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P31. IDENTIFICATION OF NEW BIOMARKERS TO ENABLE LIQUID BIOPSY FOR POMPE DISEASE

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Evaluation of patient's condition and monitoring the efficacy of Enzyme Replacement Therapy (ERT), is a critic issue for Pompe Disease (PD), a metabolic, hereditary disease, due to deficiency of lysosomal acid alpha glucosidase. In this study we are looking for a set of measurable biomarkers in plasma/serum of PD patients, based on the analysis of micro-RNA (miRNA) and oxylipins. MiRNAs are small non-coding RNAs that regulate gene expression. Oxylipins are an emerging group of metabolites, generated by oxygenation of polyunsaturated fatty acids. Both are molecoles involved in numerous biological functions and their content in circulation can alter in response to pathological conditions. We are evaluating circulating miRNAs and serum oxylipins in samples of patients (N = 52) and of mouse model. Our approach combines Next Generation Sequencing techniques, tandem mass spectrometry and bioinformatics analysis. In previous work we stated that miR-133a is upregulated in plasma of PD patients (Tarallo et al, 2018). We have evaluated others two circulating microRNAs, miR-1 and miR-206. Both are upregulated in the plasma of PD patients and correlate with the severity of the phenotype and the response to ERT. Preliminary data also suggest that the profile of serum oxylibins in PD mice is altered, compared to age matched wild type mice. The identification of multiple biomarkers measurable in blood would allow for development of so-called "liquid biopsies" in the management of PD, as tool to support the diagnosis, monitor patient's condition and evaluate the effecacy of ERT.