Introduction

This article includes discussion of fingerprint body myopathy and congenital fingerprint body myopathy. The foregoing terms may include synonyms, similar disorders, variations in usage, and abbreviations.

Overview

Fingerprint body myopathy is a rare muscle disease characterized by weakness and reduced muscle mass and by subsarcolemmal, non-membrane-bound inclusions that are frequently adjacent to mitochondria. The author addresses major concerns of this rare disease and highlights specific case studies.

Key points

• Fingerprint body is a rare congenital myopathy.
• Only 5 cases of fingerprint myopathy have been described to date.
• Fingerprint myopathy is defined by subsarcolemmal inclusions on muscle biopsy; the inclusions have a characteristic lamellar pattern on electron microscopy.
• The genetic basis of fingerprint myopathy is not yet known.
• An increasing number of protein aggregate myopathies are now recognized. The multitude of diverse proteins aggregating within muscle fibers suggests a common pathway of impaired extralysosomal degradation of proteins or defects in sarcomeric development and maturation.

Historical note and terminology

The first case of this ultrastructural myopathy to be described was in a 5-year-old girl with weakness (Engel et al 1972). Four additional children have been described, including 2 half-brothers (Fardeau et al 1976) and twin boys (Curless et al 1978). A similar abnormality was discovered in muscle obtained from a 54-year-old woman who had static muscle weakness from early childhood (Gordon et al 1974) and in 2 adult siblings with proximal weakness from early childhood (Stojkovic et al 2001).

Light microscopy reveals either type 1 predominance or normal pattern, but inclusion bodies are generally not apparent histologically. When they are seen on light microscopy, fingerprint bodies may appear as pale or eosinophilic inclusions on the Gomori trichrome stain (Stojkovic et al 2001). Of the 5 children with congenital weakness (Engel et al 1972; Fardeau et al 1976; Curless et al 1978), 3 had type 1 fiber predominance, 1 was normal, and the histology was not described in the fifth. Using their prior identification of ultrastructural fiber typing (Payne et al 1975), Payne and Curless demonstrated type 1 specificity for the fingerprint inclusions (Payne and Curless 1977).

Identical ultrastructural findings in patients without muscular signs or symptoms were noted in 3 adult patients with myotonic dystrophy (Tome 1973) and in a patient with Marfan syndrome (Jadro-Santel et al 1980). Sengel and Stoeber found fingerprint bodies in the muscle of 5 adults; 2 had emphysema and the others had a progressive myopathy, cardiomyopathy, myotonic dystrophy, or progressive ataxia. These authors also reported fingerprint bodies in a child with a central nervous system degenerative disorder (Sengel and Stoeber 1974). Fingerprint bodies were described in an adult with oculopharyngeal muscular dystrophy. There was no limb extremity weakness, and the biopsy was obtained from a deltoid muscle (Julien et al 1974).
Clinical manifestations

Presentation and course

The patients with congenital presentation have nonprogressive, diffuse weakness that delays walking until the age of 2 to 10 years. Most have significantly reduced muscle mass, and mental retardation (IQs from 45 to 65) is also common. All have a normal creatine kinase level, EMG, and nerve conduction studies. Unlike some of the other congenital myopathies, a severe neonatal form has not been described. Also, in no cases has strength deteriorated to the point of necessitating aggressive orthopedic management. Muscles innervated by the cranial nerves are not affected, and there is no typical dysmorphic appearance. However, the low number of reported cases prevents a more definite outline of the clinical course.

The reports of patients without neuromuscular signs or symptoms, and the rarity of reported cases, raise the possibility that fingerprint body myopathy may not be a distinct congenital myopathy. Although this issue has not been resolved, the 5 children and 1 adult with childhood onset strongly suggest that this ultrastructural finding can be used to counsel families. Unfortunately, the light microscopic findings often do not prompt ultrastructural examination.

Prognosis and complications

Judging by the reports of the 6-year-old twins (Curless et al 1978) and the 54-year-old woman (Gordon et al 1974), the outlook for a long life appears good. Although most of the reported cases have below normal intelligence, more reports are required to verify or dispel the possibility that fingerprint body myopathy is generally associated with cognitive deficits.

