# Electromyography (EMG) - Useful Tool for POMPE Disease Diagnosis

# Điện cơ một phương tiện hữu ích trong chẩn đoán bệnh tích glycogen typ 2

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## ABSTRACT

**Introduction:** Late onset Pompe disease (LOPD) is a treatable, rare disease. Main signs & symptoms: proximal weakness, respiratory failure, & hyper Ckemia. Difficult to R/O from other neuromuscular pathologies: SMA, LGMD, etc. Moreover, genetic test (WES) indicated at the disease onset is not feasible. EMG is one helpful attempt to LOPD diagnosis with myotonic discharge on EMG but no clinical hypertonia. So far, the present is one of the three LOPD cases in Vietnam.

**Results:**

HPI: 14 y.o. girl admitted to ER with: dyspnea, fatigue, flaccid quadriparesis, respiratory failure using ventilator. Diagnosis: severe pneumonia, suspected neuromuscular disease.

PH: normal obstetric family. Psychomotor development: 12 y.o.: dyspnea during exhaustive physical activity (running), worsening with time, normal breath when climbing or going upstairs. 14 y.o: no exhaustive physical activity, dyspnea when going upstairs with muscle cramp. Walking with foot drop. Normal learning ability. Frequent wheezing before 3 y.o. Pneumonia needing two hospitalizations at 2 and 4 y.o

FH: unremarkable

Examination: Alert, attentive and oriented. No cranial nerve palsy. Weak neck. Weakness of lower limb than upper; distal limbs weaker than proximal. Myotonia (-). Mild muscle atrophy. Reflex (-). Normal sensory exam. No CNS's sign

**Workup:**

• 1st EMG, AMBU bag via endotracheal tube: normal NCS. Needle EMG: spontaneous activity (+++), predominant large, long duration MUAPs, suspected spinal muscular atrophy (SMA). Gene test for SMA: negative result after 1 week.

• Lab tests:

CBC, CSF: normal. Increased liver function and CK. Ultrasound: Musculoskeletal: Changes of diaphragm, brachialis, biceps, quadriceps, gastrocnemius, paraspinal muscle, possible muscle dystrophy; Thyroid, Cardiovascular: normal. ECG: normal. Brain MRI: normal. Chest CT scan: pneumonia

**Disease evolution:**

Unimproved respiratory failure, ventilator dependence. Progressive limb weakness: distal to proximal. Body weight loss. Unindicated CES due to family financial problem.

• 1 month later:

2nd EMG (one month later), AMBU bag, NCS: light CMAP decrease of some motor nerves, normal SNAP. Needle EMG on anterior tibial and quadriceps femoris muscles: sound of myotonic discharge (++), alternately with CRD (++). Early recruitment over proximal - distal muscle groups. Suspected Myotonic Dystrophy.

EMG myotonic discharge: no clinical myotonic dystrophy, suspected maltase acid deficiency (Pompe disease).

Dried blood spot (DBS) and gene test to support LOPD.

• After 21 days, Gene test: compound heterozygous on GAA gene. NGS: 2 variants in GAA gene.

• After 2 months, DBS: GAA deficiency

• After 2.5 months, by clinical symptom, EMG with myotonic discharge; DBS (+), gene test with GAA (+), diagnosis: LOPD. Myozyme (Sanofi Genzyme), 20 mg/kg/dose infused every 2 wks. After 1 month: tracheotomy. After 4 months: weaned off ventilator. After 8 months: hospital discharge. With Myozyme, now she can walk, and must use ventilator only in sleep.

**Conclusion:**

• LOPD though rare should be suspected in progressive proximal muscle weakness, respiratory insufficiency, and hyper CKemia levels.

• EMG, less invasive and less expensive than muscle biopsy, is useful to attempt LOPD diagnosis, with myotonic discharge on EMG, without clinical myotonia. (2)

• This patient with distal weakness of lower limbs, aflexia, and spare sensory refers to a possible neuropathy or SMA. Moreover, 1st EMG with large, long duration MUAPs, can lead to misdiagnosis of SMA instead of LOPD. This is in line with W. Muller's study (1) of neurogenic pattern in EMG.

• Difficulty in practicing needle EMG in children is increased with this patient due to Ambu bag squeezing. Myotonic discharge by needle EMG in lower limb muscles is easier than in paraspinal muscles. (4)

## REFERENCE:

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