



AGI 1st Regional Conference: Latin America

**Maria Carolina da
Cunha
Brazil**

May 7th, 2026

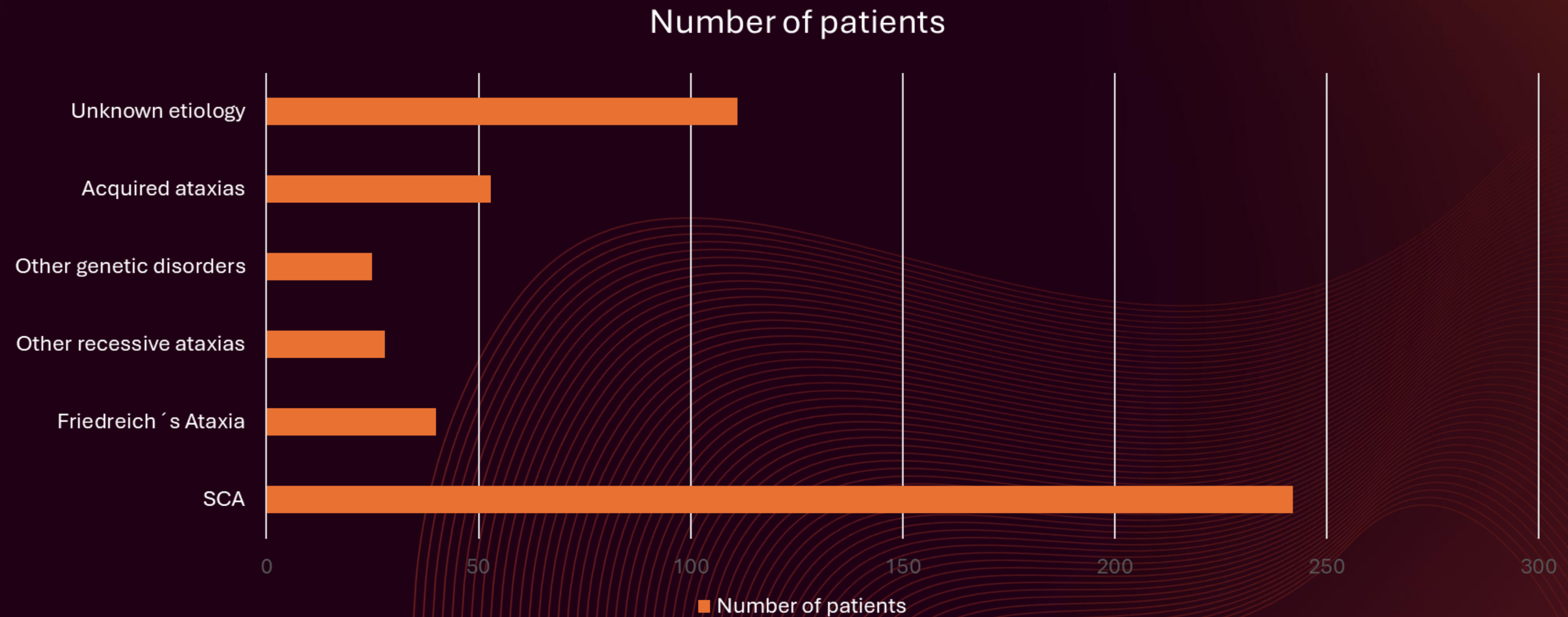
Sarah Network of Rehabilitation Hospitals



