

# Ataxias in Bolivia: What little we know, and what remains unknown.

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**ATAXIA** GLOBAL INITIATIVE  
worldwide platform for clinical research in ataxias

in  
**Latin  
America**

# Bolivia in the literature: signals, not data

- Very limited published evidence
- No national prevalence studies
- No Bolivian molecular cohort
- No structured national registry
- Main evidence: isolated family reports

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## Epidemiology of Autosomal Dominant Spinocerebellar Ataxias in Latin America: A Systematic Review and Meta-Analysis

Milagros Galecio-Castillo <sup>1 2</sup>, Jesus Gutierrez-Arratia <sup>3</sup>, Alonso Abad-Murillo <sup>1 3</sup>,  
Elison Sarapura-Castro <sup>1 3</sup>, Ismael Araujo-Aliaga <sup>1 3</sup>, Ana Saldarriaga-Mayo <sup>1 3</sup>,  
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# The strongest evidence: SCA2/SCA10 family

- **Genetically confirmed Bolivian kindred**
- SCA2 + SCA10 in the index case and mother
- SCA10 only in maternal uncle
- Hybrid phenotype:
  - slow saccades
  - cerebellar ataxia
  - seizures
- Hypothesis: possible local enrichment, still unproven



## HHS Public Access

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### **Bolivian Kindred with Combined Spinocerebellar Ataxia Type 2 and 10**

**José Fidel Baizabal-Carvalho, M.D., M.Sc.<sup>1</sup>, Guangbin Xia, M.D.<sup>2</sup>, Peter Botros<sup>3</sup>, Janeth Laguna, M.D., Tetsuo Ashizawa, M.D.<sup>2</sup>, and Joseph Jankovic, M.D.<sup>1</sup>**

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# A second report: probable Friedreich ataxia, but not confirmed

- **Three siblings from Potosí**
- Childhood onset gait disorder
- Hyporeflexia
- Foot deformities / scoliosis
- Visual abnormalities / nystagmus
- Clinically compatible with Friedreich ataxia
- No genetic testing available

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## INFORME DE CASO

**Ataxia de Friedreich. Informe de tres casos realizado por la Brigada Médica Cubana en Bolivia**

**Friedreich's ataxia. Report of three cases carried out by the Cuban Medical Brigade in Bolivia**

Dra. Mayra López Milián<sup>1</sup>, Dra. María Victoria Fajardo Gómez<sup>2</sup>, Dra. Leyda Méndez López<sup>3</sup>

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# Local experience in context: Bolivia and Neurocenter

- Bolivia:
  - 11.36 million inhabitants
- Neurocenter:
  - 3,188 first-time outpatient neurology visits
- Movement disorders/ Parkinsonism:
  - 313 patients
- Separate internal review:
  - 4 suspected genetic ataxia cases
- No local molecular confirmation
- Real-world signal, not epidemiology





# What Bolivia needs next

Preliminary discussions with Peruvian neurogenetics colleagues are underway to explore future sequencing pathways.

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From isolated cases to systematic data:

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Multicenter registry

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Standardized phenotype collection

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Access to genetic testing

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Regional collaboration through AGI

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Inclusion of Bolivia in Latin American ataxia research