

Accepted Manuscript

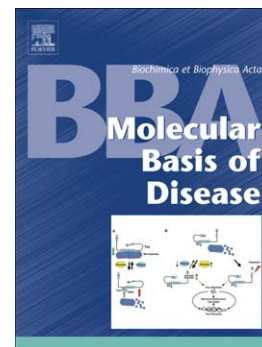
The Neuronal Ceroid Lipofuscinoses Program: A Translational Research Experience in Argentina

Romina Kohan, Favio Pesaola, Norberto Guelbert, Patricia Pons, Ana María Oller-Ramírez, Gisela Rautenberg, Adriana Becerra, Katherine Sims, Winnie Xin, Inés Adriana Cismondi, Inés Noher de Halac

PII: S0925-4439(15)00146-5
DOI: doi: [10.1016/j.bbadis.2015.05.003](https://doi.org/10.1016/j.bbadis.2015.05.003)
Reference: BBADIS 64219

To appear in: *BBA - Molecular Basis of Disease*

Received date: 28 April 2015
Revised date: 29 April 2015
Accepted date: 5 May 2015



Please cite this article as: Romina Kohan, Favio Pesaola, Norberto Guelbert, Patricia Pons, Ana María Oller-Ramírez, Gisela Rautenberg, Adriana Becerra, Katherine Sims, Winnie Xin, Inés Adriana Cismondi, Inés Noher de Halac, The Neuronal Ceroid Lipofuscinoses Program: A Translational Research Experience in Argentina, *BBA - Molecular Basis of Disease* (2015), doi: [10.1016/j.bbadis.2015.05.003](https://doi.org/10.1016/j.bbadis.2015.05.003)

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The Neuronal Ceroid Lipofuscinoses Program: A Translational Research Experience in Argentina

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Highlights

- The integrated study of NCL aimed to overcome underdiagnose in Latin America.
- Seven/13 known NCL forms were found in the region.
- Genotypes showed mostly heterozygous combinations of mutations.
- CLN2 disease is the most frequent genotype (65.3%), followed by CLN3 disease (12.2%).
- The most frequent mutation in the region is *TPP1*-p.Asp276Val (30.5%).

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