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REVIEW

Hereditary Ataxias in Argentina

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Abstract
Hereditary or genetic ataxias are hundreds of disorders characterized by large phenotypic, genetic, and epidemiological heterogeneity. In Argentina, 35 genetic ataxias have been identified, with SCA1 (ATX-A*TXN1*), SCA2 (ATX-*ATXN2*), SCA3 (ATX-*ATXN3*), and Friedreich ataxia (ATX-*FXN*) as the most prevalent causes, reflecting the epidemiology of most Western European countries, the main origin of immigration to the country. Genetic diagnostic studies of ataxia cohorts in Argentina have found high rates of undiagnosed patients, ranging from 65 to 82%. Deep phenotyping, comprehensive genetic testing, and knowledge of the prevalence of different genetic ataxias are essential for an accurate diagnostic and treatment approach in clinical practice. This narrative review proposes a targeted, tiered genetic diagnostic approach for undiagnosed patients based on the Argentinian epidemiological and healthcare system data. Future national efforts should support comprehensive screening studies on ataxia cohorts, including testing for repeat expansions in *RFC1* and *FGF14* genes. In addition, establishing a trial-ready patient registry for genetic ataxias, enhancing networking with international clinical and research initiatives, and developing specialized centers for interdisciplinary care of genetic ataxia patients are recommended.

Keywords Hereditary ataxia · Genetic testing · Epidemiology of ataxia · Diagnostic approach

Introduction

Hereditary or genetic ataxias are the most common cause of chronic ataxia, with over 400 disorders, most of which are autosomal dominant cerebellar ataxias (ADCA), also commonly referred to as spinocerebellar ataxias (SCAs) and autosomal recessive cerebellar ataxias (ARCA) [1, 2]. In about 140 disorders, ataxia is a predominant and consistent feature, variably accompanied by spastic paraplegia or movement disorders, such as dystonia, myoclonus, parkinsonism, or chorea [3, 4].

Argentina is a middle-income country with a population of 46 million people based on the 2022 national census [5]. The healthcare system is divided into three sectors: public, social security, and private [6]. Health subsectors are decentralized at provincial and municipal levels, where health planning and financing occur, leading to major fragmentation, inefficiency, and inequities [7]. Neurologists and other healthcare professionals may work in one or across multiple sectors, engaging solely in clinical practice or combining it with academic and research activities. Patients with ataxia generally have access to neurologists, particularly those practicing in neurology departments within hospitals, clinics, or specialized medical institutions. However, there are currently only a few movement disorders specialists with dedication and expertise in ataxia, but still no interdisciplinary groups of health and allied health professionals solely dedicated to treating patients with ataxia. Several laboratories, with either privately or publicly managed services at universities and hospitals, perform genetic tests for ataxia, such as targeted gene testing, next-generation sequencing (NGS), or repeat expansion detection methods [8, 9]. A list of publicly managed genetics laboratories, along with their locations in the country, can be found on the Argentina.gob.

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ar website [10]. Currently, there is no official list of privately managed genetics laboratories. Even though several genetics laboratories exist in the country, patients usually face barriers to being referred to genetic services or accessing genetic testing due to a lack of insurance coverage or high costs [11]. The vulnerable socioeconomic situation in Argentina over the past 25 years has exacerbated healthcare disparities and created barriers to genetic testing due to the continuous devaluation of local currency, foreign exchange restrictions, logistics challenges, and rising costs of genetic reagents. Possible solutions include advocating for coverage of genetic testing through private or non-governmental organizations and, in the long term, improving the country's socio-economic conditions to make genetic testing more accessible to patients. Limitations to genetic testing and access to geneticists and genetic counsellors also are common problems in most Latin American countries [12]. Genetic testing is key to ending patients' diagnostic 'odyssey', supporting genetic counselling, and enabling precise clinical actions, such as preventing comorbidities and effectively managing treatable forms of ataxia [13–15].

