

Diagnosis Journey of Brazilian Friedreich's Ataxia patients



PERSONAL PROFILE:

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OBJECTIVE:

- Understand the disease burden and diagnostic odyssey of FA patients in Brazil



METHODOLOGY:

- Online survey sent to patients and legal guardians containing from 24 to 32 questions related to disease and diagnosis journey
- Comparison between classical and LOFA diagnosis journey was performed



RESULTS:

	Classical (n=66)	LOFA (n=16)	p-value
Mean age at first symptoms (years ± SD)	12.9 ± 5.2	32.2 ± 6.2	<0.0001
Number of physicians visited up to the diagnosis (n ± SD)	6.01 ± 3.8	7.2 ± 5.2	0.386
Diagnosis delay (years ± SD)	7.6 ± 6.8	7.6 ± 6.4	0.978
Mean age at FA diagnosis (years ± SD)	20.3 ± 8.6	39.8 ± 9.3	<0.0001
Misdiagnosis (%)	43.9 %	50%	0.396
Mean age (years ± SD)	34.2 ± 12.9	50.1 ± 10.5	<0.0001

Most reported first symptom: unsteadiness and gait abnormalities
 First specialty visited: Neurologists and Orthopedical surgeons



IMPORTANCE TO THE COMMUNITY:

- Understand the disease burden and diagnostic odyssey is crucial to accelerate diagnosis, avoid unnecessary tests and medicines, and provide suitable management to patients.