GENÉTICA
SERVIÇO DE GENÉTICA MÉDICA
HOSPITAL DE CLÍNICAS DE PORTO ALEGRE

UFRGS
UNIVERSIDADE FEDERAL DO RIO GRANDE DO SUL

HOSPITAL DE CLÍNICAS
PORTO ALEGRE RS

ATAXIA GLOBAL INITIATIVE
worldwide platform for clinical research in ataxias

In Latin America

Hereditary ataxias: the 30 years Porto Alegre experience

Porto Alegre, May 7, 2026

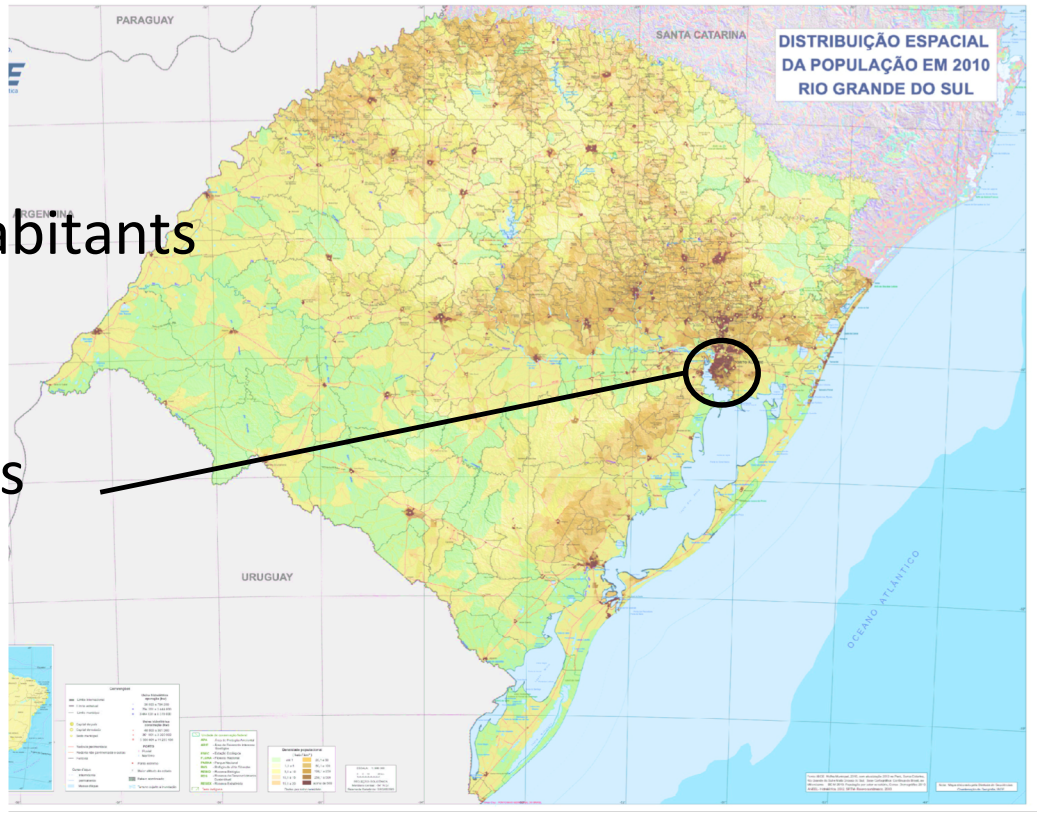
Laura Bannach Jardim
DMI, FAMED, UFRGS
SGM, HCPA

I worked at Medical Genetics Service (SGM), Hospital de Clinicas de Porto Alegre (HCPA), since 1987.

SGM-HCPA is the reference center for the care of ataxia patients in the Public Health System SUS, Rio Grande do Sul

RS has 11 million inhabitants

Porto Alegre has
1,5 million inhabitants



1989

I identified the first ADCA families in our region

1992 to 1996

Discoveries relating SCA1, SCA2, SCA3, SCA6 and SCA7 to CAGexp

1997

Jorge Sequeiros helped me sending the first ADCA families to be tested for SCA1, SCA2, SCA3 and SCA6 at McGill Uni.

2001

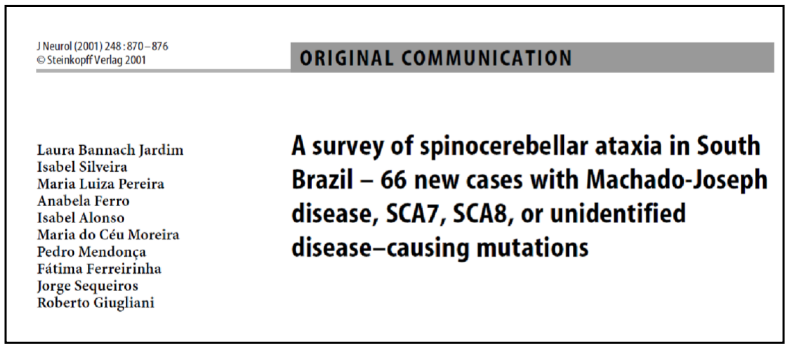
Maria Luiza Saraiva Pereira standardized the diagnostic technique for SCA3 and other CAGexp diseases.

Tests were performed in-house since then.

A large number of SCA families were looking for care.

Since 2000 we detected a large population of Machado-Joseph disease (SCA3/MJD) living in Porto Alegre and other cities.

By 2020, 760 symptomatic subjects were living in RS.



