Myopathy with rods and cores
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INTRODUCTION

Congenital myopathies are muscle disorders with variable clinical presentation. We present a case with characteristic history that was diagnosed late in the life, with typical findings on pathologic specimen.

CASE REPORT

The 65-year-old female presents with worsening weakness, restricting daily activities. Weakness is described as proximal muscle weakness which has been slowly progressive. Duration of symptom is since birth and patient has a history of delayed motor development. Patient also has strong family history of similar symptoms with her mother having scoliosis, weakness and high arch feet. Patient has a daughter who also suffers from proximal muscle weakness. The patient’s daughter has 5 kids and 3 of them have similar weakness. Proximal muscle weakness, scoliosis and high arched feet were evident on examination. EMG/Nerve conduction studies revealed findings consistent with “mild myopathy process”. Right deltoid muscle biopsy confirmed myofiber atrophy and myopathy with cores and rods.

Conclusions: Core-rod myopathy is a relatively rare form of congenital myopathy. It is known by the presence of cores and rods in separate regions of same or different muscle fibers. This case highlights the need to obtain neuromuscular evaluation for patients who present early in life with minimal weakness pattern, which is also slowly progressive. Timely evaluation and diagnosis will allow patients to make informed decision regarding their career choices as well as starting a family.

Key Words: Congenital myopathy, Myopathy with cores and rods

DISCUSSION

Congenital myopathies are a heterogeneous group of primary muscle disorders (1). Clinical presentation is variable and common symptoms include weakness, muscle cramps and gait dysfunction. There are many types of congenital myopathies which are divided into broad categories as follows:

Congenital myopathy with cores

Figure 1A) Gomori trichrome stain, 935 x resolutions. The nemaline rods are well-demystated in this image (shown in arrows).

Figure 1B) Normal muscle biopsy with gomori trichrome stain.

Cores are well-demarcated, round areas in the cytoplasm of muscle fibers, which lack oxidative staining. The cores have decreased adenosine triphosphatase (ATPase) activity but in many cases the ATPase activity is preserved (2). It has further types:

- Central core disease: This is an autosomal inherited muscle disorder, characterized by the presence of cores in muscle fibers. Presentation
placed nuclei in rows in muscle fibers. Its various types are:

### Congenital myopathies

- *Autosomal centronuclear myopathies:* It is characterized by triad of morphological features which are characteristic of DNM2-related CNM: (1) radiating sarcoplasmic strands (RSS); (2) increased numbers of central and internal nuclei; (3) type 1 muscle fiber predominance and hypotrophy (8).
- "Necklace" fiber myopathy: Necklace fibers are characterized by a cytoplasmic basophilic ring deposit, a few micrometers under the sarcolemma membrane that follows the contour of the cell, in which myonuclei are aligned.

### Congenital myopathies with abnormal fiber ratios or sizes

Skeletal muscle is made up of two types of fiber, type 1 and type 2. In this myopathy, patients have abnormal number of type 1 fibers. As a result, they have normal daily functioning but reduced stamina (9).

- **Conventional fiber type disproportion (CFTD)** and congenital neuromuscular disease with uniform type 1 fibers (CNMDU1).

We have described the above case with rare form of myopathy - The Core-rod myopathy. This relatively rare form of congenital myopathy is known by the presence of cores and rods in separate regions of same or different muscle fibers. Clinical onset can range from mild presentation (as in our patient) to fetal akinesia. Most of these myopathies are due to mutations in skeletal muscle Ryanodine receptor (RYR1).

No specific treatment is available for these disorders. Management mostly consists of physical therapy, nutritional support, assisted ventilation (if indicated), and genetic counseling.

This case highlights the need to obtain neuromuscular evaluation for patients who present early in life, with minimal weakness pattern which is also slowly progressive. Timely evaluation and diagnosis will allow patients to make informed decision regarding their career choices as well as starting a family.

### REFERENCES