


Hereditary Ataxias in Chile

What do we expect, what is unexpected, and what remains unresolved?

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worldwide platform for clinical research in ataxias



in Latin America

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**Porto Alegre, Brazil. On site and online meeting
May 7, 2026**



What is expected/ known?

Published Chilean data is sparse, and evidence is limited

Published cases 2015-2026

17 SCA3	1 SCA2
3 SCA19	9 <i>FGF14</i>
7 FRDA	7 <i>RFC1</i>
2 <i>ARSACS</i>	1 <i>SYNE1</i>

Unpublished cases Followed in 2 specialized centers

22 SCA3	1 SCA1
1 SCA44-45	1 SCA10
16 FRDA	6 <i>RFC1</i>
2 <i>ATM</i>	1 <i>SYNE1</i>

Summary of data

SCA3/ FRDA most frequent
SCA27B/ *RFC1* increasingly recognized

Limited epidemiological data
- Case reports/ case series underreport
Limited access to genetic testing
-Lack national registries

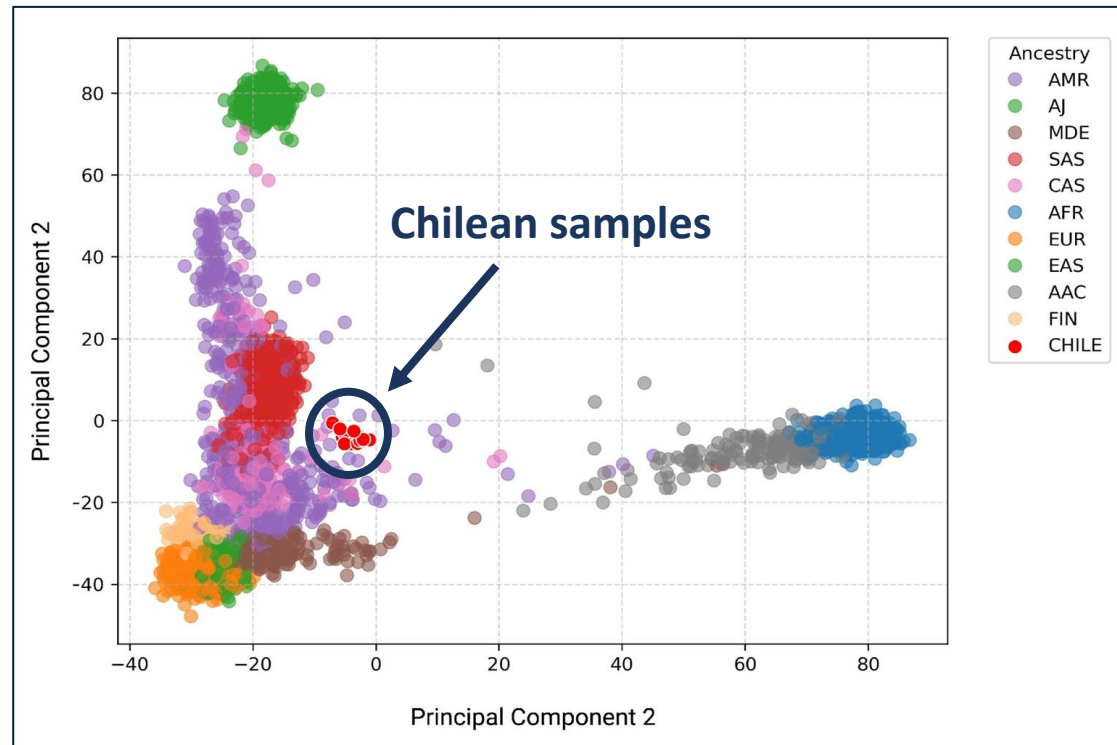
What is unexpected?