J Neurol (2001) 248 : 870 – 876
© Steinkopff Verlag 2001

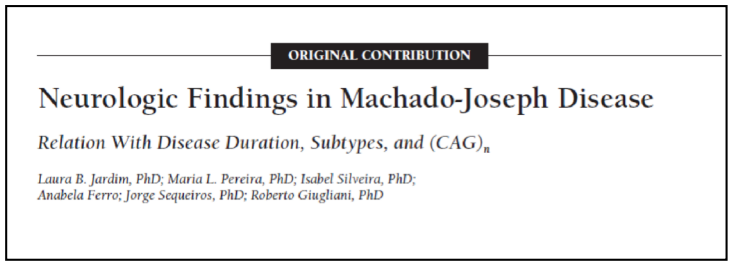
ORIGINAL COMMUNICATION

Laura Bannach Jardim
Isabel Silveira
Maria Luiza Pereira
Anabela Ferro
Isabel Alonso
Maria do Céu Moreira
Pedro Mendonça
Fátima Ferreirinha
Jorge Sequeiros
Roberto Giugliani

A survey of spinocerebellar ataxia in South Brazil – 66 new cases with Machado-Joseph disease, SCA7, SCA8, or unidentified disease–causing mutations

Machado–Joseph disease in South Brazil: clinical and molecular characterization of kindreds

Jardim LB, Pereira ML, Silveira I, Ferro A, Sequeiros J, Giugliani R. Machado-Joseph disease in South Brazil: clinical and molecular characterization of kindreds. Acta Neurol Scand 2001: 104: 224–231. © Munksgaard 2001.

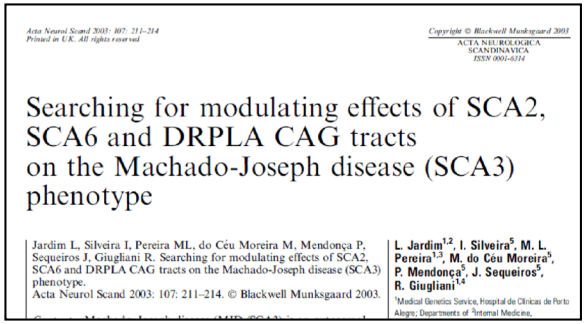


ORIGINAL CONTRIBUTION

Neurologic Findings in Machado-Joseph Disease

Relation With Disease Duration, Subtypes, and $(CAG)_n$

Laura B. Jardim, PhD; Maria L. Pereira, PhD; Isabel Silveira, PhD; Anabela Ferro; Jorge Sequeiros, PhD; Roberto Giugliani, PhD



Searching for modulating effects of SCA2, SCA6 and DRPLA CAG tracts on the Machado-Joseph disease (SCA3) phenotype

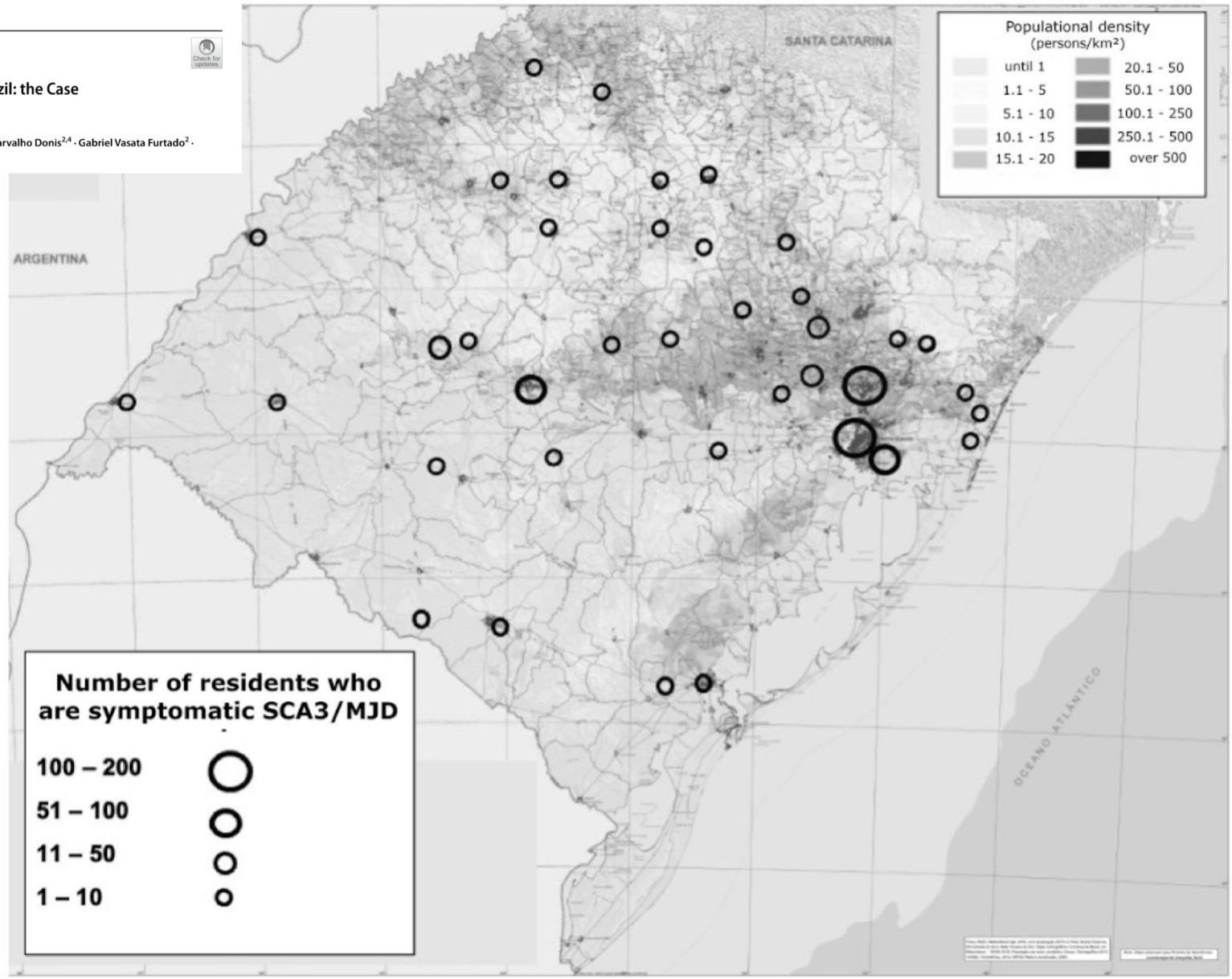
Jardim L, Silveira I, Pereira ML, do Céu Moreira M, Mendonça P, Sequeiros J, Giugliani R. Searching for modulating effects of SCA2, SCA6 and DRPLA CAG tracts on the Machado-Joseph disease (SCA3) phenotype. Acta Neurol Scand 2003: 107: 211–214. © Blackwell Munksgaard 2003.

