMA as in ALS.
- Trials therefore with riluzole, subjecting the patient to unnecessary long term hepatic complications has not yet gained enough ground.

Summary

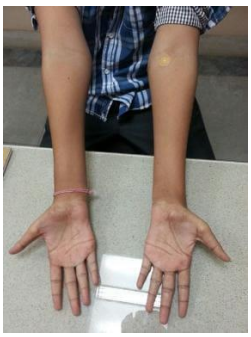
- Monomelic MND (MMA), or atrophy of a single limb (upper much commoner than lower limbs).
- Very rarely both upper limbs may be involved –BAD (bibrachial amytrophic dystrophy)
- History and Clinical examination gives the diagnosis –Always check for intact *Brachioradialis.*
- Vriants and DD to be always thought of.
- Not many investigations requied –EMG is th main, though MRI of cercicalspine, Riboflavin level.
- MRI to rule out cervical involment as welas in certain DD may be done. Of similar help is Magnetoencephalography
- USG of affected muscles may be useful
- A of date, the disease is benign and may initially progress bu becomes static in 3- 5 years
- Hence occupational therapy is enough rather than conventional pharmacological treatment.



Single limb wasting (Especially 1st DI)



Sparing of brachioradialis



Wasting of upper limbs