The objective of this narrative review was to describe the available data on the epidemiology of hereditary ataxias in Argentina. We searched our institutional database for the occurrence of the various causes of genetic ataxia, including genetically confirmed patients diagnosed at our center, a tertiary referral institution specializing in Neurology, receiving both walk-in consultations and referrals from throughout the country, as well as diagnosed patients referred from other institutions. Additionally, we conducted a literature search of PubMed from inception to November 2024, without language restrictions, using the terms 'ataxia,' 'spinocerebellar,' 'SCA,' 'epidemiology,' 'Argentina,' and 'genetic.' We also included additional publications identified in reference lists if they provided data on patients with genetically confirmed ataxia. The occurrence of the various causes of genetic ataxia was extracted from case reports, case series, and cohort screening studies. The genetic ataxias' mean frequencies were calculated using data from screening studies on ataxia cohorts.

Occurrence and Frequency of Genetic Ataxias

No studies have been conducted to determine the prevalence of genetic ataxias in Argentina for comparison with the global prevalence of SCAs (2.7/100,000), and ARCAs (3.3/100,000) [16]. Three cohort studies of unselected pediatric or adult patients with progressive ataxia, conducted in Buenos Aires, the capital city of Argentina, with coverage extending across the entire country, were identified (Table 1) [17–19]. Additionally, a retrospective study of a large cohort of 2,948 patients with movement disorders, in which whole exome sequencing (WES) was performed for diagnostic purposes in 54 patients, was also included (Table 1) [20].

Overall, ARCAs (18.3%) were more common than SCAs (15.9%) when screening both inheritance patterns within the same study sample [17]. Friedreich ataxia (ATX-*FXN*) was the most common ARCA (8.6%), followed by 19 other disorders, each with an average frequency of 1% or less (Fig. 1). Among SCAs, SCA2 (ATX-*ATXN2*) was the most frequent (6.6%), followed by SCA3 (ATX-*ATXN3*) (4.1%), and SCA1 (ATX-*ATXN1*) (3.2%). Other reported SCAs averaged frequencies of 1% or less (Fig. 1). This distribution of SCAs in Argentina reflects the common SCAs reported in Western European countries [21, 22], the primary origin of immigration to Argentina. SCA10 (ATX-*ATXN10*), characterized by progressive ataxia variably associated with epilepsy, is uncommon in Argentina, in contrast to other Latin American countries, such as Brazil or Peru, where it is among the most common SCAs [23–25]. SCA36 (ATX-*NOP56*), characterized by progressive ataxia usually combined with nystagmus, ptosis, gaze palsy, sensorineural hearing impairment, and lingual fasciculations represents the most common SCA in Costa da Morte (Galicia) and Valencia, Spain due to a founder effect [26]. Surprisingly, this genetic ataxia has been seldom reported, despite the large wave of immigration from Galicia to Argentina in the late 19th and early 20th centuries. This low diagnostic rate could be due primarily to repeat expansions in the *NOP56* gene not being included in most screening panels and its infrequent consideration by neurologists in the diagnostic approach for undiagnosed patients. Similarly, SCA38 (ATX-*ELOVL5*), a slowly progressive cerebellar ataxia variably associated with nystagmus, hyposmia, and pes cavus, and treatable with docosahexaenoic acid with several patients reported in Italy [27, 28], has been identified in only isolated cases. This is also surprising given the influx of Italian immigrants in the late 19th and early 20th centuries. Other SCAs, prevalent in some European countries, such as SCA8 (ATX-*ATXN8*) and SCA17 (ATX-*TBP*), have not been identified in screening studies or described in case reports to date.

The distribution of SCAs in Argentina differs from that observed in other Latin American countries [29]. To further explore these regional variations, Table 2 compares the relative frequencies of the most prevalent SCAs in Argentina, Brazil, Peru, Cuba, Venezuela, and Mexico. The main reasons for these differences include founder effects and variations in migration patterns [30], which contribute to distinct genetic backgrounds in some Latin American countries, such as Portuguese migration to Brazil, which explains the higher prevalence of SCA3 [29, 31]. Indigenous ancestry and founder effects also influence the prevalence of certain SCAs, like SCA10, which is more prevalent in Peru,