Ataxia outpatient clinic Multidisciplinary team



Types of ataxia

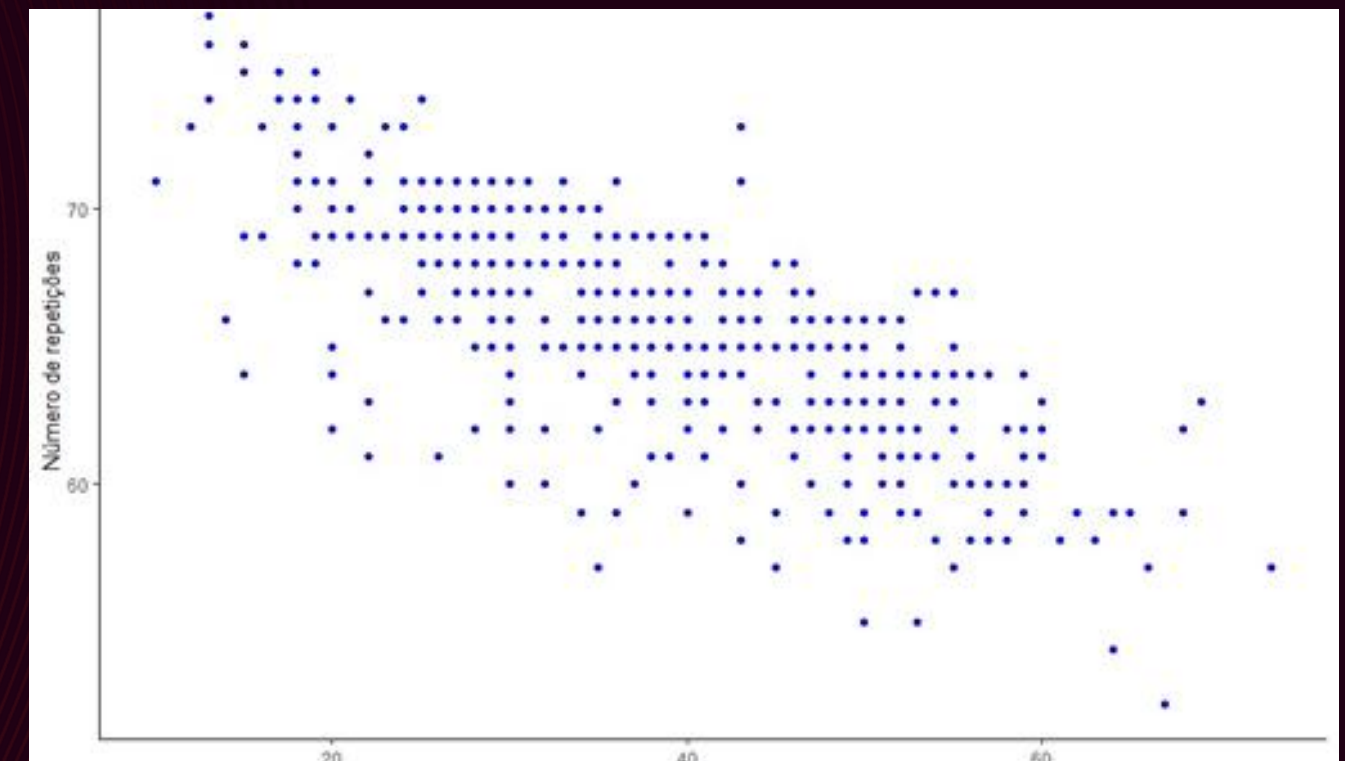
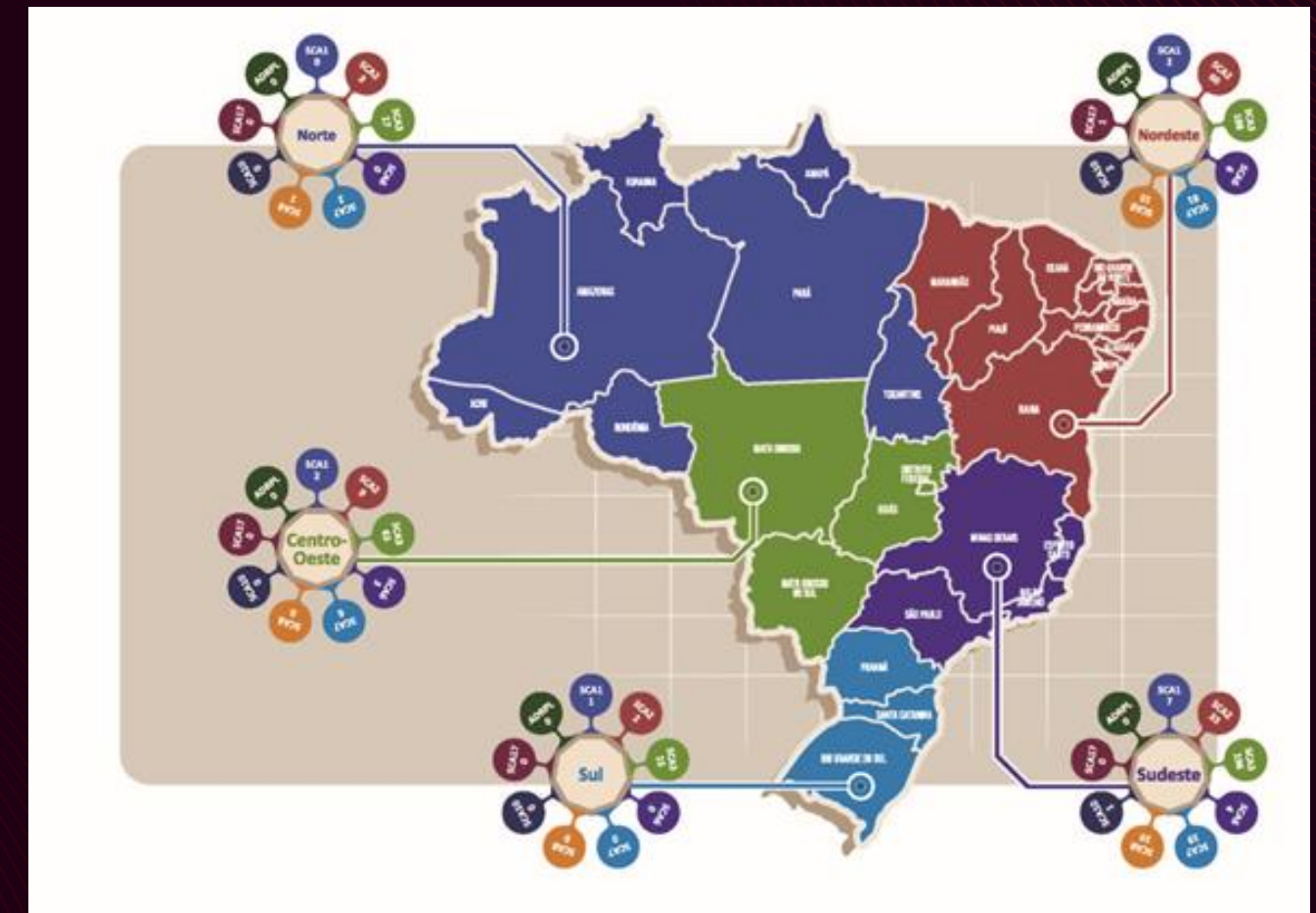




