



Clinical case report

Unclear dyspnea

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Estonia

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Patient overview

- 70 yrs old woman
- May 2009, progressive weakness, dyspnea on exertion; episodic partial ptosis of the right eyelid

Consultations:

- Jun 09 ophthalmologist (no specific findings)
- Jun 09 endocrinologist (thyroid gland tests, ultrasonography were normal)
- Jul 09 neurologist (electroneuromyography, CT of brain – normal findings)

- **Past medical history**: hypertension about 3 years.
- **Medications**: ramipril, metoprolol, hydrochlorothiazide.
No history of alcohol consumption, smoking.



Suspecting heart disease

September 2009, four months later

Last 3 weeks, exacerbation of symptoms: breathlessness on mild exertion, sometimes at rest, could sleep only with elevated head. On the suspicion of heart failure, she was admitted to cardiology department.

Examination:

- **Obesity** (BMI 41 kg/m²)
- **Heavy dyspnea and orthopnea, bilateral basal crepitant rales**
- **Partial ptosis of the right eyelid**
- Regular heart rate (90 bpm), no cardiac murmurs
- Blood pressure 140/90 mmHg
- No palpable organomegaly
- No extremity edema



Suspecting heart disease

Analyses

- Blood gases: normal
- CK-MBm 3.20 (<3.77 ng/mL)
- Troponin T <0.010 (<0.030 ng/mL)
- NT-proBNP 230 (<125 pg/mL)
- D-Dimers 0.87 (<0,41 µg/mL)
- CRP 19 (<5 mg/L)
- Normal liver, kidney tests
- Borrelia IgM, IgG negative

Investigations

- Normal ECG
- Chest X-ray: Moderate pulmonary edema
- Echocardiography: Normal systolic function, EF 60-65%. Normal pulmonary pressure. No segmentary contractility disorder. Heart valves' structure and function in order.

->No serious cardiac disease



Suspecting lung disease

Pulmonary edema was resolved with diuretic treatment (body weight 100 -> 90 kg), but dyspnea did not recede enough.

Further investigations:

- CT angiography of lungs
 - Ventilation/perfusion scan
- } normal

-> **Pulmonary embolism and other lung diseases excluded**

-> Dyspnea and decreasing of physical performance were related to high body weight and possible recent respiratory viral infection. Patient was permitted home.

At home

During one week, general weakness increased; ptosis of the left eyelid, diplopia, lump feeling in throat, difficulties with chewing and swallowing developed.



Suspecting neurological disease

In neurological department:

Examination: Speech clear. Deep tendon reflexes normal (dex=sin). No paresis, ataxia, nor pathological reflexes. Symmetrical facial movement, normal swallowing reflex and throat reflex. Normal pupils.

- Partial **ptosis** (sin>dex). Compensatory raising of eyebrows, head tilted back.



- **Diplopia** by looking left (left eyebulbs turns up)





Description of illness

Electroneuromyography:

Remarkable postsynaptic impairment in facial muscles and extremities. Jitter was not measurable because a lots of blocks in test-muscles. Diagnose - **Myasthenia gravis**.

Myasthenia gravis (MG)