The Cerebellum
https://doi.org/10.1007/s12311-022-01404-5

ORIGINAL ARTICLE

Diagnostic Delay of Hereditary Ataxias in Brazil: the Case of Machado-Joseph Disease

Jordânia dos Santos Pinheiro[1,2] · Lucas Schenatto Sena[2,3] · Karina Carvalho Donis[2,4] · Gabriel Vasata Furtado[2] · Maria Luiza Saraiva-Pereira[2,3,4,5] · Laura Bannach Jardim[2,3,4,6]



SCA3/MJD prevalence in RS: 7/100.000

doi.org/10.1007/s12311-022-01404-5

Brazil territory after Madrid treaty, 1750

Azorean settlers sent to South Brazil since 1770.

Sixty Azorean couples founded Porto Alegre in 1772.

Our scientific production related to CAGexp diseases (SCA3, SCA2, SCA7, SCA10, HD) included:

1) Frequency reports
2) Clinical studies
   - Age at onset, clinical, biochemical, molecular characterization
   - Design of a clinical scale
   - Statistical modelling
   - Natural history: survival estimates, cohort studies
3) Formal genetics: selective forces, mutation origins
4) Modifying factors affecting the phenotype
   - Genes
   - Environment
5) Biomarkers
   - Circulating proteins
   - Neurophysiology
   - Neuroimaging
   - Nutritional pattern
6)Management
   - Open label trials
   - Double blind clinical trial on lithium
   - Pre-symptomatic testing.

