Periorbital amyloid myoneuropathy: case report

Martha Lilia Tena-Suck¹, Carlos Guillermo Patiño-Camacho², Emmanuel Antonio Luna³ and Francisca Fernández Valverde⁴

¹Departamento de Neuropatología, Instituto Nacional de Neurología y Neurocirugía Manuel Velasco Suárez, México
²Departamento de Patología, Hospital Civil antiguo de Guadalajara Jalisco, México
³Residente de Neurología, Instituto Nacional de Neurología y Neurocirugía Manuel Velasco Suárez, México
⁴Laboratorio de Neuropatología Experimental, Instituto Nacional de Neurología y Neurocirugía Manuel Velasco Suárez, México

Abstract

Periorbital amyloid myopathy is a very rare tumor. A 42 year old female, presented to our hospital with ocular pain, and progressive increase in the volume of the left eye. Brain MRI showed increased density in the periorbital tissue of the left eye. Soft tissue biopsy was performed and periorbital fat with violet indurated areas was observed. Histologically hyperplastic nerve fibers were seen with tank fibrillary hypertrophic hyaline material as well as muscle fibers of the fibrillary material deposition. Fibro fatty tissue infiltrations positive to Congo red staining, also showed eosinophil fibrillary material tanks. Immunohistochemistry of the muscle fibers was positive for GLUT4, dystrophin and nerve fibers were positive for s-100. Deposits were identified in both IgG and kappa chains confirming the diagnosis of periorbital amyloid myopathy. Conclusion: Dystrophin positive come up with the possibility of amyloid to provoke to the muscle fibers with glucose metabolic disturbance.

Introduction

Amyloidosis is a rare group of diseases characterized by extracellular amyloid deposits in different organs [1,2] and is a disorder caused by an alterations in the structure of the proteins. The deposition of these proteins in tissues and organs can affect their normal function [1-3]. There are different types of amyloid, including 25 human and 8 animal fibril proteins [2], such as, transthyretin, apolipoprotein A-1, cystatin C, gelsolin, amyloid beta protein, beta 2-microglobulin, scrapie protein, and islet amyloid polypeptide etc. [3] Clinically amyloidosis has been described as localized or systemic disease, as well as, hereditary or acquired form [1-5]. The extent of amyloid deposit clinically affects organ function [1-3]. The deposition of amyloid can be of different proteins and the most common form of local amyloidosis is the local deposition of monoclonal immunoglobulin light chains produced by B-cell or plasma-cell clones. Chronic inflammatory diseases can also cause localized amyloid light chain amyloidosis [5].

Periorbital and orbital amyloidosis is an extremely uncommon disease that presents with a variety of symptoms, depending on the location and depth of the amyloid deposition [5,6]. Amyloid deposition causes tissue destruction and progressive disease which effects long term survival [3,5]. Early identification and differentiation of the protein depositions is important for the prognosis and treatment [5].

The authors report a rare case of periorbital amyloidosis in a 42-year-old woman, who developed a periorbital mass.

Clinical case

A healthy 42 years old woman over a 3months time period developed progressive left eye pain, diplopia, and headache with enlargement of the left periorbital. Her ophthalmologic examination showed no significant abnormalities and her vision was within normal limits. Cerebral CT scan and MRI was conducted and a left periorbital mass was identified (Figures 1a-1c). Meningioma was the preliminary diagnosis and she was subjected to surgical resection. During the surgical procedure a discrete tumor was not identified. Indurated purplish fibrous lesion affecting soft tissues with fatty infiltration was observed. Histologically observed abundant nerve fibers and skeletal muscle tissue with adipose infiltrated were seen (Figure 2a). Nerves fibers were hypertrophic and showed deposits of homogeneous eosinophil fibrillar material without aspect of fibrosis (Figure 2b). Lymphocytic inflammatory infiltrates and plasma cells were also observed. Some vessels were thickened with the presence of fibrillary material tank (Figure 2c). Hypertrophy of type I and 2 fibers was found, with edema and presence of numerous nerves (Figure 2d), but no fatty infiltrations.

Correspondence to: Dr. Martha Lilia Tena Suck, Departamento de Neuropatología. Instituto Nacional de Neurología y Neurocirugía Manuel Velasco Suárez, México Av. Insurgentes sur no. 3877, colonia la joya, Delegación tlalpan, México D.F México, Tel: 56063822 ext. 2011; E-mail: mltenasuck@gmail.com

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Amyloid myopathy is characterized by muscle pseudo hypertrophy and infiltration of amyloid material in the interstitium as well as within muscle fibers, particularly the type II variety, are diminished in size, because it can be found in several types of myopathies [11-13]. Muscle fibers, particularly the type II variety, are diminished in size, because it can be found in several types of myopathies [11-13]. Amyloid myopathy is characterized by muscle pseudo hypertrophy and infiltration of amyloid material in the interstitium as well as within the fibers [10], as we observed in this case. Chronic myopathy can be caused by immunoglobulin light chain amyloidosis deposition [11,12]. Amyloid myoneuropathy can resemble inclusion body myositis [13,14]. Small amounts of mononuclear inflammatory cells in endomysium or perivascular spaces without invasion of nonnecrotic muscle fibers may also be seen, leading to an erroneous diagnosis of polymyositis. Polymyositis and myositis of inclusion bodies must be considered as a differential diagnosis.

Sporadic inclusion-body myositis has been associated with abnormal accumulated proteins, of amyloid-beta precursor protein and of its proteolytic fragment, amyloid-beta, while, several aberrant proteins accumulate inside muscle fibers, including β-amyloid, α-synuclein and tau protein. Several key components of stress mechanisms of proinflammatory cells, such as nitric oxide production and macro processing autophagy contribute to muscle fiber damage associated with the aging cellular muscle fiber environment and this could be the etiology of these events [14].

We observed in our case both muscle fiber hypertrophy and that
these fibers were immunoreaction positive for light chain kappa, immunoglobulin IgG, Glut4 and dystrophin2.

Muscle fiber is the major tissue for postprandial glucose disposal and it facilitates glucose consumption into muscle fibers, and is mediated by increases in surface membrane levels of the glucose transporter GLUT4 via insulin- and/or muscle contraction-mediated GLUT4 translocation [15]. Glucose transporter GLUT4 is a larger regulator of glycemic homeostasis in skeletal muscle that is usually trapped at perinuclear spaces of myocytes. Systemic inflammation is a major risk factor for critical-illness myopathy, and could be activated by inflammation mediator NF-κB and interleukin 6, that mediate and regulate GLUT4 [15,16]. However, it is well known that the amyloid A (A-SAA) is associated with insulin resistance [17]. Therefore, we hypothesize that the expression of GLUT4 and anti-dystrophin2 could be in association with the same muscle fiber damage [18].

In conclusion, we present a rare case of periorbital amyloidosis with myopathy, and myeloid deposit was associated with the expression of IgG, kappa light chain, dystrophin and glucose receptor Glut4 in a patient which presented as with a progressive increase in volume of the left orbit with mass effect and loss of vision.

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