



SERVIÇO DE GENÉTICA MÉDICA
HOSPITAL DE CLÍNICAS DE PORTO ALEGRE



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 Clínicas 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



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

Acta Neurol Scand 2001; 104: 224–231
Printed in U.K. All rights reserved

Copyright © Munksgaard 2001
ACTA NEUROLOGICA
SCANDINAVICA
ISSN 0904-5172

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.

Objective – To examine the clinical, genetic, and molecular characterization of a group of MJD patients recently identified in the southernmost state of Brazil, and compare these data with studies from the literature. **Methods** – Seven 62 individuals from 13 families, mostly of Asorian ancestry, had their clinical data and their MJD expanded regions examined. **Results** – The present patients had an earlier age of onset, on average, than Portuguese–Asorian cases. Their survival, proportion of types, average anticipation, proportion of affected versus non-affected siblings, neurological signs and molecular findings are similar to those observed in patients previously described. Type 1 patients with male transmission showed worse anticipations than type 1 patients with female transmission. Patients with type 1 had also larger CAG expansions than other patients. **Conclusions** – The Brazilian origin seemed to affect the age of onset. We also noted that there were no differences other than the neurological between types 2 or 3, since both are similar in age of onset, disease duration and length of CAG repeats. We addressed the question of maintaining or not subtypes 2 and 3 separated, among patients with genetic and geographical backgrounds like the presented patients here.

L. B. Jardim^{1,2}, M. L. Pereira^{3,4}, I. Silveira⁵, A. Ferro⁶, J. Sequeiros⁷, R. Giugliani⁸
¹Neurologia, Hospital de Clínicas de Porto Alegre, Universidade de Porto Alegre, ²Neurociência, Universidade Federal do Rio Grande do Sul, ³Genética, Instituto de Biologia Molecular e Celular, Universidade de Porto Alegre, ⁴Genética, Universidade de Porto Alegre, ⁵Genética, Universidade de Porto Alegre, ⁶Genética, Universidade de Porto Alegre, ⁷Genética, Universidade de Porto Alegre, ⁸Genética, Universidade de Porto Alegre

Key words: Machado–Joseph disease; polyglutamine expansion; spinocerebellar ataxia

Laura B. Jardim, Medical Genetics Service, Hospital de Clínicas de Porto Alegre, Rua Financas Sules 2006, 91290-900 Porto Alegre, Brazil
Tel: + 55 51 336 8011
Fax: +55 51 336 8010
e-mail: lbgardim@terra.com.br

Accepted for publication April 22, 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

Acta Neurol Scand 2003; 107: 211–214
Printed in U.K. All rights reserved

Copyright © Blackwell Munksgaard 2003
ACTA NEUROLOGICA
SCANDINAVICA
ISSN 0904-5172