Table 1 Screening studies of genetic ataxias in Argentina

Author (Year)	City location of laboratory or medical institution (region covered)	Sample source (*n*)	Genetic testing	Genetic ataxias (%)	Diagnostic yield (%)
Millar Vernetti P, et al. (2022) [20]	Buenos Aires (all over Argentina)	Adult patients with movement disorders (54)	WES	*POLG* (1.8%), *ELOVL5* (1.8%), *SPG7* (1.8%), *ATP1A3* (1.8%), *PMM2* (1.8%)	26%
Guarnaschelli M, et al. (2020) [19]	Buenos Aires (all over Argentina)	Adult patients with progressive ataxia (272)	*ATXN1*, *ATXN2*, *ATXN3*, *CACNA1A*, *ATXN7*, *ATXN8*	*ATXN2* (8.1%), *ATXN3* (4.8%), *ATXN1* (3.7%), *CACNA1A* (0.7%), *ATXN7* (0.4%), *ATXN8* (0%)	18%
Perez Maturo J, et al. (2020) [17]	Buenos Aires (all over Argentina)	Pediatric and adult patients with progressive ataxia (334)	Multigene panels, WES, and WGS *FXN*, *ATXN1*, *ATXN2*, *ATXN3*, *CACNA1A*, *ATXN7*, *NOP56*	*FXN* (10.8%), *ATXN2* (6.0%), *ATXN3* (3.9%), *ATXN1* (2.4%), *PRNP* (1.2%), *NPC1* (1.2%), *CACNA1A* (0.9%), *ATM* (0.9%), *SCN2A* (0.9%), *STUB1* (0.6%), *APTX* (0.6%), *SETX* (0.6%), *SCARB2* (0.6%), *OPA1* (0.6%), *SYNE1* (0.6%), *SACS* (0.3%), *TPP1* (0.3%), *ATXN7* (0.3%), *AFG3L2* (0.3%), *NOP56* (0.3%), *CYP27A1* (0.3%), *KCNA2* (0.3%), *CC2D2A* (0.3%)	34%
Rodriguez-Quiroga S, et al. (2015) [18]	Buenos Aires (all over Argentina)	Pediatric and adult patients with progressive ataxia (387)	WES, Sanger *FXN*, *ATXN1*, *ATXN2*, *ATXN3*, *CACNA1A*, *ATXN7*, *ATXN8*, *TBP*	*FXN* (6.4%), *ATXN2* (5.7%), *ATXN3* (3.6%), *ATXN1* (3.6%), *PRNP* (1.4%), *CACNA1A* (0.7%), *ATXN7* (0.7%), *STUB1* (0.7%), *ATXN8* (0%), *TBP* (0%)	35%

WES: whole exome sequencing, WGS: whole genome sequencing

Mexico, and Brazil than in other regions [30, 32, 33], and SCA7, with a founder effect reported in Veracruz, Mexico [34, 35]. Also, the geographical disparities can be attributed to differences in healthcare access and the methodological heterogeneity of ataxia cohort screening studies, leading to varying diagnostic rates.

The diagnostic yield of genetic testing for undiagnosed ataxia patients varies globally due to methodological differences across screening studies but generally averages between 30 and 50% [18, 36–41]. A recent systematic literature review on the diagnostic yield of NGS tests for hereditary ataxias found a median diagnostic yield of 43% (IQR = 9.5–100%) [42]. Higher diagnostic yields were obtained for episodic ataxia (68%), late-onset ataxia (54%), parental consanguinity (52%), and presumed ARCA (63%) [42]. Cohort screening studies of ataxia cohorts in Argentina have found high rates of undiagnosed patients, ranging from 65 to 82% [17–19]. This may be due to the limited number of genes investigated or to other as-yet-unidentified genetic causes of ataxia, such as SCA27B (ATX-*FGF14*) and ATX-*RFC*1, which have been identified in recent years and for which commercial testing is not yet universally available or accessible. Neurologists should be aware of SCA27B (ATX-*FGF14*), caused by GAA repeat expansions in the *FGF14* gene in patients with progressive cerebellar ataxia with downbeat nystagmus, and also episodic ataxia, visual disturbances, vertigo, and dysarthria, often responding well to 4-aminopyridine [43, 44]. This disorder is an increasingly common late-onset SCA, with frequencies up to 60%, particularly in Western Europe and Canada [43, 44]. Additionally, ATX-*RFC*1, causing Cerebellar Ataxia with Neuropathy and Vestibular Areflexia Syndrome (CANVAS), was identified in 2019 [45] and can be as frequent as 16% in European patients with undiagnosed ataxia and up to 67% in patients with two or more features of CANVAS or ataxia with chronic cough [46].

