## CR59
## NOT EVERY RESPIRATORY FAILURE NOWDAYS IS COVID. POMPE DISEASE

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INTRODUCTION/OBJECTIVES: Pompe disease, also known as glycogen storage disease type II, is an autosomal recessive disorder caused by deficiency of the lysosomal enzyme acid-α-glucosidase. It is a chronic and progressive disease characterized by storage of glycogen mostly in muscles. Late onset cases typically present with proximal muscle weakness and respiratory insufficiency or exertional dyspnea. Treatment is now available with intravenous infusion of recombinant acid α-glucosidase.
CASE PRESENTATION: We present a 45-year-old patient that was healthy up to 2017. when he started developing mild muscle weakness. He was not able to stand up from sitting position. At the beginning of 2020. his symptoms aggravated following a respiratory tract infection (SARS CoV-19 was suspected but test was negative). He developed severe respiratory failure and was treated in the ICU. Given the clinical presentation, late onset form of Pompe's disease was suspected. Diagnose was confirmed with low Alpha glucosidase enzyme concentration and muscle biopsy (glycogen deposits) and 32-13T>G- missense gene variation. At the time he was tracheostomized and dependent on home oxygen concentrator. Enzyme replacement therapy with Myozyme was introduced. After a year on Myozyme the patient is subjectively well and has been decannulated. His 6MWT is improving and he is starting to live a more active life.
CONCLUSION: Pompe disease is rare, and the awareness about it is low. Pompe disease should be always in differential diagnosis of patients presenting with muscle weakness and respiratory insufficiency lacking clear pulmonal pathology. Early diagnosis and treatment improves outcome and significantly improves the quality of patients' lives.

## CR60
## Pemetrexed treatment for adenocarcinoma of unknown primary origin with BAP-1 mutation – implementing NGS analysis into clinical practice

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Keywords: BAP-1 mutation, Next-generation sequencing (NGS), pemetrexed

INTRODUCTION/OBJECTIVES: Genome analysis like „Next-generation sequencing" (NGS) has impacted research of complex diseases including cancer. NGS allowed a cost- and time-effective sequencing of tumor DNA, introducing us to a „genomic era" of cancer research and treatment.
CASE PRESENTATION: The 65-year-old female patient with a history of arterial hypertension and hypothyroidism presented with palpable lesion and pain under her left rib cage. After comprehensive diagnostic evaluation, other lesions were found in the left lung hilum, pleura and XI left rib. The patient was operated and histopathological analysis showed adenocarcinoma possibly originating from pancreas, biliary tract or gallbladder. During the diagnostic procedure and operation, the primary origin was not found. Adjuvant therapy included cisplatin and gemcitabine but, because of disease progression, it was changed to docetaxel. Despite the systematic treatment, progression was noticed both intrathoracic and intra-abdominal. NGS analysis of tumor material revealed a BAP-1 (BRCA1- Associated Protein 1) mutation linked to several tumors including malignant mesothelioma. The treatment was continued with pemetrexed indicated for malignant mesothelioma and NSCLC (non-small cell lung cancer). Regression of metastatic lesions was achieved.
CONCLUSION: This case represents that finding the molecular background of cancer can lead us to more specific and effective treatment and better outcomes in the future.