Epidemiological Update on Hereditary Ataxias in Uruguay

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A (not so) short timeline

March 2019

Ataxia Clinic

Hospital de Clínicas Dr. Manuel Quintela

November 2022

Adult´s Rare Diseases

CRENADECER BPS

October 2024

First Uruguayan Report

May 2026

Second Uruguayan Report

> Cerebellum. 2025 Apr 28;24(4):89. doi: 10.1007/s12311-025-01839-6.

Clinical Characteristics of Spinocerebellar Ataxia Type 3 in Uruguay

Nicolás Sommaruga [1, 2], Nicolás Labaure [3, 4], Andrea Zamora [3, 4], Santiago Giménez [3, 4], Gonzalo Pérez-Hornos [3, 4], Cristina Vázquez [3]

Affiliations + expand

PMID: 40289053 DOI: 10.1007/s12311-025-01839-6

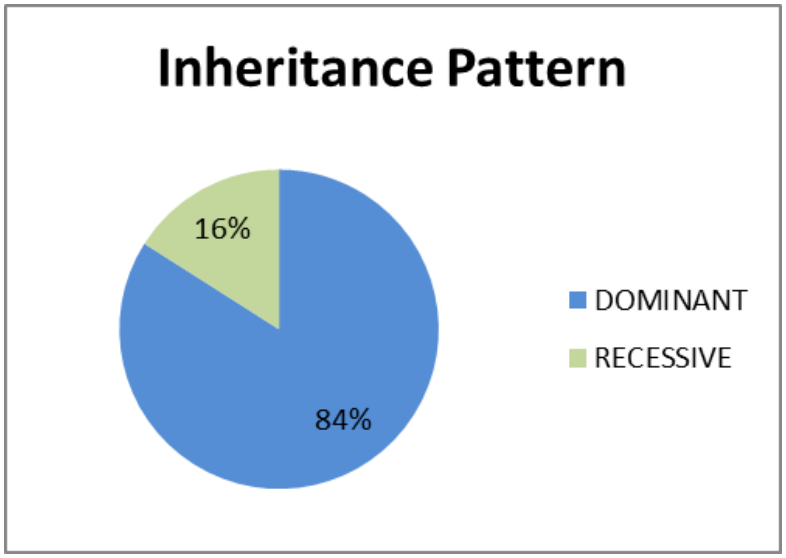
Initial Data

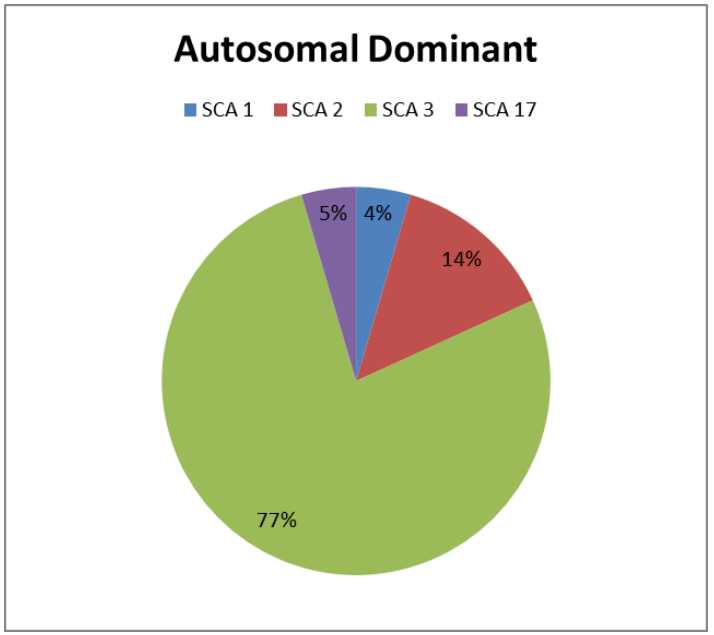
(Adult Terciary Center)