Regarding other inheritance patterns than ARCA and ADCA, *FMR1* premutation expansions causing Fragile X-associated Tremor/Ataxia Syndrome (FXTAS) were identified in isolated cases, but no mitochondrial causes of ataxia were found. The occurrence of the 35 genetic ataxias identified in Argentina is shown in Fig. 1. No clusters of any specific genetic ataxia were found in particular regions of Argentina or among specific ethnicities. Regarding phenotypic aspects, published patients and those in our institutional database exhibited typical clinical manifestations,

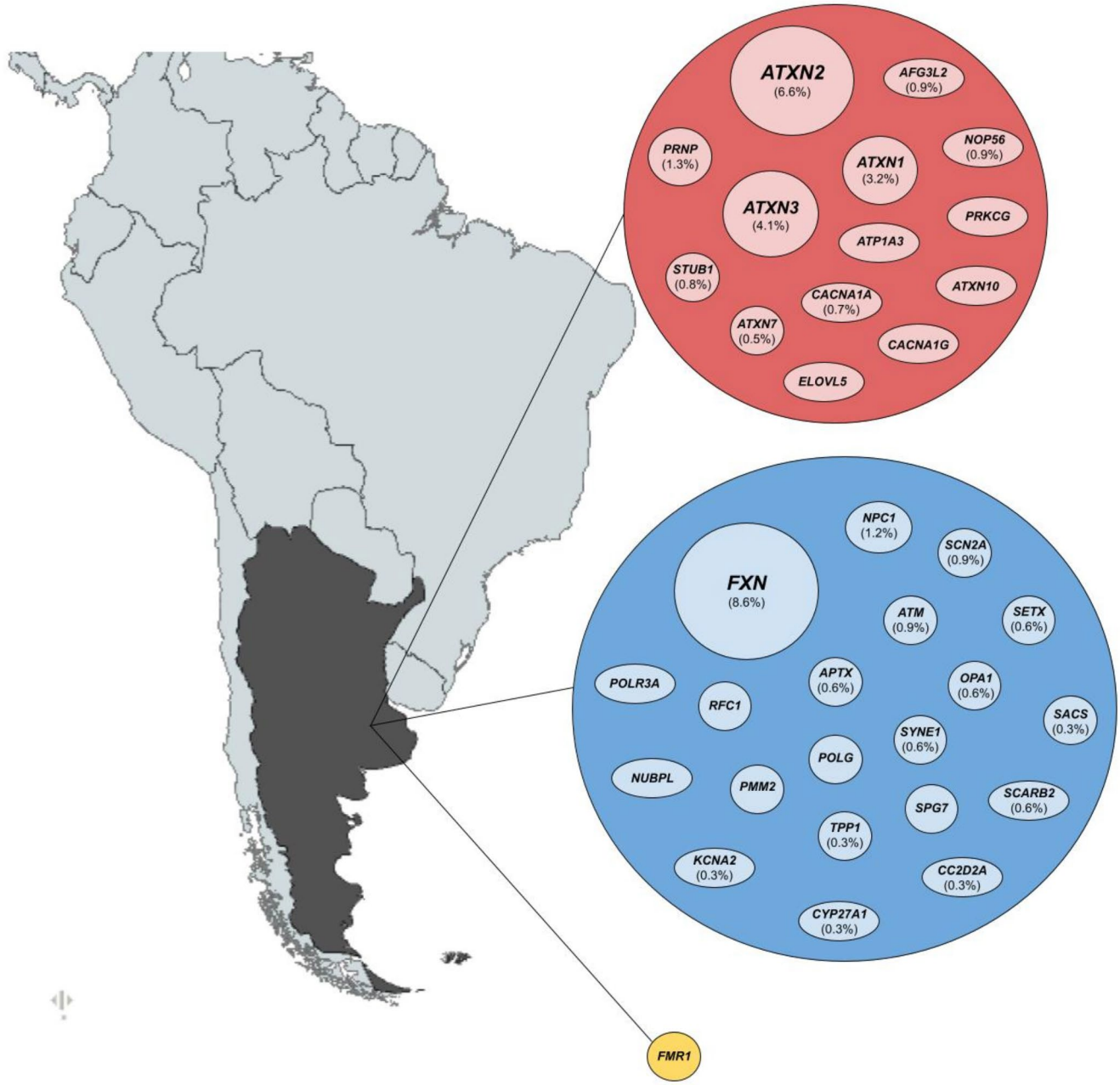

Fig. 1 Occurrence and frequency of genetic ataxias in Argentina. Red: SCAs; Blue: ARCAs; Yellow: X-linked inheritance. Mean frequencies were calculated based on data from the cited references [17–19]. Circle sizes are based on the total frequency of ataxias according to their inheritance pattern

with no differences from standard phenotypes or peculiar features related to age at onset or disease progression.

Diagnostic Approach

The diagnostic approach of undiagnosed patients with ataxia is challenging considering the large phenotypic and genetic heterogeneity of genetic ataxias [3, 47–52]. Factors such as age of onset, regional frequency, and ancillary test results are key to neurologists in clinical practice [2, 14, 21, 53, 54]. Clinical algorithms help narrow the differential diagnoses of ataxia often combined with other clinical manifestations [2, 54–58]. If the cause of the ataxia is presumed to be genetic, different testing approaches and algorithms have been proposed [51, 54, 59–61]. Based on the Argentinian epidemiological and healthcare system data provided here, we propose to follow in clinical practice the genetic diagnostic approach for undiagnosed patients as

Table 2 Comparison of the relative frequencies of the most prevalent SCAs in Latin America

SCA type	Argentina	Brazil	Peru	Venezuela	Cuba	Mexico
SCA1	2.4 − 3.7% [17–19]	0 − 8.9% [24, 95–104]	0.9% [23]	13% [105]	0.2% [106]	0% [35,107]
SCA2	5.7 − 8.1% [17–19]	0 − 18.4% [24, 95–104]	6.9% [23]	15.6% [105]	84.7% [106]	14.1 − 23.8% [35,107]
SCA3	3.6 − 4.8% [17–19]	34.1 − 92% [24, 95–104]	1.7 − 5.3% [23,108]	13.9% [105]	2.0 − 16.2% [106,109]	0 − 5.6% [35,107]
SCA6	0.7 − 0.9% [17–19]	0 − 5.2% [24, 95–104]	2.6% [23]	-	0% [106]	0% [35,107)
SCA7	0.3 − 0.7% [17–19]	1.7 − 11.9% [24, 97–104]	4.3% [23]	21.7% [105]	0.9% [106]	(3.5 − 85.9%) [35,107]
SCA10	-	0.4 − 18.3% 97–104	23.5% [23]	5.2% [105]	-	6.5% [35,107]

Frequencies, presented as percentages from ataxia cohort studies, include ranges when multiple values for a single gene were reported within a country