Biological basis

Etiology and pathogenesis

The etiology is not known, but reports of affected siblings suggest an inherited basis.

Fingerprint bodies are subsarcolemmal, non-membrane bound inclusions, which are frequently adjacent to mitochondria. Their ultrastructural lamellar pattern produces a fingerprint-like appearance. The bodies usually are juxtaposed to myofibrils. Engel and associates noted no change in the ultrastructure following digestion with ribonuclease or glycerination (Engel et al 1972). Although Sarnat was able to demonstrate the presence of desmin in several other congenital myopathies, a similar study has not been published of patients with fingerprint body myopathy (Sarnat 1992). No postmortem cases are available.

Fingerprint bodies may derive from mitochondria (Curless et al 1978) or be the consequence of abnormal muscle innervation (Engel et al 1972).

Epidemiology

The number of reported cases is insufficient to predict the incidence of this condition. Muscle biopsy is not invariably diagnostic in children with congenital myopathies, and the diagnosis of fingerprint myopathy may well be missed if ultrastructural study is not undertaken.

The two reports of affected siblings strongly suggest an inherited basis. The half-sibling relationship in one family suggests an autosomal dominant pattern.

Differential diagnosis

Fingerprint bodies have been observed in a number of other myopathies, including myotonic dystrophy and oculopharyngeal muscular dystrophy (Tome and Fardeau 1973; Julien et al 1974) as well as in patients suffering from neurodegenerative disorders (Sengel and Stoebner 1974). There are an increasing number of congenital myopathies with excessive protein aggregation, many of which have a defined genetic basis (Goebel and Blaschek 2011). In most cases the disease pathology is underpinned by either abnormal protein catabolism or genetic defects of sarcomeric protein formation and/or aggregation.
Diagnostic workup

The decision to obtain a muscle biopsy should be based on the clinical picture: onset during infancy of a non-progressive weakness with reduced muscle mass and a normal creatine kinase level, EMG, and nerve conduction velocity. Before the biopsy, instructions must be given to maintain a portion of muscle in glutaraldehyde for electron microscopy. If light microscopy reveals either normal tissue or type 1 predominance, an electron microscopy study should be considered.

Management

No treatment is available. It is possible that a wider range of motor and intellectual abilities may be present than the few reports indicate.

Special considerations

Anesthesia

There is a single report of malignant hyperthermia in an adult with fingerprint body myopathy. Pathologic findings in this case also included central cores and Z-line streaming. Malignant hyperthermia precautions are appropriate in patients with fingerprint body myopathy or a suspected congenital myopathy.

References cited


Goebel HH, Blaschek I. Protein aggregation in congenital myopathies. Semin Pediatr Neurol 2011;18(4):272-6. PMID 22172423


Payne CM, Curless RG. Fingerprint inclusions; ultrastructural demonstration of muscle fiber type specificity. J Neurol Sci 1977;31:379-86. PMID 191570


**References especially recommended by the author or editor for general reading.**

**Former authors**

Richard G Curless MD

**ICD and OMIM codes**

**ICD codes**

ICD-9:
Congenital hereditary muscular dystrophy: 359.0

ICD-10:
Congenital hereditary muscular dystrophy: G71.0

**OMIM numbers**

Fingerprint body myopathy: 305550

**Profile**

**Age range of presentation**

0-01 month
01-23 months
02-05 years
06-12 years
13-18 years

**Sex preponderance**

male=female

**Family history**

family history may be obtained

**Heredity**

heredity may be a factor
autosomal dominant

**Population groups selectively affected**

none selectively affected

**Occupation groups selectively affected**

none selectively affected

**Differential diagnosis list**

myotonic dystrophy
oculopharyngeal muscular dystrophy
neurodegenerative disorders

**Associated disorders**

Myotonic dystrophy
Other topics to consider

Reducing body myopathy
Myotonic dystrophy
Ataxia