Spinocerebellar Ataxia in Brazil: A Comprehensive Genotype – Phenotype Analysis

Maria Carolina Da Cunha Ganimi¹ · Christian Marques Couto¹ · Alessandra de La Rocque Ferreira¹ · Carmen Lucia Antão Paiva²

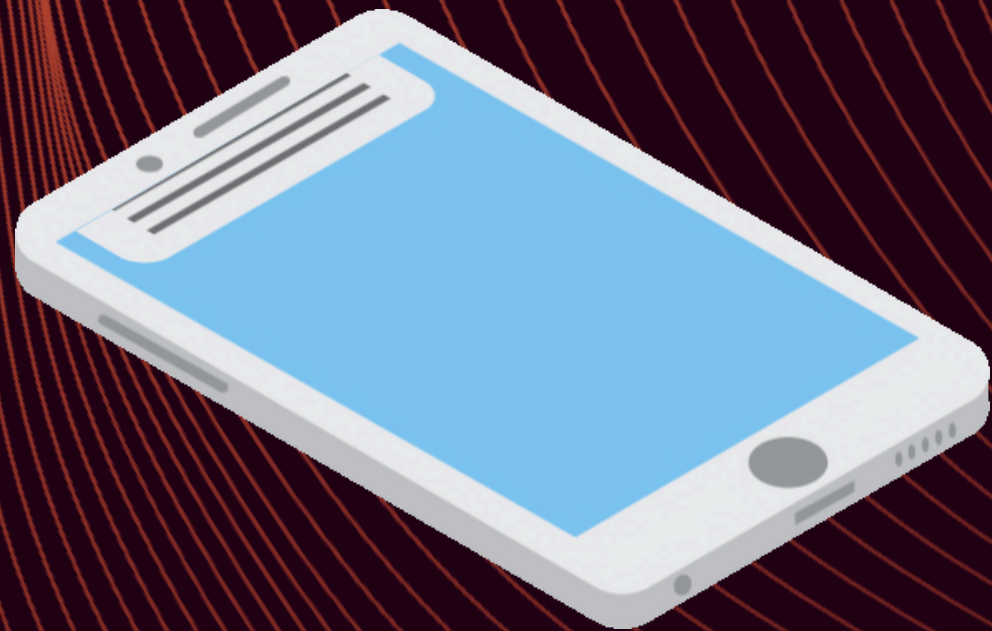
Multi center study in Brazil
Frequency of SCAs
Clinical and epidemiological aspects
Correlation with the size of the expansion



