Late Onset Pompe Disease (LOPD) is a metabolic, autosomal recessive disease due to a reduced functionality of enzyme alpha-glucosidase (GAA), which leads to an accumulation of glycogen in skeletal muscle tissue and in other organs. 70% of LOPD patients develops progressive respiratory failure (RF) with vital capacity reduction and risk of mechanical ventilation. Recent data suggest the importance of an early LOPD diagnosis.

**Prevalence of LOPD suspect in patients with respiratory failure**

Aim: to develop an early LOPD diagnostic algorithm for suspected LOPD (Fig. 2a-b) to assist clinicians in the management of adult patients with unexplained RF and follow the diagnostic flow chart to confirm the disease. Non-invasive mechanical ventilation (NIV) has been collected on DBS. The aim of the study is to implement a centralized GAA measurement of GAA, executed in a centralized (VACS) and to develop a comprehensive diagnostic algorithm for suspected LOPD (Fig. 2a). The overall goal of the study was to optimize the diagnostic process in pulmonology units, encouraging early diagnosis and management of LOPD.

**Methods**

- Real-life study, conducted in 18 Pulmonology Italian Centers (Fig. 1), aimed to enroll 500 adult patients with unexplained RF and follow the diagnostic algorithm for suspected LOPD (Fig. 2a-b).
- For each patient, clinicians collected a drop of blood on Dried Blood Spot (DBS) for the measurement of GAA, executed in a centralized laboratory.
- Muscular disability was assessed with Walton Gardner Scale (WMS).

**Preliminary Results**

PneumoLoped started in February 2015 (enrollment is ongoing). 67 patients have been enrolled at the date of this analysis (characteristics in Table 1 and Fig. 3-7):

- 43% accrued for respiratory reasons
- 45% was hospitalized and 33% needed mechanical ventilation
- Hyperckemia was reported in 39% of patients
- The majority of cases (78%) presented a WMSG between 0 and 5

**Conclusions**

PneumoLoped could provide original data to better highlight the role of pulmonologists in the management of acute respiratory failure associated to neuromuscular diseases, and in the assistance of subjects with genetic rare disorders such as Late Onset Pompe Disease.

**References**


**Background**

Late Onset Pompe Disease (LOPD) is a metabolic, autosomal recessive disease due to a reduced functionality of enzyme alpha-glucosidase (GAA), which leads to an accumulation of glycogen in skeletal muscle tissue and in other organs. 70% of LOPD patients develops progressive respiratory failure (RF) with vital capacity reduction and risk of mechanical ventilation. Recent data suggest the importance of an early LOPD diagnosis.

**Objectives**

- Prevalence of LOPD suspect in patients with respiratory failure
- Optimize the diagnostic process in pulmonology units, encouraging early diagnosis

**Methods**

- Real-life study, conducted in 18 Pulmonology Italian Centers (Fig. 1), aimed to enroll 500 adult patients with unexplained RF and follow the diagnostic algorithm for suspected LOPD (Fig. 2a-b).
- For each patient, clinicians collected a drop of blood on Dried Blood Spot (DBS) for the measurement of GAA, executed in a centralized laboratory.
- Muscular disability was assessed with Walton Gardner Scale (WMS).

**Preliminary Results**

PneumoLoped started in February 2015 (enrollment is ongoing). 67 patients have been enrolled at the date of this analysis (characteristics in Table 1 and Fig. 3-7):

- 43% accrued for respiratory reasons
- 45% was hospitalized and 33% needed mechanical ventilation
- Hyperckemia was reported in 39% of patients
- The majority of cases (78%) presented a WMSG between 0 and 5

**Conclusions**

PneumoLoped could provide original data to better highlight the role of pulmonologists in the management of acute respiratory failure associated to neuromuscular diseases, and in the assistance of subjects with genetic rare disorders such as Late Onset Pompe Disease.

**References**