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

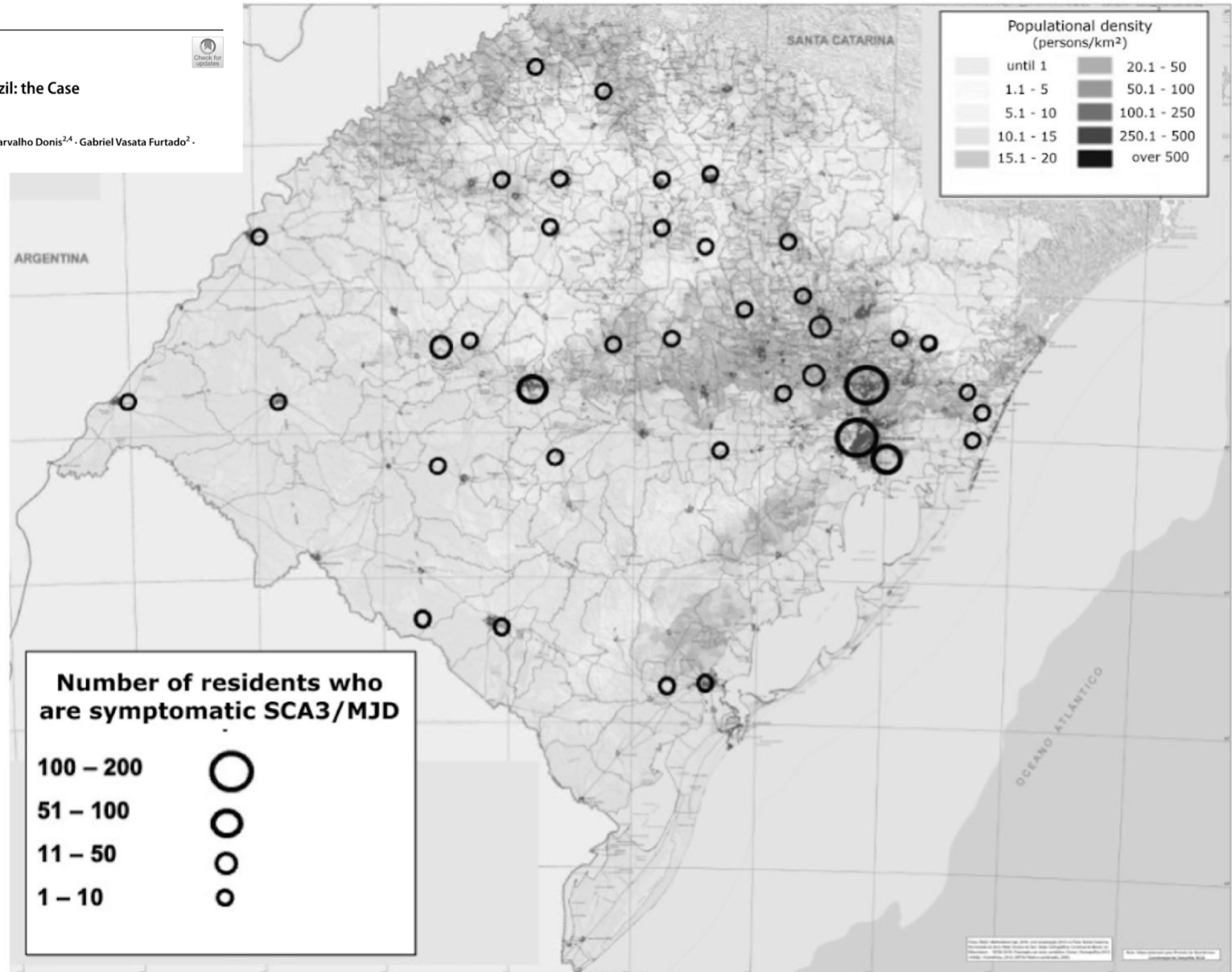
Jardim L, Silveira I, Pereira ML, do Céu Moreira M, Mendonça P, Sequeiros J, Giugliani R. Searching for modulating effects of SCA2, SCA6 and DRPLA CAG tracts on the Machado-Joseph disease (SCA3) phenotype. *Acta Neurol Scand* 2003; 107: 211–214. © Blackwell Munksgaard 2003.

L. Jardim^{1,2}, I. Silveira³, M. L. Pereira^{4,5}, M. do Céu Moreira⁶, P. Mendonça⁷, J. Sequeiros⁸, R. Giugliani⁹
¹Medical Genetics Service, Hospital de Clínicas de Porto Alegre; Departments of ²Internal Medicine,



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 Furtado² · 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