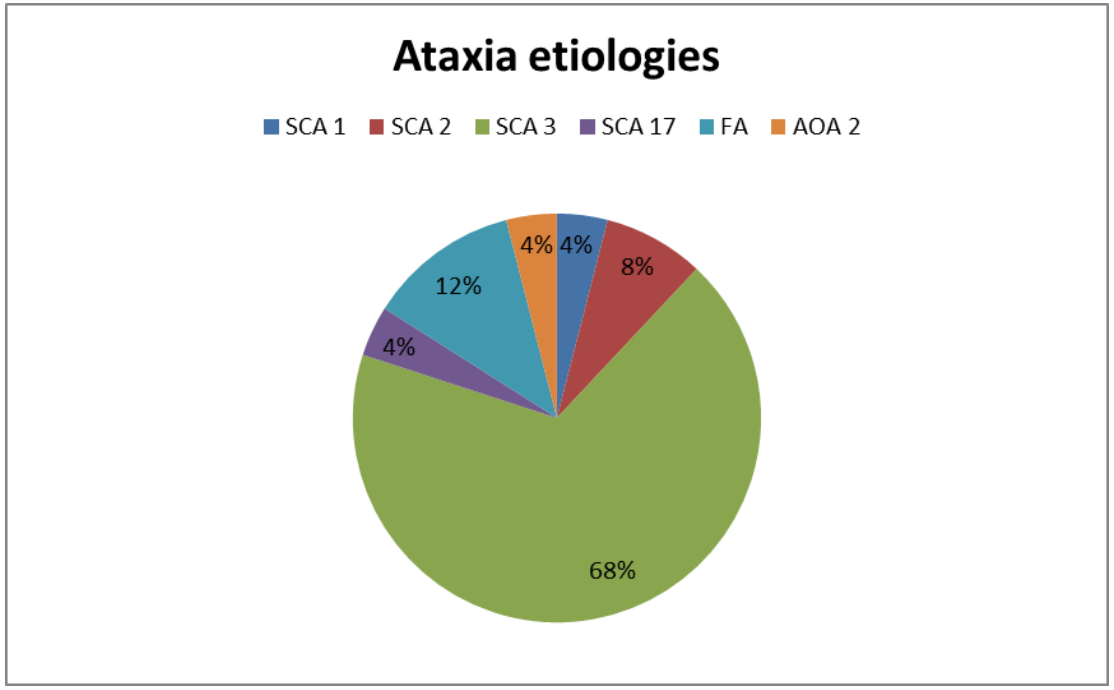
ID	Family history	Diagnosis	Genetic findings
1	No	SCA1	CAG repeats 33/36 *ATXN1*
2	AD	SCA2	*
3	AD	SCA2	CAG repeats 22/53 *ATXN2*
4	AD	SCA2	CAG repeats 21/41 *ATXN2*
5	AR	SCA3	CAG repeats 20/78 *ATXN3*
6	AR	SCA3	CAG repeats 14/76 *ATXN3*
7	AD	SCA3	CAG repeats 23/65 *ATXN3*
8	AD	SCA3	CAG repeats 23/67 *ATXN3*
9	AD	SCA3	CAG repeats 22/76 *ATXN3*
10	AD	SCA3	CAG repeats 21/68 *ATXN3*
11	AD	SCA3	*
12	No	SCA3	CAG repeats 21/72 *ATXN3*
13	AD	SCA3	CAG repeats 16/65 *ATXN3*
14	AD	SCA3	CAG repeats 14/80 *ATXN3*
15	AD	SCA3	CAG repeats 21/63 *ATXN3*
16	AD	SCA3	CAG repeats xx/63 *ATXN3*
17	AD	SCA3	CAG repeats 27/76 *ATXN3*
18	AD	SCA3	*
19	AD	SCA3	*
20	AD	SCA3	CAG repeats 23/69 *ATXN3*
21	AD	SCA 17	CAG repeats 22/46 *TBP*
22	AR	FA	GAA expansion *FTX* (pathological range), homozygous
23	AR	FA	GAA expansion *FTX* (pathological range), homozygous
24	AR	FA	GAA expansion *FTX* (pathological range), homozygous
25	AR	AOA2	SETX c.5929C>T p.Leu1977Phe
26	No	Negative	SCA 1 2 3 7 27b
27	AD	Negative	SCA 1 2 3 7 27b
28	AD	Negative	SCA 1 2 3 7 27b + WES
29	AD	Negative	Ataxia panel + WES
30	No	Negative	Ataxia panel
31	AD	Negative	SCA 1 2 3 7 27b
32	AD	Negative	SCA 1 2 3 7 27b

*No data. Ataxia panel: SCA1, SCA2, SCA3, SCA6, and SCA7. WES: Whole-exome sequencing.

(In Collaboration with UCL Queen Square Institute)


