PAHAN survey on the care for ataxics in the Americas and the Caribbean

From the Pan-American Hereditary Ataxia Network

This survey is an initiative of PAHAN (https://pan-american-hereditary-ataxia-network-pahan7.webnode.com/). We are sending it to health professionals who care for ataxics in American countries, especially from Latin America. Our goals are to help define (1) the approximate number of ataxics already identified in our countries, (2) the access that our populations have to diagnostic, genetic counseling, pre-symptomatic testing and rehabilitation facilities related to inherited ataxias, and their cost coverage; (3) if there are any health policies set for identification and prevention, diagnosis and early intervention; (4) the availability of training courses (residency, fellowships, etc) in HA for health professionals; and (5) how is the support to local clinical and experimental research on hereditary ataxias. With this, we intend to approach all professionals involved with these conditions in our countries, so that together we can prepare a general diagnostic framework and a set of recommendations on how to improve the care of hereditary ataxia in our communities. Please feel free to choose the questions below. You will see that none are mandatory and that there are different types of questions, so you can choose the ones that are easier to be answered, according to your context. We are very grateful for your time and your participation in this poll.

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Nome

First name Surname (last name)

E-mail

exemplo@exemplo.com

Professional address (institution or private clinic)

first line: name of the institution, if suitable

second line: street address

Cidade País

ZIP code

Phone Number (Country, Province, Number)

WhatsApp (if you wish to be included)

for a whatsApp group

Do you want to become a provisional member of PAHAN? As soon as possible, we will hold a debate on the topic.

○ Sim

○ Not yet

You are

○ a neurologist
○ a clinical geneticist
○ other doctor (physician)
○ a genetic counselor
○ a professional in the laboratory diagnosis of inherited ataxias
○ a rehabilitation professional
○ a nurse
○ a social worker
○ outro

Your answers to this survey will tell about your experience

○ in a public service (public health system). You will be the only respondent about your service
○ in a public service (public health system). It is possible that other colleagues of mine will respond to this survey based on our common experience.
○ in your private clinic (including health insurance patients). You will be the only respondent about your clinics
○ in your private clinic (including health insurance patients). It is possible that other colleagues of mine will respond to this survey based on our common experience.

How many cases of ataxia are in your current care?

	Number of individuals	Number of families
SCAs (autosomal dominant cerebellar ataxias)		
Isolated/sporadic ataxic cases, or clearly autosomal recessive ataxias		
non-genetic ataxias		

Can you better define your current cases?

	Number of individuals	Number of families
SCA1		
SCA2		
SCA3/MJD		
SCA6		
SCA7		
SCA8		

SCA10		
SCA12		
SCA17		
DRPLA		
SCA21		
SCA36		
Friedreich ataxia		
Ataxia-telangiectasia		
AOA1		
AOA2		
AOA3		
AOA4		
ARSACS		
CANVAS		

Another hereditary ataxia?

The above data are

○ approximated values
○ exact values, obtained from a local survey

How many cases of ataxia you followed throughout your clinical practice, in the last 20 or less years?

	Number of individuals	Number of families
SCAs (autosomal dominant cerebellar ataxias)		
isolated/sporadic ataxic cases, or clearly autosomal recessive ataxias		
non-genetic ataxias		

The above data are

○ approximated values
○ exact values, obtained from a local survey

Can you better define the ataxia cases you followed throughout your clinical practice?

	Number of individuals	Number of families
SCA1		
SCA2		
SCA3/MJD		
SCA6		
SCA7		
SCA8		
SCA10		
SCA12		
SCA17		
DRPLA		
SCA21		
SCA36		
Friedreich ataxia		
Ataxia-telangiectasia		
AOA1		
AOA2		
AOA3		
AOA4		
ARSACS		
CANVAS		

Another hereditary ataxia?

☐

You and your patients have access to

	Directly available in public service	Available in public service only under court order (judicialization)	Available in the private sector (health insurance included)	We do not have access to it
Panels for expanded repeat ataxias	☐	☐	☐	☐
Panels for ataxias (point mutations, frameshift, etc)	☐	☐	☐	☐
NGS panel for ataxias	☐	☐	☐	☐
Exome sequencing	☐	☐	☐	☐
Genome sequencing	☐	☐	☐	☐

Check what additional assessments or exams you have available to include in the diagnostic work up for ataxia

	Directly available in public service	Available in public service only under court order (judicialization)	Available in the private sector (health insurance included)	We do not have access to it.
Brain MRI	☐	☐	☐	☐
Spinal cord MRI	☐	☐	☐	☐
CT Scan	☐	☐	☐	☐
Peripheral conduction studies (electroneuromyography)	☐	☐	☐	☐
Evoked potentials	☐	☐	☐	☐
Vitamin E in plasma	☐	☐	☐	☐
Alpha-fetoprotein	☐	☐	☐	☐
Optical coherence tomography (OCT)	☐	☐	☐	☐