Some collaborative projects with bilateral public funding

fct

Jorge Sequeiros, Porto

Manuela Lima, Azores

Nuffic

Harm Kampinga, Groningen

DAAD

Thorsten Schmidt, Tübingen

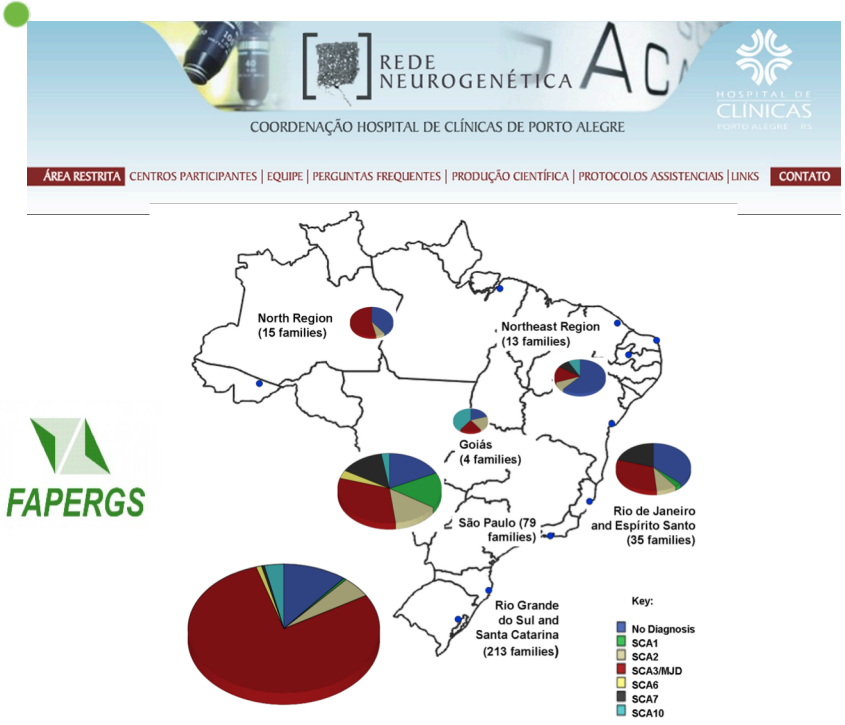
NIH
National Institutes of Health

Tetsuo Ashizawa, Houston

CNPq

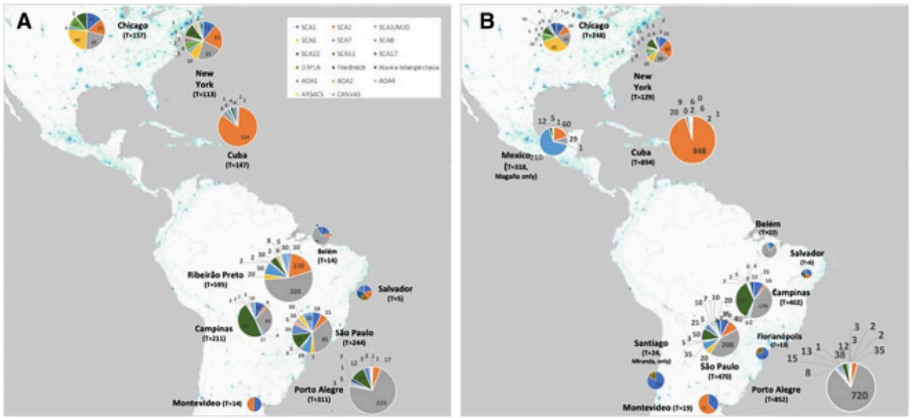
CAPES

And some networking to increase access



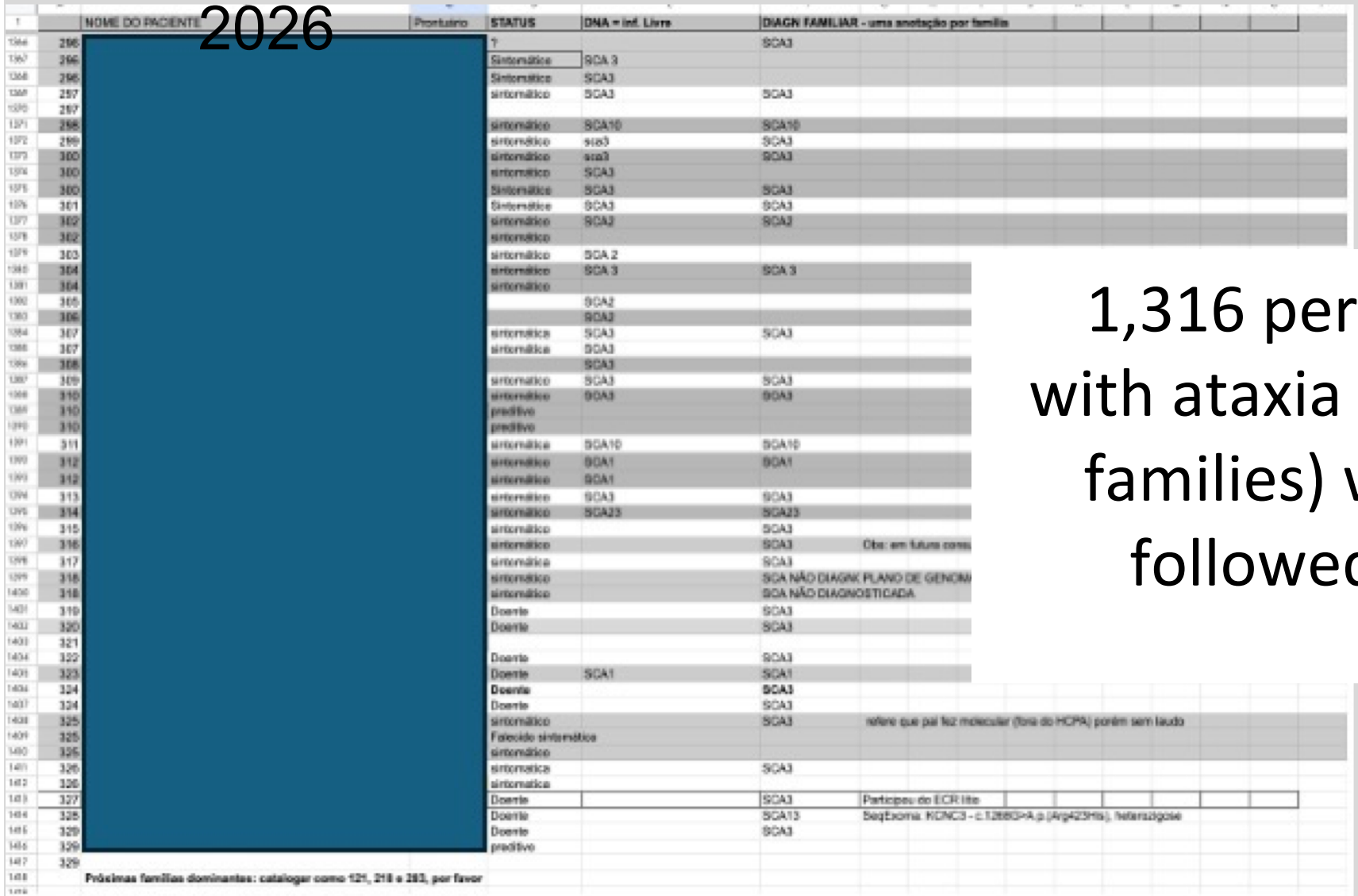
FAPERGS

pahan
pan american hereditary ataxia network



ATAXIA GLOBAL INITIATIVE
worldwide platform for clinical research in ataxias

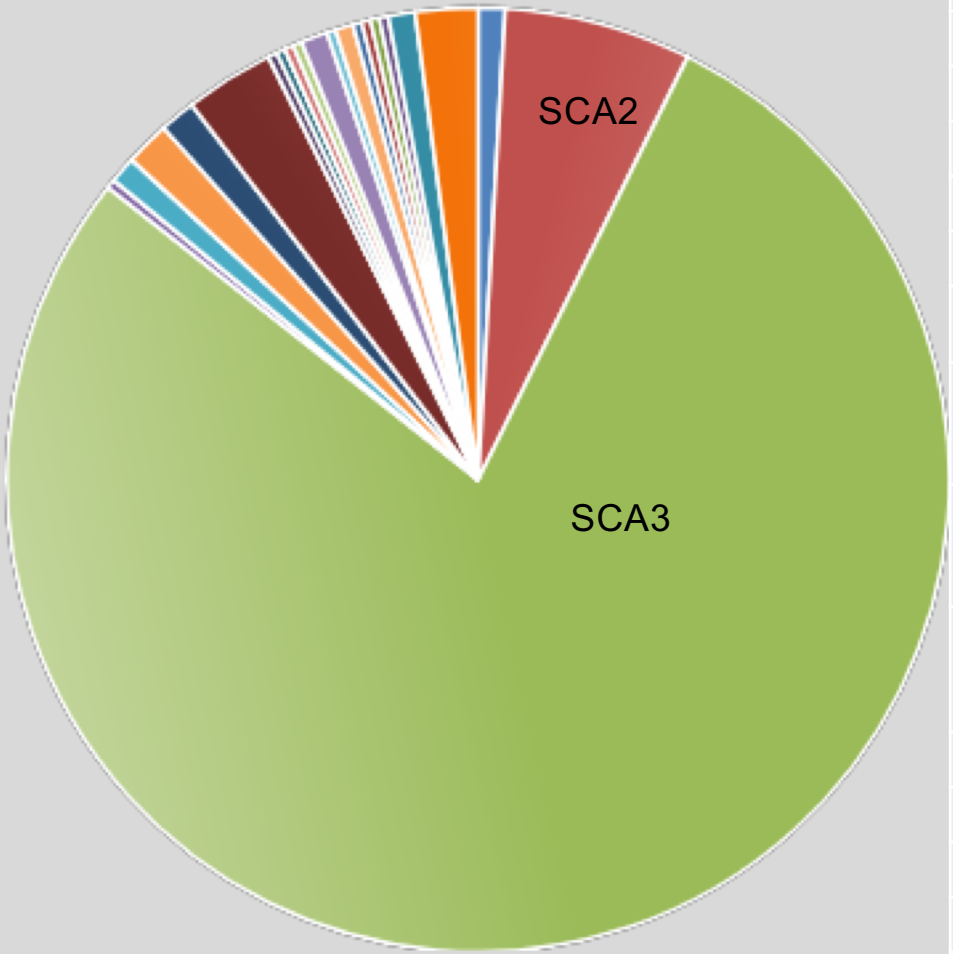
SCAs diagnosed at SGM-HCPA between 2000 and 2026



	NOME DO PACIENTE	Prontuário	STATUS	DNA = Int. Livre	DIAGN FAMILIAR - uma anotação por família	
1364	266		?		SCA3	
1367	266		Sintomático	SCA 3		
1368	266		Sintomático	SCA3		
1369	267		sintomático	SCA3	SCA3	
1370	267					
1371	268		sintomático	SCA10	SCA10	
1372	299		sintomático	sca3	SCA3	
1373	300		sintomático	sca3	SCA3	
1374	300		sintomatico	SCA3		
1375	300		Sintomático	SCA3	SCA3	
1376	301		Sintomático	SCA3	SCA3	
1377	302		sintomático	SCA2	SCA2	
1378	302		sintomático			
1379	303		sintomático	SCA 2		
1380	304		sintomático	SCA 3	SCA 3	
1381	304		sintomático			
1382	305			SCA2		
