

## MAIN FOCAL MUSCULAR ATROPHIES OF MEDICAL AND SURGICAL INTEREST

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*[Principali atrofie muscolari focali di interesse medico-chirurgico]*

### ABSTRACT

Focal muscular atrophies are common neurological diseases that require special diagnostic attention by the neurologist and neurophysiologist; the causes are many and they can express localized or systemic diseases, and be stable or progressive, acquired or genetically determined.

**Key words:** Atrophy, muscular atrophy, myogenic atrophy, neurogenic atrophy.

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### Introduction

Focal muscular atrophies are commonly observed neurological conditions that require particular diagnostic attention for the neurologist and neurophysiologist. Its frequency is undefined.

The causes are many and, at times, not even neurological; they can express localized or systemic diseases, be stable or progressive, acquired or genetically determined.

*The main etiologies include:*

- traumas
- inflammation
- compression syndromes
- spinal diseases
- genetic diseases
- vasculitis
- autoimmune diseases
- infections
- metabolic diseases
- malformations
- tumors
- endocrine diseases
- paraneoplastic syndromes.

Many diseases that cause focal muscular atrophies do not reduce the length of life, except for the

forms with an evolutionary trend. There is no particular preference of gender, except for Kennedy's disease and monomelic amyotrophy in which there is a prevalence among males.

Spinal muscular atrophy typically occurs with an atrophy whose time of onset is variable, with weakness in the atrophic muscle group.

Atrophy may affect both the upper and lower limbs distally and proximally.

*The symptoms are characterized by:*

- fibrillations and/or fasciculations
- cramps
- paresthesias (if matched by suffering with sensory fibers)
  - systemic symptoms (for systemic diseases, tumors, metabolic or endocrine diseases)
  - familiarity (familial or environmental diseases due to exposure to common toxic factors).

Apart from the most unusual or rare varieties, the most frequent forms can be attributed to:

- disuse atrophies (forced immobilization, fractures, plasters)
- traumatic injuries (nerves or plexuses)
- mononeuropathies due to diabetes or other causes<sup>(1)</sup>, vasculitis, compression, infections (polio)
- tumors

- syringomyelia
- genetic diseases.

We will describe the most salient varieties, bearing in mind that a large number of other diseases may lie at the root of focal atrophy.

### **Post-polio syndrome (PPS)**

Symptoms occur decades (at least 15 years) after the acute phase of the disease in 40% of the survivors of the infection, although in some cases probably the acute phase goes unrecognized.

The pathogenesis is still controversial: the possible reactivation of the virus, the rarefaction of the motor neuron pool of the anterior horns of the spinal cord, a possible autoimmune reaction of the body, age-related metabolic factors, persistent subclinical infection, new viral infections are considered.

The symptoms are characterized by a marked weakness that may impair the performance of common daily activities, an increase of the atrophic process previously affected by infection, increased fasciculations and cramps, muscle tenderness,<sup>(1,2)</sup> neurophysiological evidence of acute and chronic pathological spontaneous activity in the same muscles affected by the acute phase of the disease or in other locations, much slower progression compared to the pseudo-polyneuritic variant of amyotrophic lateral sclerosis, with which it is involved in the differential diagnosis and from which it distinguishes itself for:

- Earlier age
- Prevalence in women
- No bulbar signs and symptoms
- No pyramidal signs and symptoms
- Presence of pain
- Slower progression
- Focal distribution of lesions.

### **Hirayama disease (monomelic amyotrophy)**

The disease is named after the author of the first description in 1959 in Japan, characterized by a slow and progressive distal atrophy of an upper limb of a young male: progression is very slow and has a tendency to stabilize.

The pathogenesis is not defined: degenerative, infectious or ischemic factors were considered.

The prevailing hypothesis is that it is a focal ischemic myelopathy due to compression of the roots of the anterior horns of the spinal cord in the lower cervical tract following the dynamic changes

that the extradural space of the neck undergoes during the flexion movements, as it is possible to document with MRI with contrast medium, due to laxity of the epidural posterior ligaments of cervical spine<sup>(3,4)</sup>.

Though considered by some a focal variant of SMA, no genetic alteration typical of SMA has ever been documented, including that relating to the Survival Motor Neuron.

Familial recurrence is rare and the symptoms are characterized by:

- Localization with a single upper limb
- Slow progression with a tendency to stabilize
- Absence of sensory, bulbar or 1st motor neuron signs
  - Involvement of the C7-T1 motor roots
  - Typical “oblique atrophy” due to the integrity of the brachioradial spine (C5-C6)
  - Electrophysiological evidence of distal focal muscular atrophy, without progression and with little functional limitation.

### **Focal muscular atrophies (FMA)**

The myogenic causes of focal atrophy may be due to facioscapulohumeral muscular dystrophy, limb-girdle muscular dystrophy (with involvement of the upper portion of the deltoid and integrity of the lower portion), myotonic dystrophy (with atrophy of the temporal muscles, masseters and neck muscles), iatrogenic fibroses caused by repeated intramuscular injections and prevalent in the deltoids or gluteal muscles<sup>(5)</sup>.

### **Axonal multifocal motor neuropathy (AMMN)**

It is a chronic asymmetric motor neuropathy, present for at least 3 years, in the absence of bulbar signs, sensitive to immunomodulatory therapy, without signs of electrophysiological conduction block or major delay in motor nerve conduction velocity, no signs of involvement of the 1st motor neuron, absence of radiation therapy with negative cervical MRI, negative anti-GM1 antibody tests and no electrophysiological signs of sensory suffering (SSPE and VCS) with typical lack of F waves in the ulnar nerves<sup>(6,7)</sup>.

### **Thoracic outlet syndrome (TOS)**

Thoracic Outlet syndrome (TOS) is a condition of unilateral neurovascular suffering, due to

compression of the lower cervical plexus or subclavian-axillary arterial trunk at the thoracic outlet. The typical picture is characterized by an atrophy of the intrinsic muscles of the hand due to the lesion of the ulnar and median nerve and a dysfunction of the medial nerve of the forearm, without any sign of compression<sup>(8)</sup>. The treatment of thoracic outlet syndrome requires particular skill, as it also comprises the simultaneous application of several mini-invasive techniques<sup>(9,10)</sup>. In mild forms, the literature reports limited benefits from the use of L-acetyl-carnitine<sup>(11,12,13)</sup>.

In conclusion, let us briefly mention the electrophysiological findings that may be useful in differentiating the main occurrences responsible for focal muscular atrophy:

#### 1. motor nerve conduction velocity

- In the event of demyelinating damage: MMN, plexopathy

- In the event of axonal damage: TOS, AMMN, PPS, Hirayama disease

#### 2. Sensory nerve conduction velocity

- Widespread abnormalities: autosomal dominant hereditary bulbar palsy

- Circumscribed abnormalities: TOS, plexopathy

#### 3. F waves

- Abnormalities: MMN, TOS, discopathies

#### 4. EMG

- Widespread acute neurogenic damage: neuropathy, axonal neuropathies (cancer, drugs)

- Widespread chronic neurogenic damage: ALS, SMA, axonal neuropathies, Kennedy's disease

- Focal acute neurogenic damage: focal axonal neuropathy (traumatic)

- Focal chronic neurogenic damage: AMMN, Hirayama disease

- Focal myogenic damage: traumatic or injection myopathy.

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