SCA4 appears in a population background where it would not be classically expected

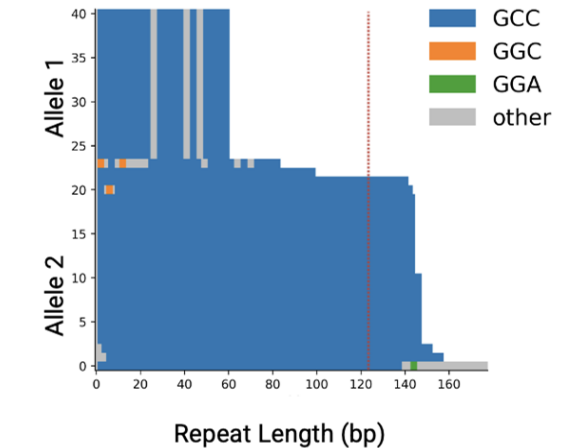
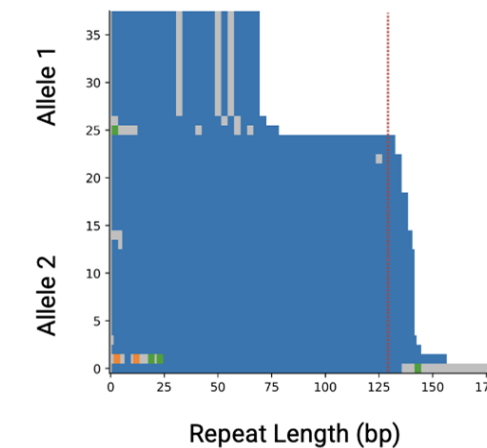
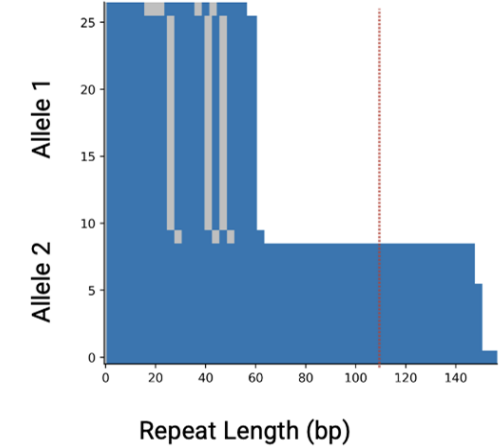
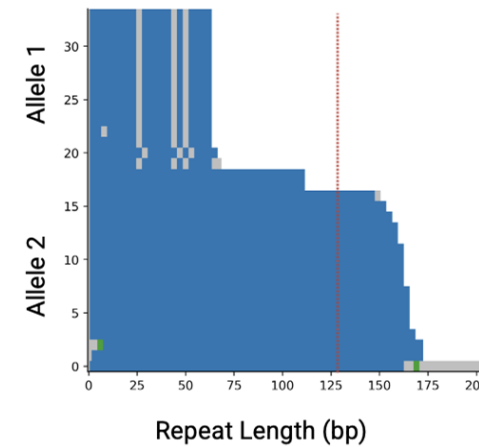
4 cases with SCA4

Why this matters

- Unexpected in a Latino admixed population
- Outside the population background where SCA4 has been classically described
- Carriers with founder-associated haplotype



All carriers had one expanded allele above the pathogenic threshold.



What still needs to be solved

Unsolved cases nationwide and specific regional cases

Chiloé Island (example)

Chiloé is an isolated archipelago in southern Chile with ~170,000 inhabitants

17

hereditary
ataxia
patients

3

Familial
clusters

12

Índex cases

~10/100,000 minimum observed prevalence

Genetic results studied cases

Result	Number / proportion
Definitive diagnosis	2/13 (15%)
Identified diagnoses	1 homozygous <i>SPG7</i> ; 1 <i>RFC1</i>
VUS burden	70% carried VUS
Unsolved cases	85% unsolved

Are these unresolved Chiloé cases due to underpowered databases, missed structural/repeat variants, founder effects, or a new genetic ataxia?

Take-home message

- High heterogeneity and a large diagnostic gap
- Unique genetic architecture in Chile
- Universal access to genetic tests is needed
- New genetic technologies, for unsolved cases



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