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Case Report

# Clinical Suspicion Refuted by Laboratory Tests

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## Abstract

Medical laboratory tests complement the medical check-up, playing an essential role in establishing a correct diagnosis. They confirm or refute certain suspicions of the disease. Depending on the specific clinical context of each patient, the physician will recommend medical laboratory tests, from routine tests to tests specific to certain conditions.

We present the case of a 73-year-old female patient, who presented to the medical office for fatigue arising from moderate exertion and breathing difficulties. Following the pulmonology check-up, it was decided to carry out a thoracic-pulmonary computer tomography (CT) which revealed fine bilateral infrahilar bronchiectasis and a voluminous round-oval process with axial dimensions of 39/26 mm located at the level of the prevascular space. It was decided to perform an anterior mediastinal puncture and the histopathological test revealed an aspect compatible with the diagnosis of type A thymoma. Considering the relationship between myasthenia gravis and tumours of the thymic gland, the diagnosis of myasthenia gravis was suspected.

Medical laboratory tests refuted the existence of anti-acetylcholine receptor antibodies. Anti-acetylcholine receptor antibodies are present in 80% of patients with generalized myasthenia gravis, which means that there are no current criteria for this diagnosis.

Medical laboratory tests can be the first line of detection of unusual events thus facilitating the establishment of an etiological diagnosis of suspicion or certainty.

Laboratory diagnosis plays an essential role in the correct clinical decision as well as in the optimal treatment for each patient.

**Keywords**: Medical Laboratory Tests; Clinical Suspicion; Thymoma; Myasthenia Gravis; Thymectomy

## Introduction

Medical laboratory tests complement the medical check-up, playing an essential role in establishing a correct diagnosis. They confirm or refute certain suspicions of the disease. Depending on the specific clinical context of each patient, the physician will recommend medical laboratory tests, from routine tests to tests specific to certain conditions.

The team approach to the patient is the key to success in order to understand the pathological complexity and make a treatment plan [1].

Patients consider fatigue to be one of the most distressing symptoms, much more distressing than nausea and vomiting, for example, which can be treated with medication [2].

The clinical hallmark of myasthenia gravis is fluctuating weakness characterized by abnormal fatigability that improves with rest [6].

Acetylcholine receptor antibodies (AChR Abs) in serum. Present in 80% to 90% of patients with generalized myasthenia gravis and 60% of patients with ocular myasthenia gravis. AChR-seronegative cases should be tested for muscle-specific kinase (MuSK) autoantibodies and anti-low-density lipoprotein receptor –related protein (LRP4) autoantibodies. Some cases remain "triple seronegative" [7].

On electrodiagnostic studies, the width of the compound muscle action potentials (CMAPs) waveform decreases by 10% to 15% with repetitive nerve stimulation [7].

Single-fiber electromicrography (EMG) shows increased "jitter". Single-fiber EMG is a technically difficult test but can be diagnostically helpful in mild or purely ocular cases [7].

The relationship between myasthenia gravis and tumours of the thymic gland was initially noted by Laquer and Weigert in 1901, and in 1949 Castleman and Norris made the first detailed description of the anatomopathological changes of the gland [3].

The role played by the thymus in myasthenia gravis is unclear; however 65% of patients have thymus hyperplasia and 10% thymoma [4].

## Clinical Case

We present the case of a 73-year-old female patient, who presented to the medical office for fatigue arising from moderate exertion and breathing difficulties. Following the pulmonology check-up, it was decided to carry out a thoracic-pulmonary computer tomography which revealed fine bilateral infrahilar bronchiectasis and a voluminous round-oval process with axial dimensions of 39/26 mm located at the level of the prevascular space.

It was decided to perform an anterior mediastinal puncture and the histopathological test revealed an aspect compatible with the diagnosis of type A thymoma.

Considering the relationship between myasthenia gravis and tumours of the thymic gland, the diagnosis of myasthenia gravis was suspected.

Medical laboratory tests refuted the existence of anti-acetylcholine receptor antibodies. Anti-acetylcholine receptor antibodies are present in 80% of patients with generalized myasthenia gravis.

The electromyography (EMG) shows that there are no criteria for myasthenia gravis (myasthenic syndrome).

Approximately 4 months later, the thoracic-pulmonary CT was repeated and the progression of the lesional process at the level of the prevascular space was noticed (45/29 mm).

Surgical intervention (thymomectomy) followed by radiotherapy is decided, and the patient's evolution is good.

## Discussions

Thymus pathology occupies a well-defined place in mediastinal pathology due to a clearly defined topography - the thymic lodge [5].

The term thymoma was coined by Grand Lomme in 1900 and has been routinely used ever since in the pathology of thymic tumours [5].

Most statistics claim that the malignancy of thymic tumours after the age of 40 is 70% [5].

There are no clinical signs characterizing thymic tumours except when there are associations with other diseases: myasthenia, leucosis [5].

Once the diagnosis of thymic tumour is established, surgical intervention is required, even if the tumour is associated with other conditions [5].

What is the thymus gland? What is a myoid cell?

The thymus gland is a small gland located in the fat pad beneath the sternum. It plays a critical role in the maturation of immunologically active cells and in the development of immune self-tolerance in the healthy patient. Myoid cells are muscle-like cells found mainly within the medulla of the thymus. Myoid cells express nicotinic AChRs, and given their location within this critical site for the development of the global immune response, these cells may play a pivotal role in autosensitization against the receptor in myasthenia gravis [8].

What evidence suggests that the thymus gland has a major role in the pathogenesis of myasthenia gravis?

Removal of the thymus seems to improve myasthenia gravis in the majority of patients.

The majority of patients with myasthenia gravis have an abnormal thymus, demonstrating either hyperplasia or thymoma.

Thymic myoid cells express AChRs proximate to the site of T-lymphocyte maturation (which includes immune self-tolerance).

AChRs in the thymic myoid cells in myasthenia gravis express the fetal gamma subunit, making them potential targets for antibody sensitization.

Thymic B lymphocytes from patients with myasthenia gravis produce more anti-AChR antibodies than other antibodies.

Tyimic cells selectively increase the production of anti-Ach antibodies when added to myasthenic B lymphocytes in the laboratory.

Myasthenia gravis thymus tissue transplanted to immunodeficient mice produces anti-AChR antibodies, which deposit at skeletal muscle endplates [8].

## Conclusions

Medical laboratory tests can be the first line of detection of unusual events thus facilitating the establishment of an etiological diagnosis of suspicion or certainty.

Laboratory diagnosis plays an essential role in the correct clinical decision as well as in the optimal treatment for each patient.

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