Thorsten Schmidt, Tübingen



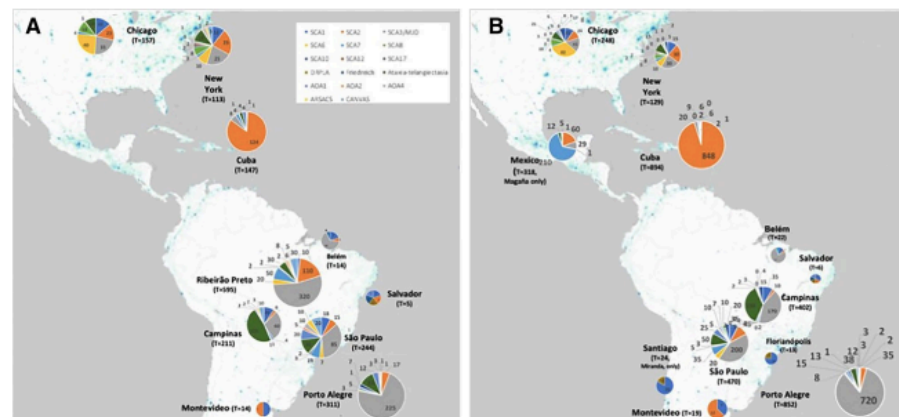
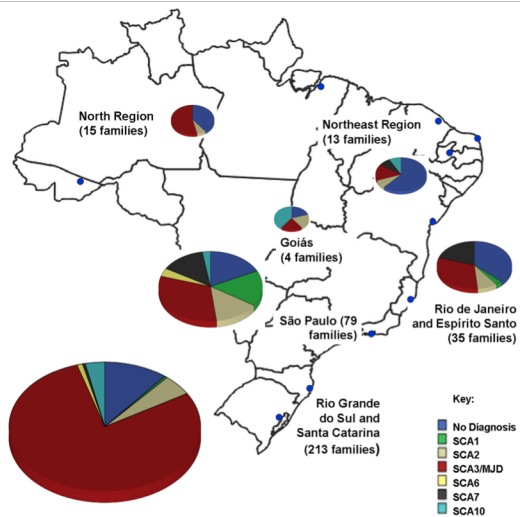
National Institutes
of Health



Tetsuo Ashizawa, Houston



And some networking to increase access



ATAXIA GLOBAL INITIATIVE
worldwide platform for clinical research in ataxias

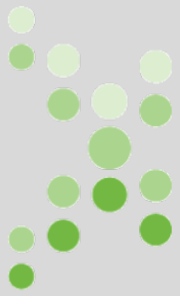




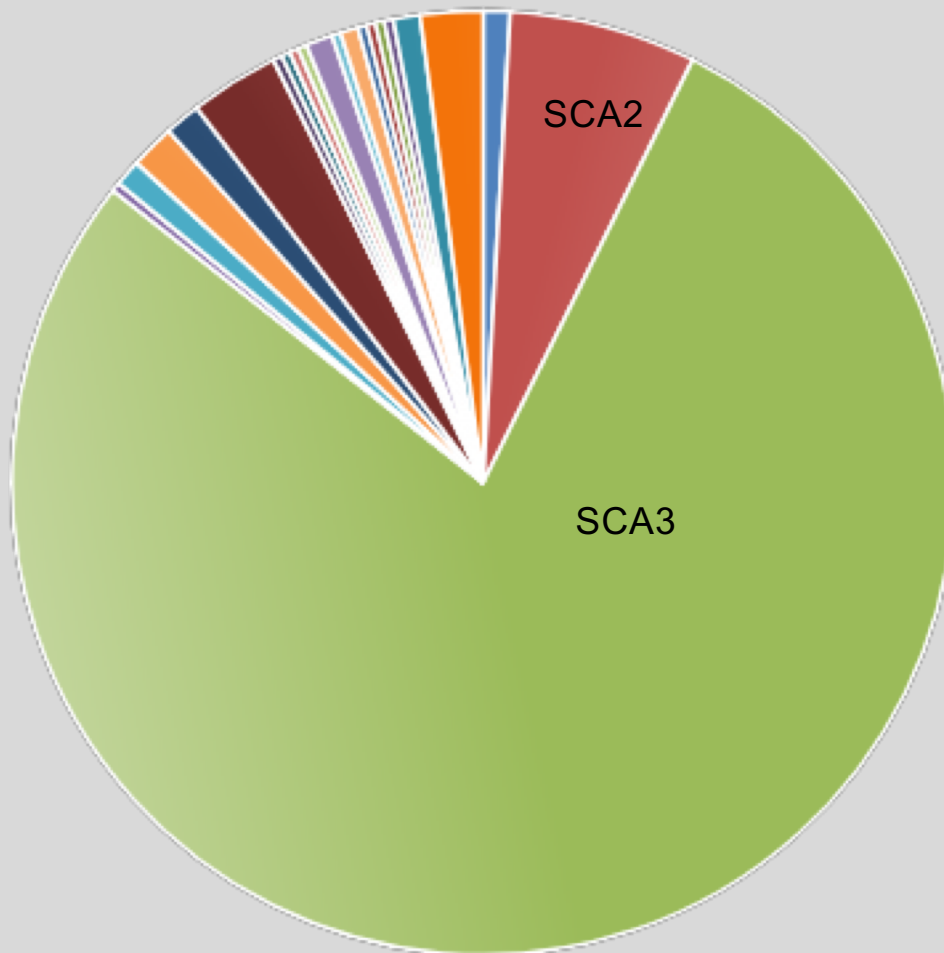
SCAs diagnosed at SGM-HCPA between 2000 and 2026