Inheritance Pattern
16%
84%
DOMINANT
RECESSIVE

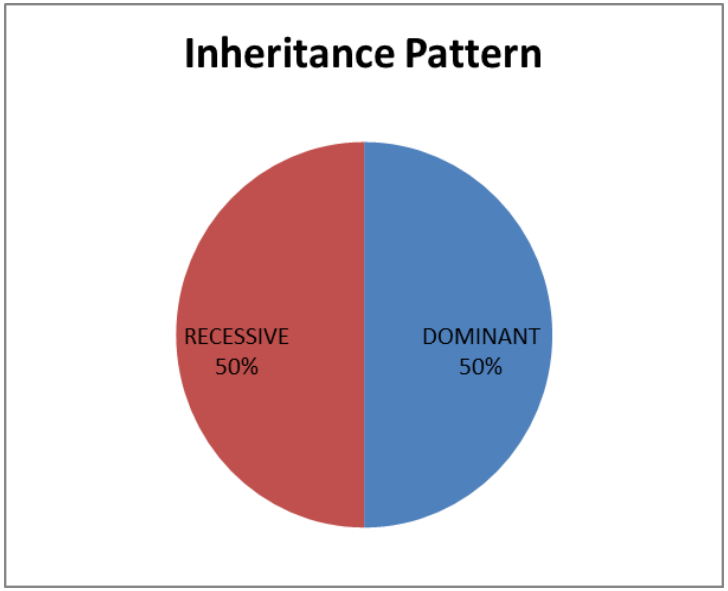

Autosomal Dominant
SCA 1
SCA 2
SCA 3
SCA 17
5%
4%
14%
77%

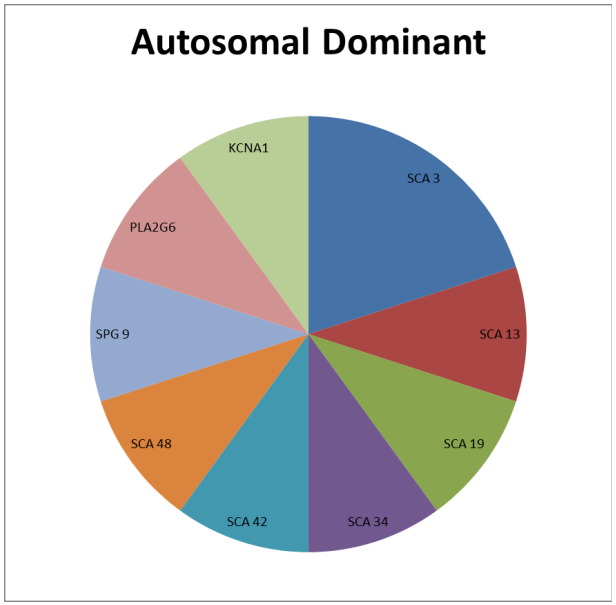

Ataxia etiologies
SCA 1
SCA 2
SCA 3
SCA 17
FA
AOA 2
4%
4%
8%
12%
4%
68%

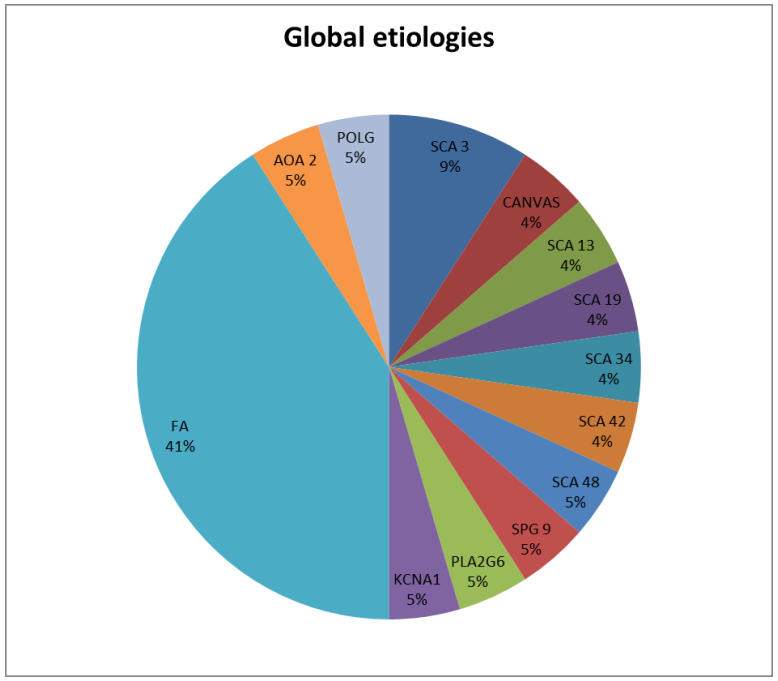
Newer Data

(Rare Diseases Center)

