

# in Latin America

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|---|--|
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|  Irene Paradisi         | Mario Cornejo-Olivas           |
|  Jonathan Javier Magaña | Miguel Angel Ramirez Garcia    |
|  Laura Bannach Jardim   | Nicolas Sommaruga              |
|  Luis Velázquez Perez   | Paula Saffie Awad              |
|  Malco Rossi            | Pedro Braga Neto               |
|  Marcelo Kauffman       | Thomas Klockgether             |
|  Marcondes França Jr    | Wilson Marques Jr              |

**online meeting, May 7, 2026,  
8:30 am-13 pm (Brasilia), 1:30-6 pm CEST**

<https://ataxia-global-initiative.net/events/agi-regional-conference-in-latin-america/>

# AGI Regional Conference

**May 7<sup>th</sup>, 2026**

***Online meeting***

**Malco Rossi**

*Movement Disorders Section  
Neurology Department, Fleni  
Instituto Fleni-CONICET (INEU)  
Buenos Aires, Argentina*



# Personal interest in ataxia, affiliations & clinical/research background

## Affiliations

### FLENI

*Buenos Aires - Argentina*



### CONICET

*Buenos Aires - Argentina*



## Clinical & research background on genetic ataxias

- Diagnostic approach for genetic ataxias - clinical algorithms
- AI-based tools for the differential diagnoses of genetic ataxias
- Epidemiological study of prevalence of SCAs in Argentina
- MDS Nomenclature of genetic ataxias (ATX prefix)
- MDSGene genotype-phenotype correlations for some ataxia genes
- MDSGene Ataxia Online Resource: [mdsgene.org/ataxia.html](http://mdsgene.org/ataxia.html)
- Deep phenotyping of rare or atypical cases
- Focal cerebellar lesion cohort

Grateful to mentors in ataxia, neurogenetics and movement disorders



# Perspectives on personal experiences in studying and managing hereditary ataxias

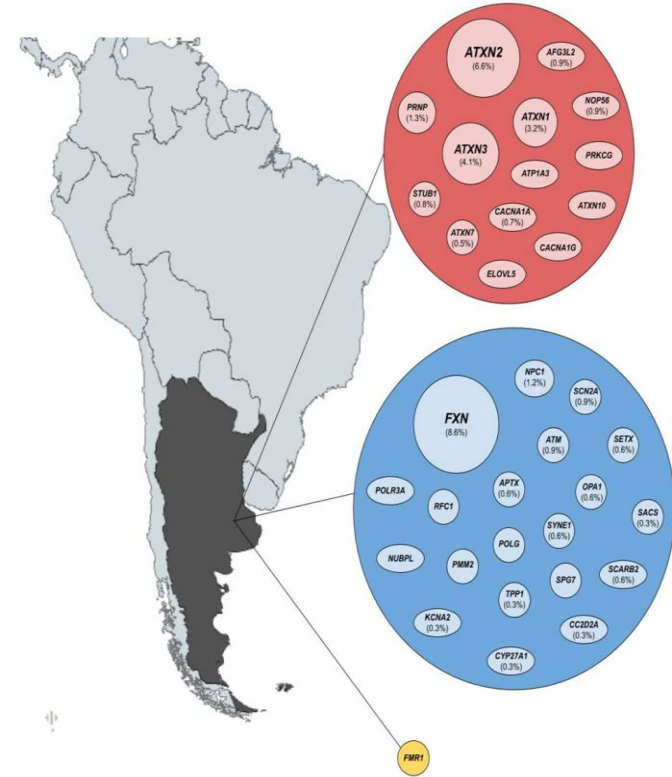
## Clinical & research team:

- Movement disorders neurologists and fellows
- Neuroimaging specialists and Neurology residents
- Bioengineers

- About 100 patients with genetic ataxia confirmed.
  - Mostly SCA2, SCA3, and Friedreich ataxia
- Patients referred from across the country and neighboring countries.
- Electronic clinical records and own patient databases.
- Standardizing patient follow-up assessments with rating scales
  - Applying this approach to patients with Friedreich ataxia receiving omaveloxolone treatment.
- 3-Tesla MRI, neurofilaments, Oxford Nanopore Technologies, DBS
- Rehabilitation Center at Fleni Escobar



## Local challenges in terms of access, organization, diagnosis, management, research and genetic counseling for ataxias



- Argentina is a middle-income country (population of 46 millions)
- Buenos Aires and greater metropolitan area (15 millions)
- The healthcare system is divided into three sectors: public, social security, and private.
- Several laboratories (privately or publicly managed) perform genetic tests for ataxia, such as **NGS, repeat expansion detection methods, and long-read sequencing.**

### Barriers to genetic testing: not always financially covered

- Genetic diagnostic studies of ataxia cohorts in Argentina found **high rates of undiagnosed patients (65% to 82%).**
- Need to test more *NOP56* (SCA36), *RFC1*, and *FGF14*

## Opportunities in clinical research and care

- Developing a **trial-readiness national registry of genetic ataxias** that is compatible with existing international multicenter registries or consortia.
- **Natural history studies**, promote collaboration between clinicians and researchers

### - **Specialized Ataxia Center, Clinic or Unit**

- In Argentina, patients with ataxia generally have access to neurologists
- There are only a few movement disorders specialists with dedication and expertise in ataxia
- Still no interdisciplinary groups of health and allied health professionals solely dedicated to treating patients with ataxia. Few trained genetic counsellors.

- Maintain a close relationship with national ataxia **patient advocacy and support groups** in Argentina and with the national federation of rare diseases (FADEPOF).

- **More research funding opportunities for screening studies on ataxia cohorts.**

Thank you for  
your attention

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**ATAXIA** GLOBAL INITIATIVE

worldwide platform for clinical research in ataxias



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