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ORAL COMMUNICATION

Proteomic Analyses And Integrative Omics Approach To Highlight The Common Molecular Signatures For Different NCL Forms

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Neuronal ceroid lipofuscinoses (NCLs) are a group of inherited lysosomal storage disorders characterized by neurodegeneration, mainly with childhood onset. To date, there is no curative therapy to delay or halt disease progression but only treatments aimed to alleviate the symptoms. Currently, 14 different forms (CLN1-CLN14, CLN = ceroid lipofuscinosis, neuronal) of this kind of disorders have been identified associated with mutations in 14 distinct genes, but mounting evidence seems to indicate that the resulting protein products act in shared or convergent biological pathways [1].
In the last years, mass spectrometry has been exploited for the identification and quantification of proteins differentially expressed in specific NCL forms [2]. However, these proteomic studies have only provided an incomplete picture of the common molecular mechanisms underlying this group of neurodegenerative diseases.
To fill this gap, we propose here a comprehensive bioinformatic analysis, using the software QIAGEN IPA, of CLN proteomic and transcriptomic datasets already present in the literature or generated by ourselves. From an experimental point of view our laboratories contributed to this integrative omics study carrying out proteomic investigations on SH-SY5Y cell lines (used as in vitro NCL models) through the highly informative and accurate SWATH-MS (Sequential Window Acquisition of all THeoretical fragment ion Mass Spectra) methodology.
We strongly believe that the results deriving from this multi-omics approach could represent a promising starting point for the identification of both molecular networks common to different CLNs as well as shared biomarkers related to disease onset and progression, thus paving the way to the design of new effective treatment options for all NCLs.

Keywords: Proteomics, SWATH-MS, NCLs

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