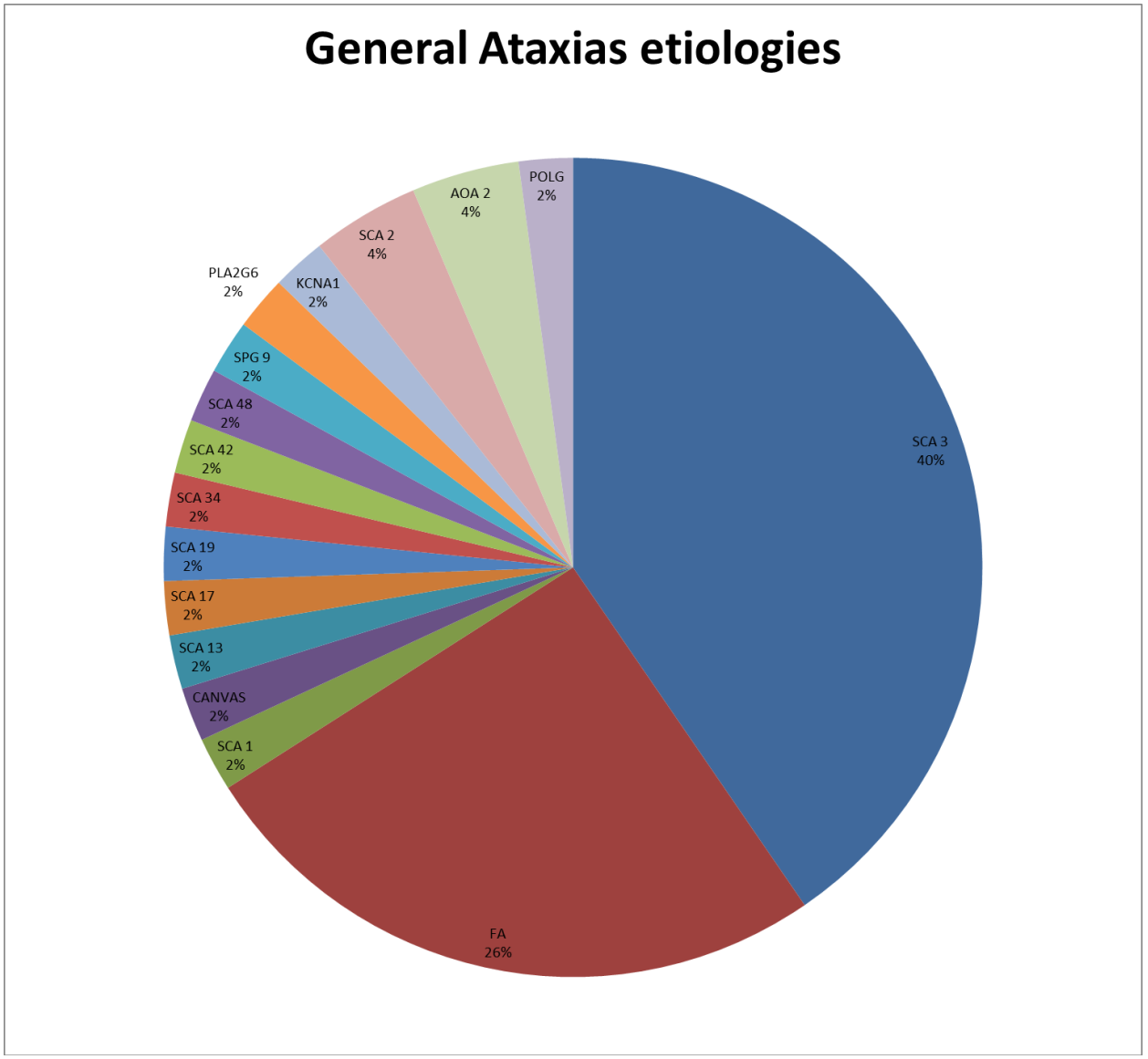
Patient	Age	Diagnosis
1	16	FA
2	19	FA
3	18	FA
4	16	FA
5	16	FA
6	40	FA
7		FA
8	41	AOA 2
9	38	SCA 3
10		SCA3
11	64	SCA 14
12	51	SCA 19
14	81	SCA 42
15	38	SCA 48
16	78	CANVAS
17	29	SPG 7
18	48	POLG
19	18	EA Type 1
20	33	EA Type 2
21	39	PLA2G6


Inheritance Pattern
RECESSIVE
50%
DOMINANT
50%


Autosomal Dominant
KCNA1
SCA 3
PLA2G6
SCA 13
SPG 9
SCA 19
SCA 48
SCA 42
SCA 34


Global etiologies
POLG
5%
SCA 3
9%
AOA 2
5%
CANVAS
4%
SCA 13
4%
SCA 19
4%
SCA 34
4%
SCA 42
4%
FA
41%
SCA 48
5%
SPG 9
5%
PLA2G6
5%
KCNA1
5%

Considering all the information…



Take Away Messages

- Historical point where diagnosis is linked to the technology availability
- Uruguay has a diverse landscape with a high prevalence of SCA3 and more uncommon Ataxias, which diagnosis are still limited by access to molecular studies
- Rare Ataxias, viewed as a group, have a prevalence that compels us to consider them in daily clinical practice.
- Constant epidemiological updates keep us alert to diagnose less prevalent diseases even more easily and frequently.

THANK YOU

Dr. Sue Ferraz
Lic. Paola Gonzalez
Dr. Rosario Guecaimburu