Effects of an intensive rehabilitation program in patients with spinocerebellar ataxias



Research Lines



01

Prevalence of
Friedreich's ataxia in
Brazil

02

Association between
SCA2 intermediate
expansions and
Amyotrophic lateral
sclerosis

03

Evaluation of
impulsivity
symptoms in SCAs

04

Assessment of
osteosarcopenia in
patients with
hereditary ataxias

Challenges in diagnosis



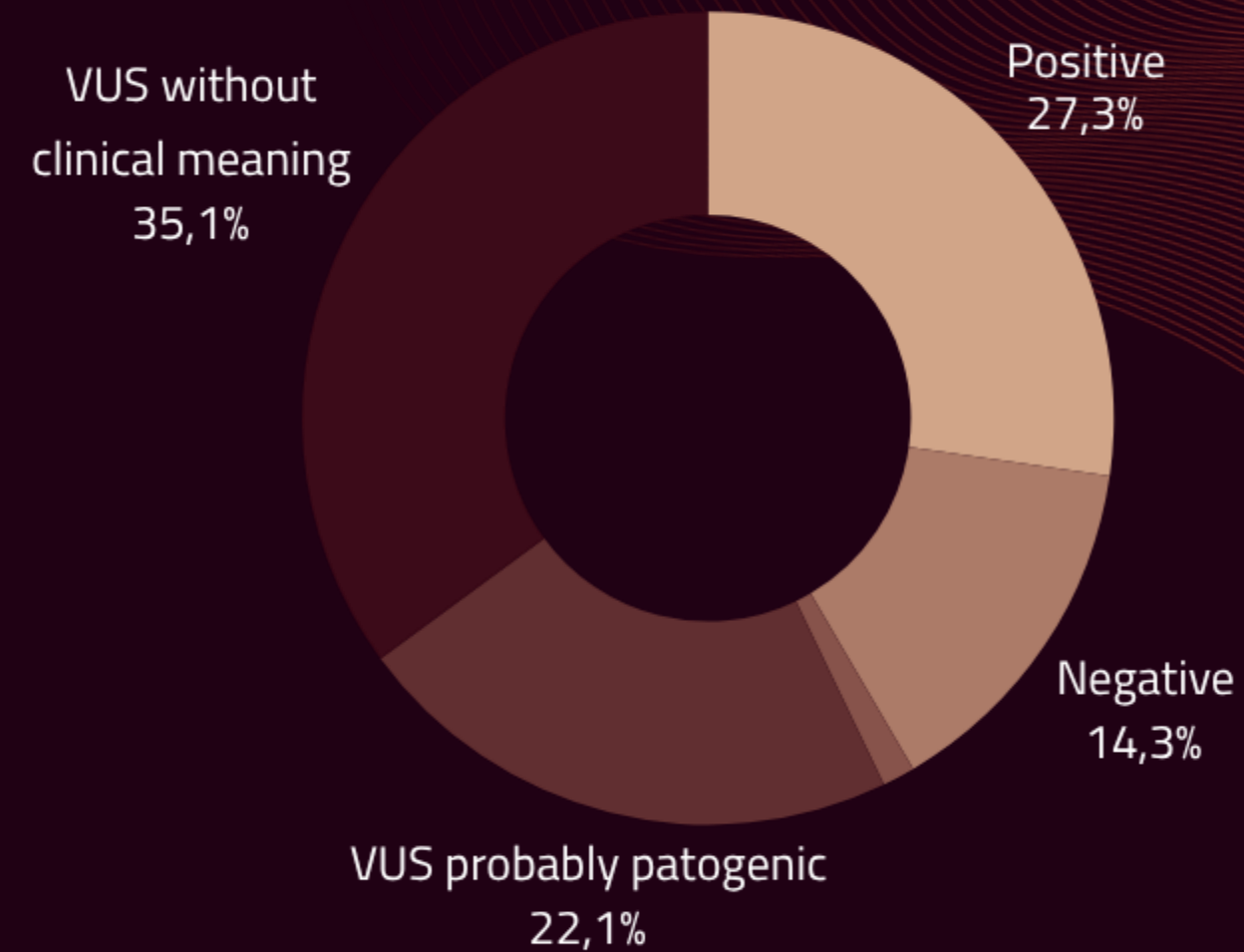
01 Limited access to NGS panels