1383	306			SCA2		
1384	307		sintomatica	SCA3	SCA3	
1385	307		sintomática	SCA3		
1386	308			SCA3		
1387	309		sintomatico	SCA3	SCA3	
1388	310		sintomático	SCA3	SCA3	
1389	310		preditivo			
1390	310		preditivo			
1391	311		sintomática	SCA10	SCA10	
1392	312		sintomático	SCA1	SCA1	
1393	312		sintomático	SCA1		
1394	313		sintomático	SCA3	SCA3	
1395	314		sintomático	SCA23	SCA23	
1396	315		sintomático		SCA3	
1397	316		sintomático		SCA3	Obs: em futuro cons
1398	317		sintomática		SCA3	
1399	318		sintomático		SCA NÃO DIAGNÓ	PLANO DE GENOM
1400	318		sintomático		SCA NÃO DIAGNOSTICADA	
1401	319		Doente		SCA3	
1402	320		Doente		SCA3	
1403	321					
1404	322		Doente		SCA3	
1405	323		Doente	SCA1	SCA1	
1406	324		**Doente**		**SCA3**	
1407	324		Doente		SCA3	
1408	325		sintomático		SCA3	refere que pai fez molecular (fora do HCPA) porém sem laudo
1409	325		Falecido sintomático			
1410	325		sintomático			
1411	326		sintomatica		SCA3	
1412	326		sintomatica			
1413	327		Doente		SCA3	Participou do ECR I lite
1414	328		Doente		SCA13	SeqExoma: KCNC3 - c.1288G>A,p.(Arg423His), heterozigose
1415	329		Doente		SCA3	
1416	329		preditivo			
1417	329					
1418	Próximas famílias dominantes: catalogar como 121, 218 e 283, por favor					