T	NOME DO PACIENTE	Prontuário	STATUS	DNA = Inf. Livre	DIAGN FAMILIAR - uma anotação por família				
1366	296		T		SCA3				
1367	296		Sintomático	SCA 3					
1368	296		Sintomático	SCA3					
1369	297		sintomático	SCA3	SCA3				
1370	297								
1371	298		sintomático	SCA10	SCA10				
1372	299		sintomático	sca3	SCA3				
1373	300		sintomático	sca3	SCA3				
1374	300		sintomático	SCA3					
1375	300		Sintomático	SCA3	SCA3				
1376	301		Sintomático	SCA3	SCA3				
1377	302		sintomático	SCA3	SCA3				
1378	302		sintomático	SCA3	SCA3				
1379	303		sintomático	SCA 2					
1380	304		sintomático	SCA 3	SCA 3				
1381	304		sintomático						
1382	305			SCA2					
1383	306			SCA2					
1384	307		sintomático	SCA3	SCA3				
1385	307		sintomático	SCA3					
1386	308			SCA3					
1387	309		sintomático	SCA3	SCA3				
1388	310		sintomático	SCA3	SCA3				
1389	310		preditivo						
1390	310		preditivo						
1391	311		sintomático	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		sintomático		SCA3				
1399	318		sintomático		SCA NÃO DIAGNÓC 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		sintomático		SCA3				
1412	326		sintomático						
1413	327		Doente		SCA3	Participou do ECR lite			
1414	328		Doente		SCA13	SeqExoma: NCNC3 - c.12880>A.p (Arg423His), heterozigose			
1415	329		Doente		SCA3				
1416	329		preditivo						
1417	329								
1418									
1419									

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



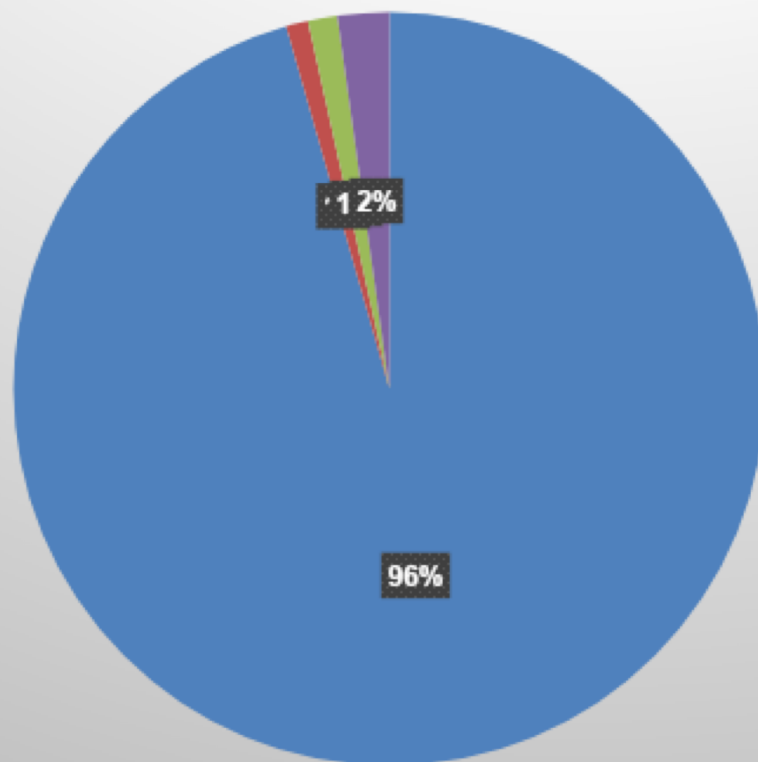
SCAs diagnosed at SGM-HCPA between 2000 and 2026