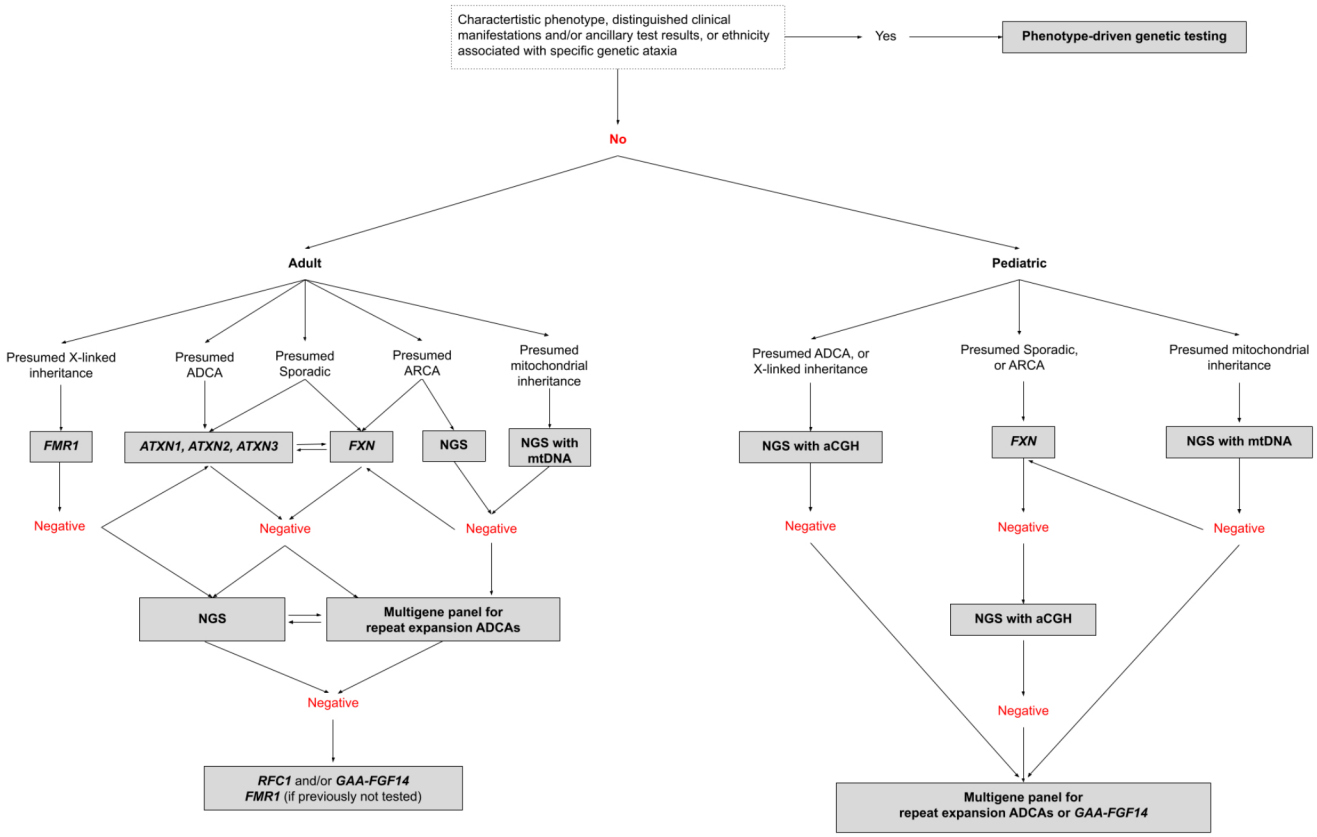

Fig. 2 Diagnostic approach for presumed genetic ataxias in Argentina. NGS: next-generation sequencing: it includes whole exome sequencing (WES), whole genome sequencing (WGS), or multigene panels of conventional variants; all of them should include copy number variations (CNVs) analyses. mtDNA: mitochondrial deoxyribonucleic acid, aCGH: array comparative genomic hybridization

illustrated in Fig. 2. A targeted, tiered approach is preferred, starting genetic testing either with a single focused gene detection for patients with particularly distinguished phenotypes (e.g. *FXN* for a Friedreich-like phenotype, *ATM* for conjunctival telangiectasias, etc.) or with a subset of genes (*ATXN1*, *ATXN2*, and *ATXN3*) for presumed SCA. Treatable causes of ataxia, such as ataxia with isolated vitamin E deficiency (ATX-*TTPA*), cerebrotendinous xanthomatosis (ATX-*CYP27A1*), Wilson disease (DYT/ATX-*ATP7B*), Niemann-Pick disease (ATX-*NPC1*, ATX-*NPC2*), or coenzyme Q10 deficiency type 4 (ATX-*ADCK3*), and, as recently suggested, Friedreich ataxia (ATX-*FXN*), should be prioritized based on clinical suspicion or ancillary test results [13, 15]. First-line laboratory tests that are accessible and can favor single-focused gene detection include serum levels of vitamin E (*TTPA*), α-fetoprotein (*ATM*, *APTX*, *SETX*, *PNKP*,