02 High cost of whole-exome sequencing

03 Variants of uncertain significance or findings that do not fully explain the clinical phenotype

NGS Panels

72 NGS panels
May 2023 and December 2025



Challenges

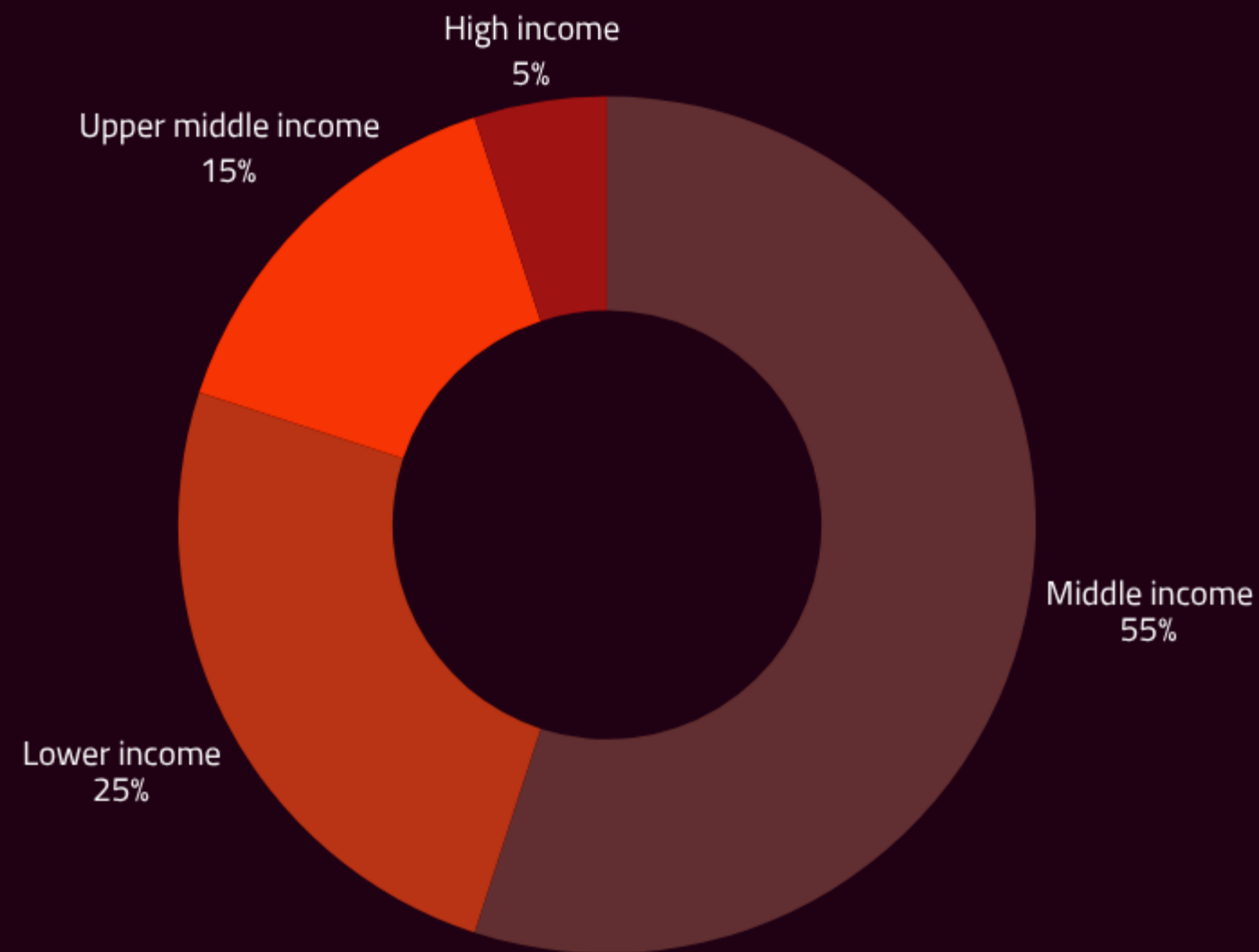
Social barriers

Generally low educational and economic levels

Difficulty in commuting to Sarah Rio

Disease severity

Low attendance and adherence to treatment programs



Thank you