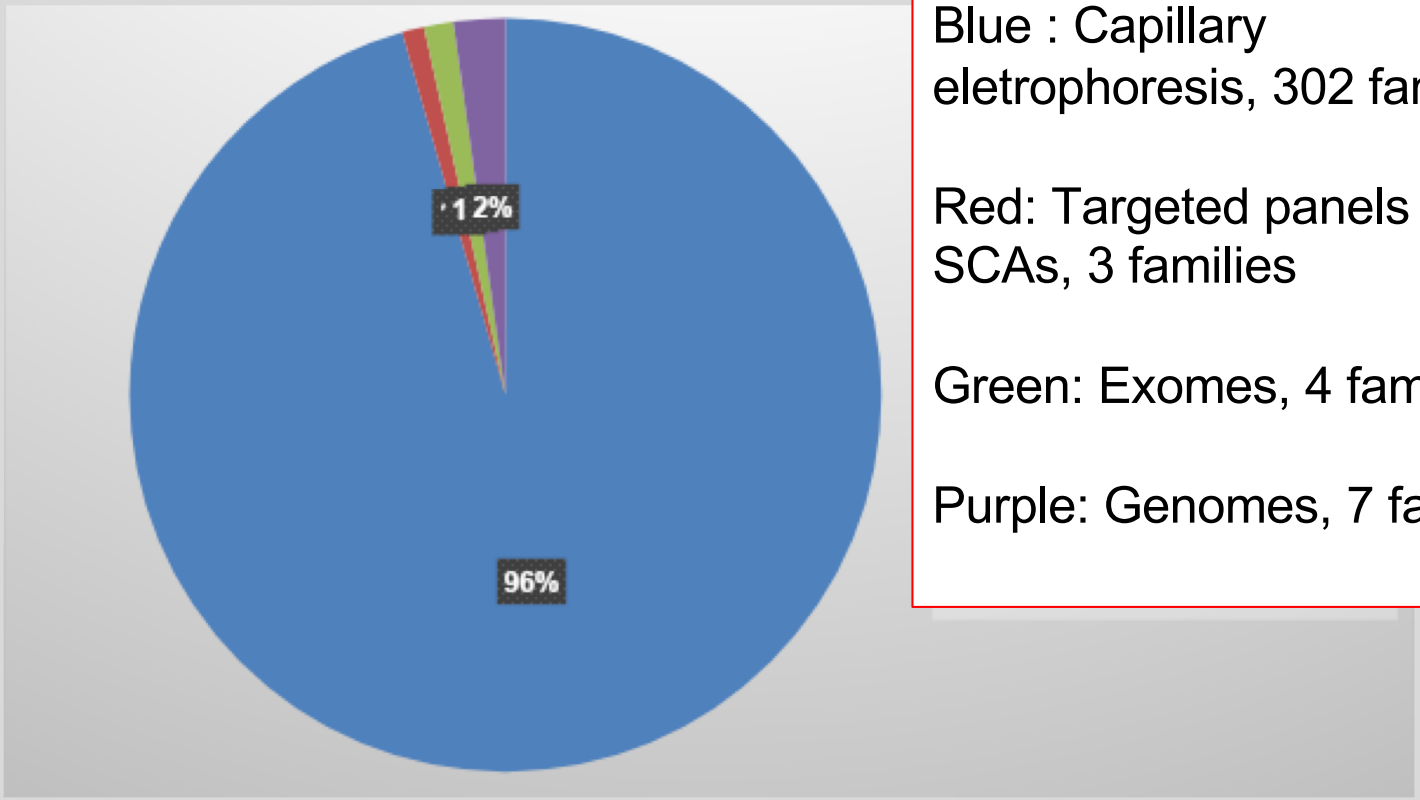
1,316 persons with ataxia (328 families) were followed up.

SCAs diagnosed at SGM-HCPA between 2000 and 2026



Diagnósticos	N de famílias
SCA3/MJD	256
SCA2	21
SCA10	10
SCA7	5
SCA8	4
SCA1	3
SCA6	3
SCA27b	3
SPG7	2
SCA5	1
SCA13	1
SCA15	1
SCA19	1
SCA23	1
SCA48	1
EA tipo 2	1
Gerstmann-Straussler-Scheinker	1
PARK8	1
HD	1
Perdas	7
Pending	4
Total	328

Diagnostic methods



Blue : Capillary eletrophoresis, 302 families.