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 electrophoresis, 302 families.

Red: Targeted panels for SCAs, 3 families

Green: Exomes, 4 families

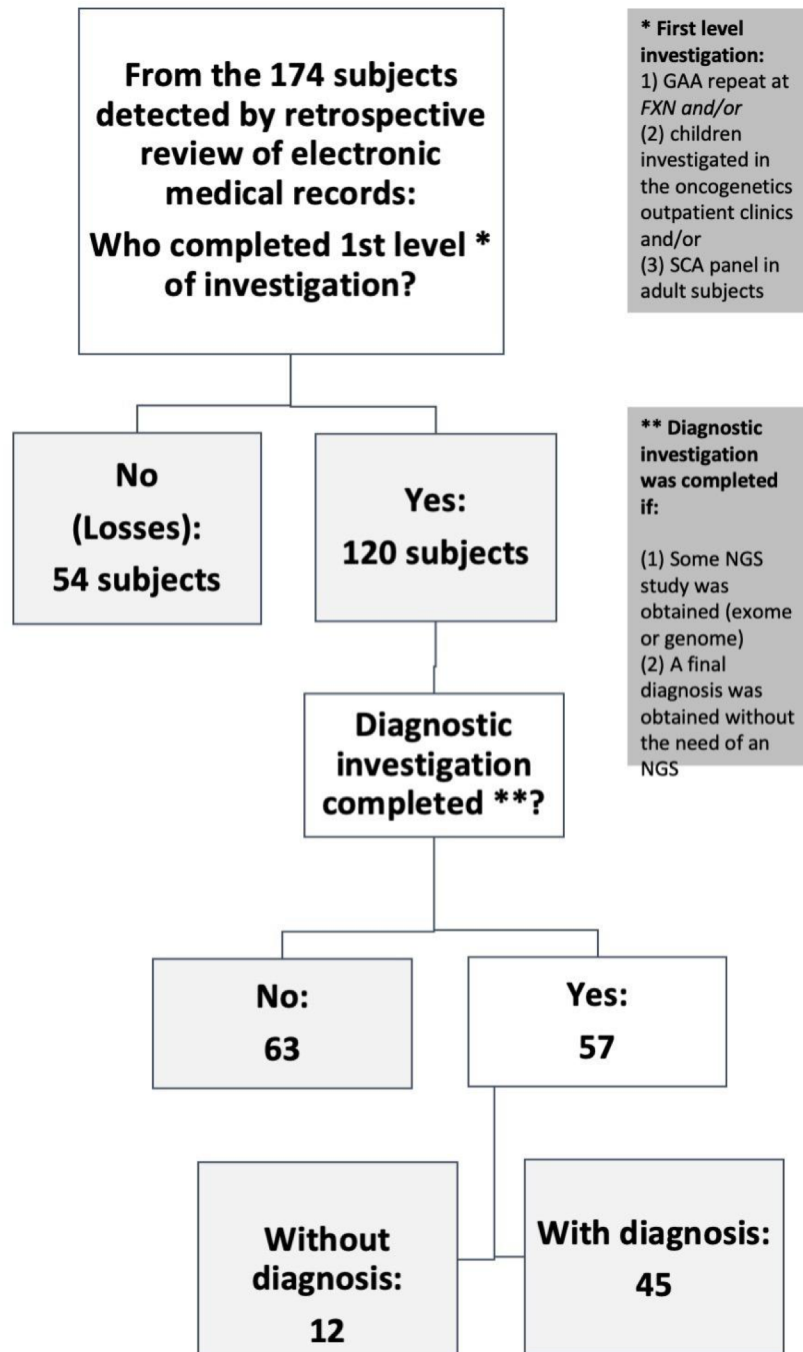