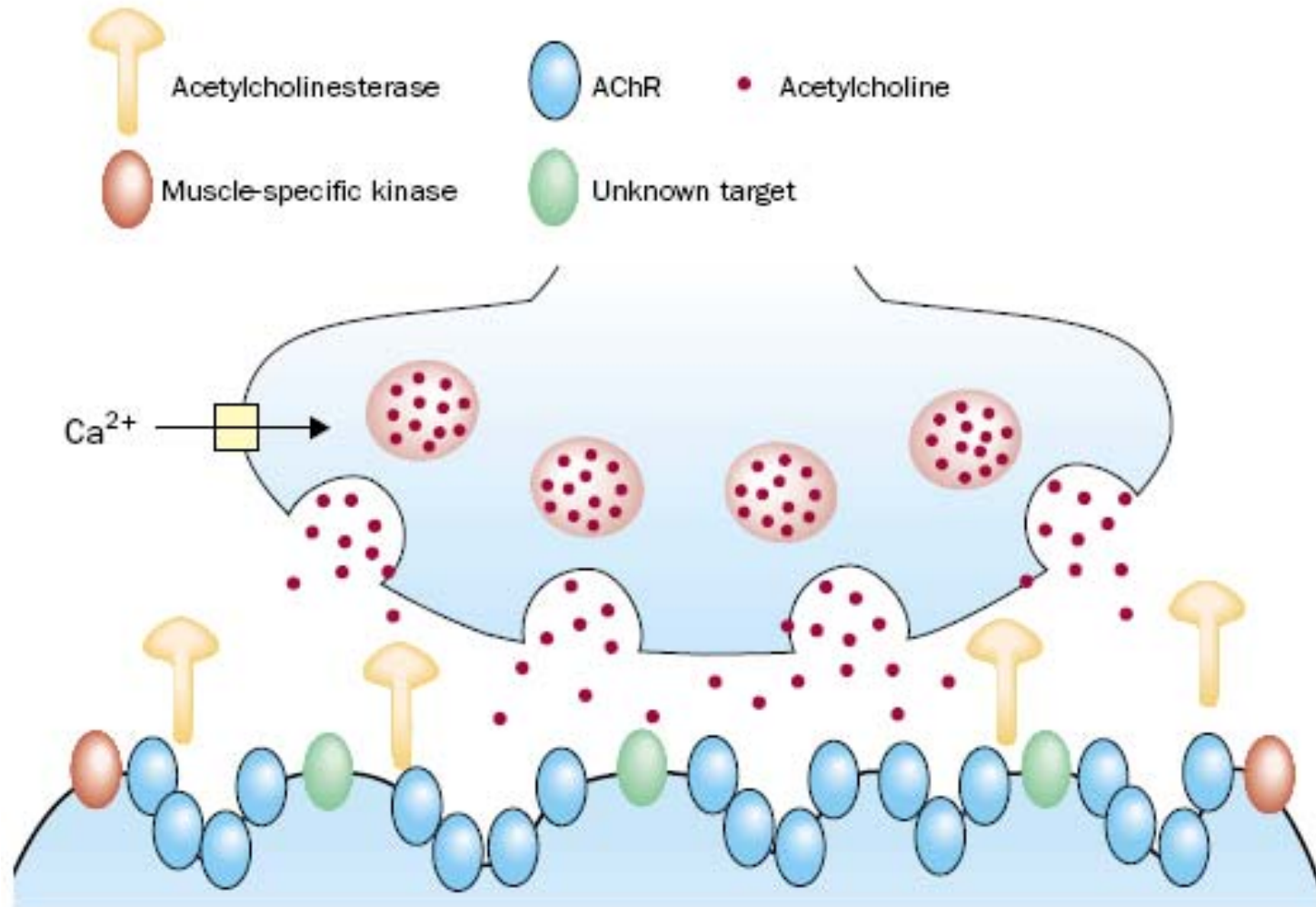
Autoimmune disorder of the neuromuscular junction.

- Prevalence 14-15/100 000
- Bimodal peak of incidence: 30 y. ♀, 60-70 y. ♂

Main feature is weakness of muscles, which increases during repeated use and improves after rest. The most commonly affected muscles: levator muscle of upper eyelid, facial muscles, proximal limb muscles and neck extensors. Ocular palsies are often asymmetrical.



Neuromuscular junction





Diagnostic tests for MG

- Tensilon test
 - Does the intravenous short-acting acetylcholinesterase inhibitor (Edrophonium) influence symptoms?
- Presence of anti-AChR and anti-MuSK antibodies
- Electrophysiological tests to assess NMJ
 - Repetitive nerve stimulation and single fiber EMG
- Chest MRI/CT
 - Pathology in thymus?
- Additional tests to detect other autoimmune diseases



Treatment options of MG

- Symptomatic therapy, first line treatment:
 - Cholinesterase inhibitors (Pyridostigmine) 30–90 mg every 4–6 h
- Long-term immune therapies
 - First line: prednisone (adjust to minimum dose), or prednisone plus azathioprine or mycophenolate mofetil
 - Second line: ciclosporin or tacrolimus, IVIG
 - Third line: cyclophosphamide, rituximab, serial PE or IVIG
- Short-term immune therapies (treatment of MG exacerbations):
 - Plasma exchange (PE): 4–6 exchanges on alternate days
 - Intravenous immunoglobulin (IVIG) 1–2 g/kg (over 2–5 days)

Consider thymectomy: Therapeutic option in anti-AChR-positive generalised MG with disease onset before the age of 50 years.



In our case ...

Treatment

- Pyridostigmine 60 mg x 3->120 mg x 3
- Azathioprine 50 mg x 1
- Aspirin 150 mg x 1
- Metoprolol retard 25 mg x 1
- Ramipril 2.5 mg x 2
- Furosemide 40 mg x 1 (every other day)
- Diclophenac 100 mg x 1

Treatment effect positive: improved general feeling, chewing and swallowing disorder receded, dyspnea and eye symptoms decreased significantly.



Learning points

- Generalized myasthenia gravis can mimic symptoms of other diseases.
- In the beginning of illness the symptoms are mild and controversial. Repeat the investigations, when you have clinical suspicions!
- There is a risk, that specialisation in medicine may compromise integral approach to the patient



Never underestimate ocular signs!

Thank you!