Red: Targeted panels for SCAs, 3 families

Green: Exomes, 4 families

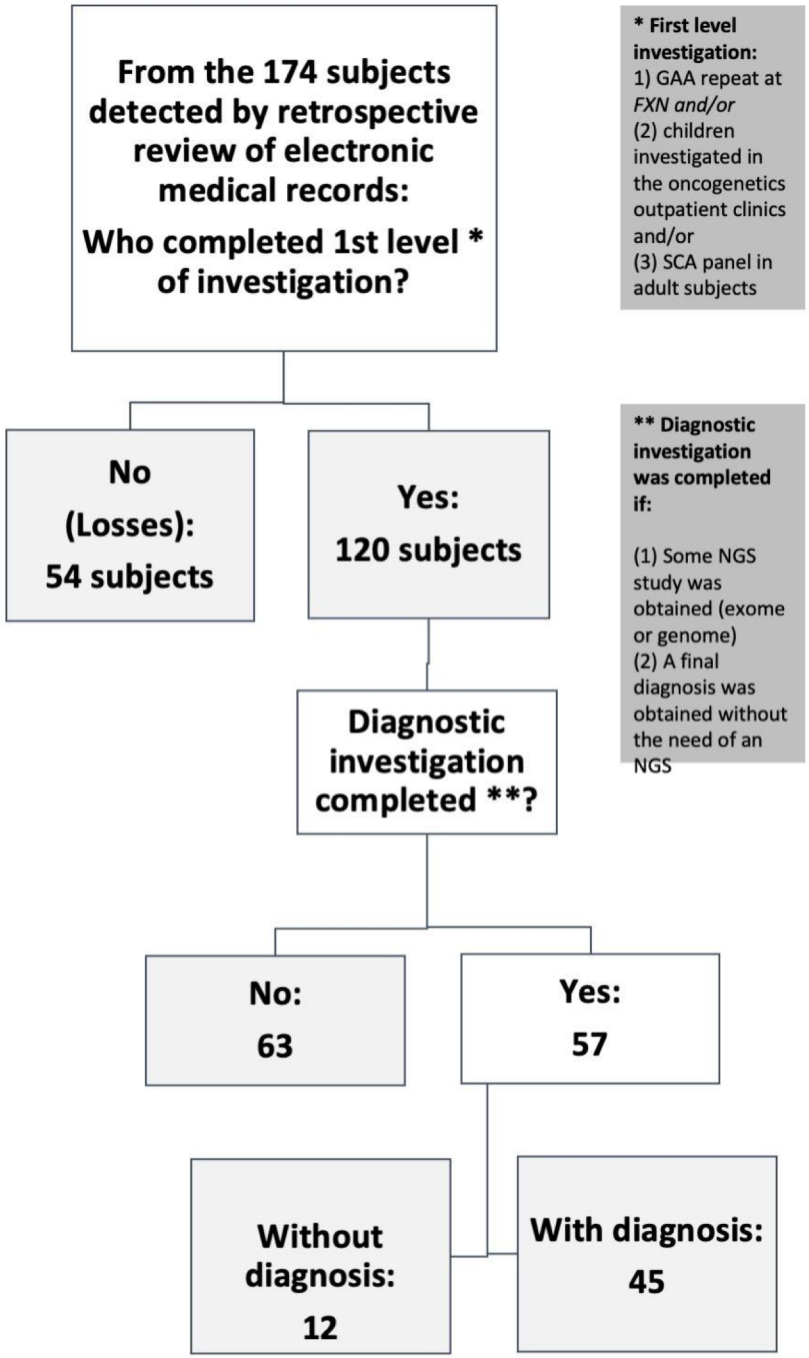
Purple: Genomes, 7 families

Recessive ataxias at SGM-HCPA

Retrospective review of subjects with ataxia,

without a vertical inheritance,

that started evaluation from 2003 to 2020 (Aschoff et al, in preparation)



Recessive ataxias at SGM-HCPA

First level of investigation:

GAA repeat at FXN or alpha-fetoprotein or SCA panel.

Diagnostic work was completed after:

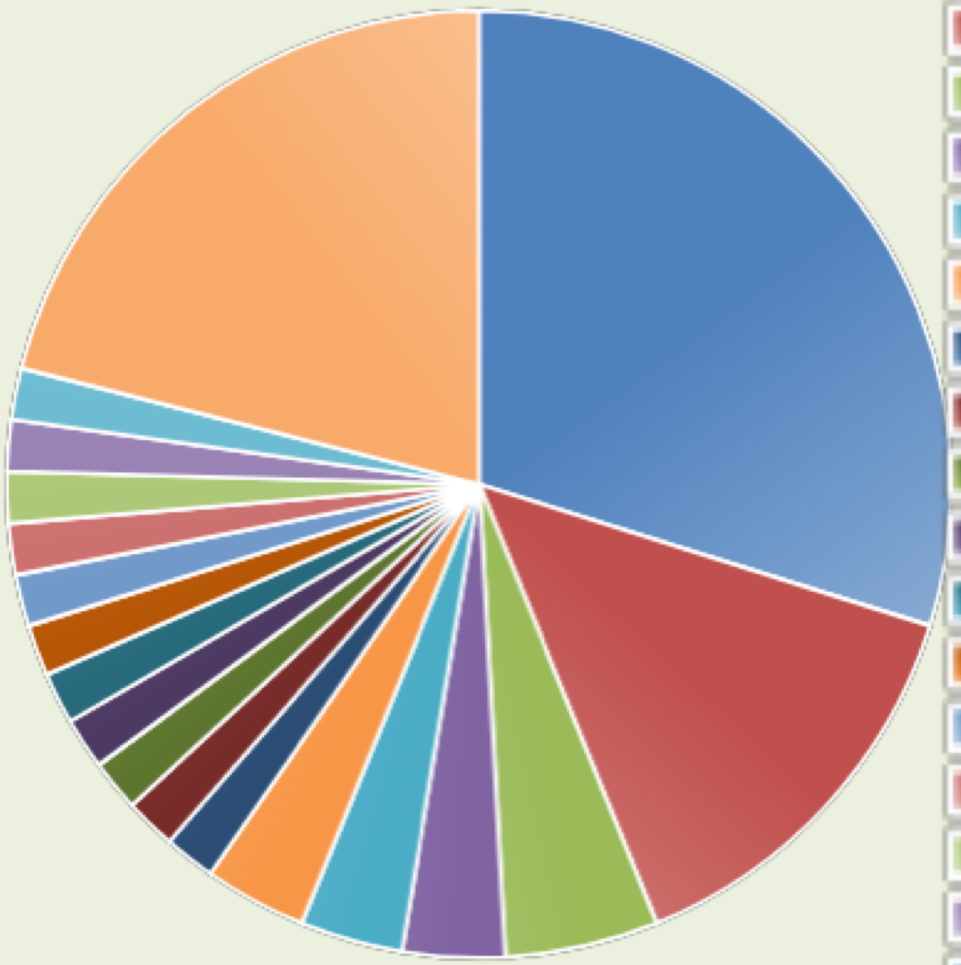
- A diagnosis
- an NGS study with negative results.