TDP1), cholesterol (*APTX*, *SETX*, *PNKP*, *TDP1*, *MTTP*, *CYP27A1*, *PMM2*), albumin (*APTX*, *PNKP*, *TDP1*, *PMM2*, *WDR73*), ceruloplasmin (Wilson's disease, aceruloplasminemia), and creatine kinase (*ADCK3*, *GOSR2*, *SIL1*, *SETX*). Additionally, certain distinctive neuroimaging patterns can indicate specific genetic ataxias, such as the hot cross bun sign observed in several SCAs [62], the middle cerebellar peduncle sign (ATX-*FMR1*) [63], bilateral hyperintensities of the dentate nuclei with a central hypointensity in the deep cerebellar nuclei related to deposition of hemosiderin and focal calcifications (ATX-*CYP27A1*) [64], superior cerebellar peduncles atrophy and hyperintensity (ATX-*FGF14*) [65], and the varied combination of superior cerebellar vermis atrophy, posterior mid-body corpus callosum thinning, bilateral hypointense pontine striations, hyperintense peri thalamic rims, and enlarged pons (ATX-*SACS*) [66]. A special consideration should be made for CANVAS (ATX-*RFC1*), and SCA27B (ATX-*FGF14*), for which no epidemiological data exists in Argentina; therefore, they should be investigated based on clinical suspicion or after other genetic causes were ruled out. First-tier testing of repeat expansion SCAs, including SCA6 (ATX-*CACNA1A*), SCA7 (ATX-*ATXN7*), SCA8 (ATX-*ATXN8*), SCA10 (ATX-*ATXN10*), SCA12 (ATX-*PPP2R2B*), SCA17 (ATX-*TBP*), SCA31 (ATX-*BEAN1*), SCA36 (ATX-*NOP56*), SCA37 (ATX-*DAB1*), and DRPLA (ATX-*ATN1*), which are more prevalent in other countries or regions, is not recommended due to cost-effectiveness unless there are strong clinical indications or complementary studies suggesting otherwise. NGS genetic testing, such as WES or multigene panels for conventional variants, as a first step in the diagnostic process, is suggested for presumed ARCA or mitochondrial inheritance (Fig. 2).

Future Perspectives

In clinical practice, genetic testing is no longer an option but a standard care, considering the need to end the patient´s diagnostic odyssey and appropriate management of treatable ataxias [13, 15, 67]. Friedreich ataxia, the most common genetic ataxia worldwide, recently received Food and Drug Administration (FDA) and European Medicines Agency (EMA) approvals for a new treatment, omaveloxolone, an Nrf2 activator that improves mitochondrial function [68, 69]. The studies on omaveloxolone in Friedreich ataxia demonstrate promising results as a potential treatment that slows disease progression as it significantly improved neurological function at 48 weeks on the modified Friedreich's Ataxia Rating Scale (mFARS) compared to placebo and showed persistent benefits in an open-label extension over three years, outperforming matched patients from a natural history study (FACOMS) [68, 70, 71]. This emerging clinical scenario requires neurologists to be aware of correctly and timely diagnosing Friedreich ataxia.