Purple: Genomes, 7 families



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:

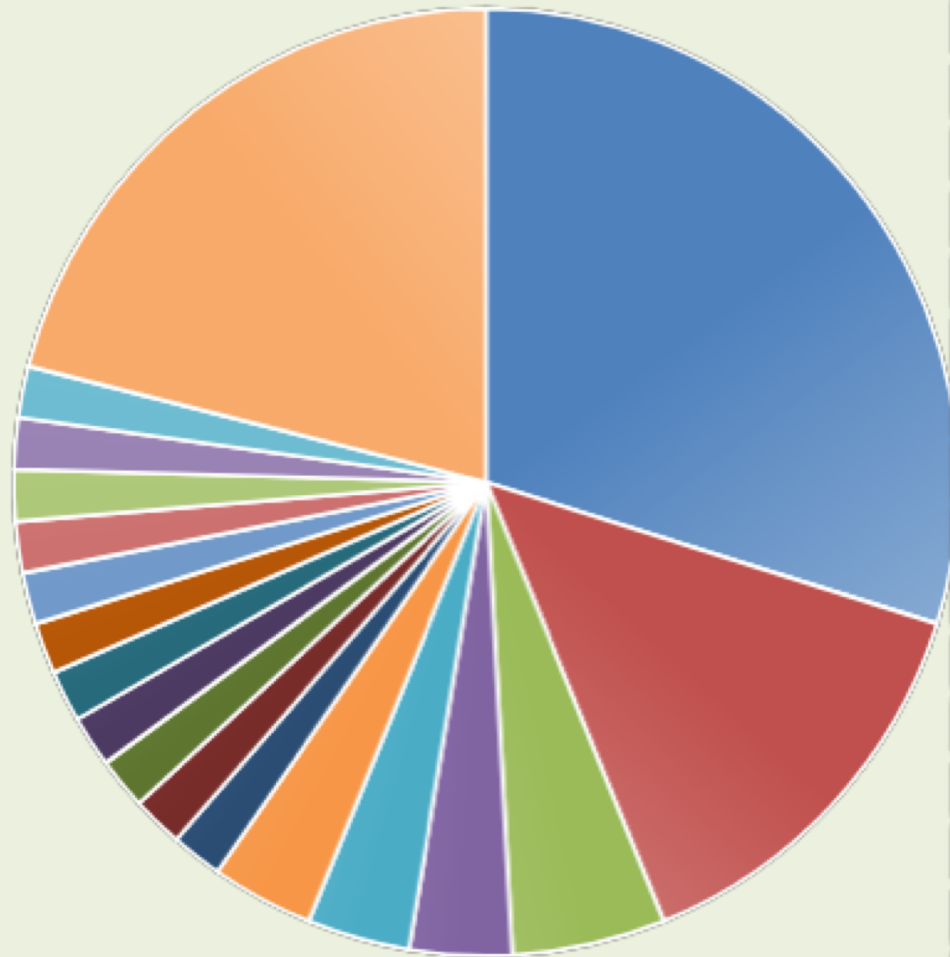
- A diagnosis
- an NGS study with negative results.