Conclusion:

45/57 (79%) with

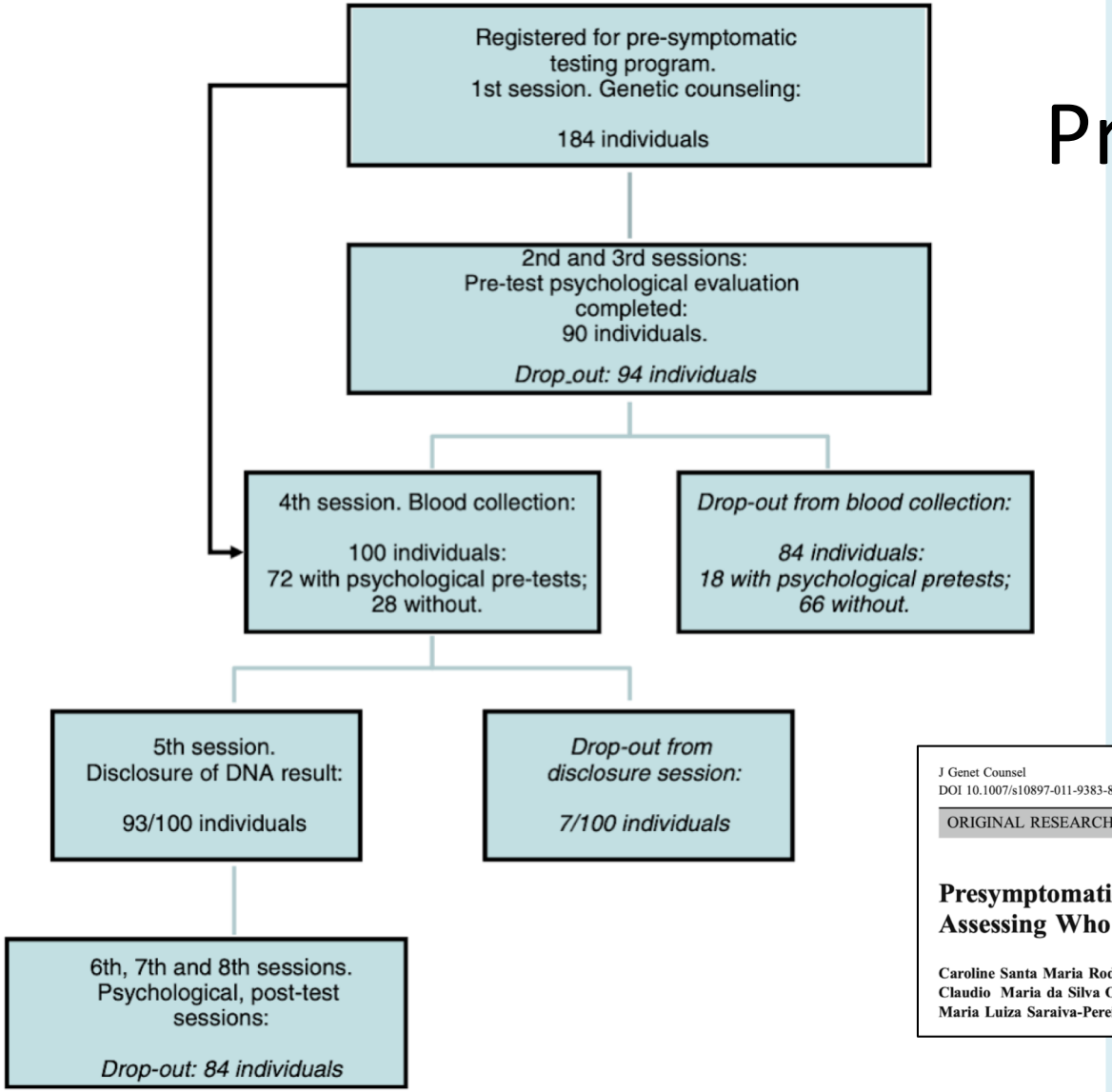
12/57 (21%) without a final diagnosis

Recessive ataxias at SGM-HCPA



- Friedreich ataxia, 17 families
- Ataxia-telangiectasia, 8 families
- Co Q10 deficiency, 3 families
- SCA2, 2 families
- NPC, 2 families
- AOA 2, 2 families
- AOA1, one family
- POLG
- Joubert
- SPG 7
- SPG 11
- SPG 76
- ECHS 1 deficiency
- SCAR 16
- Perrault syndrome
- KCNB 1 encephalopathy
- SCA27b
- No diagnosis after NGS, 12 families

Care: Pre-symptomatic testing



J Genet Counsel
DOI 10.1007/s10897-011-9383-8

ORIGINAL RESEARCH

Presymptomatic Testing for Neurogenetic Diseases in Brazil: Assessing Who Seeks and Who Follows through with Testing

Caroline Santa Maria Rodrigues · Viviane Ziebell de Oliveira · Gabriela Camargo · Claudio Maria da Silva Osório · Raphael Machado de Castilhos · Maria Luiza Saraiva-Pereira · Lavínia Schuler-Faccini · Laura Bannach Jardim

Obrigada!

My students

Ana Laura Brandi

Ana Carolina Martins

Carlos Aschoff

Gabriel Furtado

Ingrid Tuchtenhagen

Karine Caregnatto

Lucas Sena

Rafaella Mergener

Profa. Maria Luiza Saraiva-Pereira

SGM/HCPA staff

Sandra Leistner

Thayne Kowalski

Leonardo Medeiros

Projeto Genomas Raros - PROADI (SUS)

ljardim@hcpa.edu.br