In Argentina, there is an underdiagnosis of globally common genetic ataxias whose molecular underpinnings have been identified in recent years, such as CANVAS (ATX-*RFC1*) and SCA27B (ATX-*FGF14*) [44, 45]. Future cohort screening studies are needed to determine the prevalence of these disorders, as well as SCA36 (ATX-*NOP56*), which is common among Spanish people from Galicia, a region with significant emigration to Argentina. The lack of studies on the prevalence of genetic ataxias in Argentina can be attributed mainly to limited research funding, the absence of centralized patient registries, and competing public health priorities. Potential strategies to address this gap include implementing cost-effective screening methods, such as targeted gene panels and sequencing strategies tailored to the population's specific needs, establishing partnerships with international collaborative studies and networks, and integrating epidemiological data collection into existing healthcare systems. Additionally, sharing laboratory and clinical genomic data is essential for enhancing genetic healthcare [41, 72–74]. Partnering with international research platforms, such as The Ataxia Global Initiative (AGI) (https://ataxia-global-initiative.net/), The Movement Disorders Society (MDS) Ataxia Study Group (https://www.movementdisorders.org), or the Pan American Hereditary Ataxia Network (PAHAN) [75], is vital for advancing research and improving clinical care of patients with genetic ataxia. Additionally, developing a trial-readiness national registry of genetic ataxias that is compatible with existing international multicenter registries or consortia is crucial for epidemiological research [76–78]. Establishing such a registry requires standardized clinical outcome assessments and systematic follow-up of patients, utilizing validated rating scales [79]. This approach will enhance natural history studies, collect real-world data, and promote collaboration between clinicians and researchers, ultimately leading to improved understanding and advancements in the field of ataxia [80–85]. Such a comprehensive, systematic, and interdisciplinary approach is most effectively conducted in specialized Ataxia Centers or Clinics [78, 79, 86]. Comparisons between specialist Ataxia Centers and non-specialist services in terms of healthcare have found that the former provides better management, more personalized care, and higher patient satisfaction, with no differences in costs [87–89]. It is recommended to maintain a close relationship with national ataxia patient advocacy and support groups in Argentina, such as ATAR (https://atar.org.ar/) and GPATAX (https://www.gpatax.org/), or the national federation of rare diseases (FADEPOF) (https://fadepof.org.ar/), as well as with social media outreach groups like AtaxiasARG (https://x.com/ataxiasarg?lang=es). Identifying and removi

ng barriers to accessing neurologic evaluation and genetic testing is crucial for advancing healthcare equity and providing optimal care to patients with ataxia. Furthermore, the implementation of advanced genetic testing techniques such as Long-Read Sequencing (LRS) in Argentina has the potential to transform the genetic diagnosis of hereditary ataxias by overcoming the inherent limitations of short-read sequencing methods like WES and short-read Whole-Genome Sequencing (WGS) in detecting repeat expansions, structural variants, and deep intronic mutations, ultimately increasing diagnostic yields in ataxia cohorts [90–92]. LRS offers significant advantages for detecting and characterizing repeat expansions while simultaneously identifying point mutations, copy number variations (CNVs), and noncoding pathogenic variants, potentially refining ataxia genetic testing workflow [93]. Given its emerging role in ataxia genetics, LRS has the potential to become the preferred first-line genetic test for undiagnosed ataxia cases; however, further optimization is needed before clinical implementation to ensure accuracy, scalability, and cost-effectiveness [93, 94].

Summary

The most prevalent genetic ataxias in Argentina are Friedreich ataxia (ATX-*FXN*), SCA1 (ATX-*ATXN1*), SCA2 (ATX-*ATXN2*), and SCA3 (ATX-*ATXN3*). Future national efforts should focus on large cohort screening studies using NGS and repeat expansion testing including recently identified common genetic ataxias, establishing a trial-ready patient registry for genetic ataxias, enhancing networking with international clinical and research initiatives, and developing specialized centers for interdisciplinary care of patients with genetic ataxia.

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Author Contributions Author contributions: Idea for the article: MR and MMLiterature search and data analysis: MRDrafted the manuscript: MRCritically revised the work: MMAll the authors approved the final manuscript as submitted and agreed to be accountable for all aspects of the work.

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Data Availability No datasets were generated or analysed during the current study.

Declarations

Ethical Approval this article does not involve any patient-derived clinical data, so there is no ethics involved.

Competing Interests MR has served on an advisory board for Biogen and has received honoraria for talks from the International Parkinson and Movement Disorder Society and Biogen. MM has no competing interests related to this work.

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