Conclusion:

45/57 (79%) with

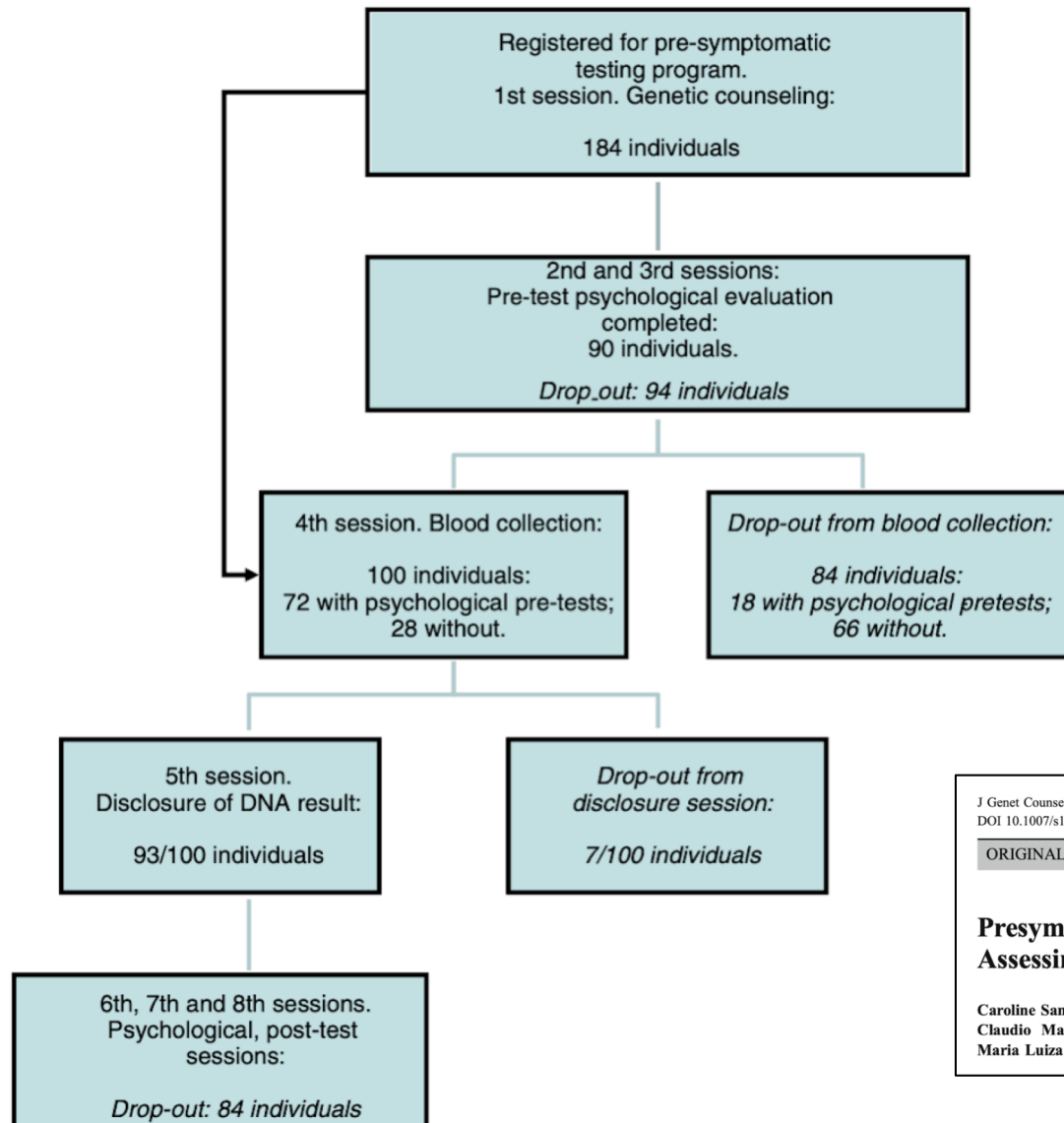
12/57 (21%) without a final diagnosis

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 Luíza Saraiva-Pereira · Lavinia 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