Conventional ophthalmic evaluation - with fundoscopy and slit lamp	☐	☐	☐	☐
Evaluation of peripheral smear by hematologist (acanthocytes)	☐	☐	☐	☐
Abdominal imaging	☐	☐	☐	☐
Lysosomal enzymes in leukocytes and plasma	☐	☐	☐	☐
Ceruloplasmin	☐	☐	☐	☐
Copper, 24h urine	☐	☐	☐	☐
Amino acids in blood or urine	☐	☐	☐	☐
Organic acids in urine	☐	☐	☐	☐
TSH	☐	☐	☐	☐
B12	☐	☐	☐	☐
anti-HIV	☐	☐	☐	☐
anti-HTLV1	☐	☐	☐	☐
Jejunum biopsy (for Whipple)	☐	☐	☐	☐
Anti-transglutaminase IgA	☐	☐	☐	☐
Quitotriosidase	☐	☐	☐	☐
Oxysterols	☐	☐	☐	☐
Filipin in fibroblasts' culture	☐	☐	☐	☐

Do you or your workgroup have access to a clinical protocol for ataxias with therapeutic guidelines?

☐ Yes, we follow an evidence-based protocol built after public consultation by the country's health authority.
☐ Yes, we follow a protocol built by our local institution
☐ Não

After diagnosis, do the patient and her/his family have access to genetic counseling sessions?

○

○ Não

After the diagnosis of the index case, do other family members have access to pre-symptomatic tests?

○ Sim
○ Não

The genetic counseling sessions are performed

☐ in the public health system
☐ in private clinics

The genetic counseling sessions are performed

☐ by a multidisciplinary team
☐ by the same doctor who take care of diagnosis

Do you want to comment about the above issues?

What rehabilitation services are available for ataxics in your community?

	Public Health System	private health clinics	We do not have access to it
physical therapy	☐	☐	☐
speech therapy	☐	☐	☐
occupational therapy	☐	☐	☐

How do you rate the availability of the following care for your ataxic patients:

	Adequate	Limited by scarcity of professionals or structure	Unavailable
Clinical diagnosis and follow-up visits	○	○	○
Laboratory work up	○	○	○
Genetic Counseling	○	○	○
Physical therapy	○	○	○

Speech therapy	○	○	○
Occupational therapy	○	○	○

What molecular studies are performed in your laboratory/institution?

	Funded by the public health system	Private service (including health insurance)	No available in our institution
SCA1	☐	☐	☐
SCA2	☐	☐	☐
SCA3/MJD	☐	☐	☐
SCA6	☐	☐	☐
SCA7	☐	☐	☐
SCA8	☐	☐	☐
SCA10	☐	☐	☐
SCA12	☐	☐	☐
SCA17	☐	☐	☐
DRPLA	☐	☐	☐
FRDA	☐	☐	☐
ATM	☐	☐	☐
AOA1	☐	☐	☐
AOA2	☐	☐	☐
AOA3	☐	☐	☐
AOA4	☐	☐	☐
ARSACS	☐	☐	☐
CANVAS	☐	☐	☐
NGS panel	☐	☐	☐
Exome sequencing	☐	☐	☐
Genome sequencing	☐	☐	☐

Another molecular analysis?

Do you supervise students in research activities related to ataxia projects?

	The state pays for the student's education	The student pays for their education
undergraduate students	☐	☐
MSc students	☐	☐
PhD students	☐	☐
Post-doc students	☐	☐

Please check if you are conducting research project(s) on the following subjects related to hereditary ataxias

	Clinical studies (on human data), with public funding	Clinical studies, sponsored by corporations/private companies	Experimental studies in cells or other models, with public funding	Experimental studies in cells or other models, sponsored by corporations/private companies
SCA1	☐	☐	☐	☐
SCA2	☐	☐	☐	☐
SCA3/MJD	☐	☐	☐	☐
SCA6	☐	☐	☐	☐
SCA7	☐	☐	☐	☐
SCA8	☐	☐	☐	☐
SCA10	☐	☐	☐	☐
Other SCAs	☐	☐	☐	☐
Friedreich ataxia	☐	☐	☐	☐
Ataxia-telangiectasia or any AOA	☐	☐	☐	☐
ARSACS	☐	☐	☐	☐

CANVAS	☐	☐	☐	☐
Other	☐	☐	☐	☐

Another subject under your research lines related to ataxia?

☐

In your opinion, what must be improved in your state or country for the care of ataxic people? Number from 1 to 10 in order of priority. You need more and better:

	Priority
Doctors	☐
GC professionals	☐
Rehabilitation professionals	☐
Molecular tests	☐
MRI	☐
Clinical protocols and therapeutic guidelines	☐
Physical infrastructure for clinical care	☐
Physical infrastructure for rehabilitation	☐
Funding for local scientific research	☐
Professional training	☐

We intend to present the results of this survey to all participants by email, and also to health authorities in each region. If the results are robust, they might result in scientific communications as well. We thank you again for your participation.