

Genetic Modifiers of Spinocerebellar Ataxias (GeM-SCAs)

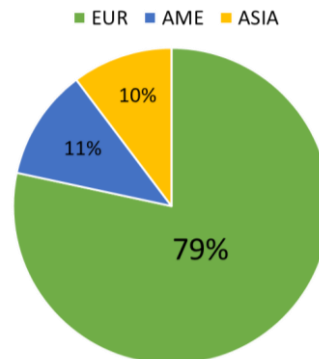
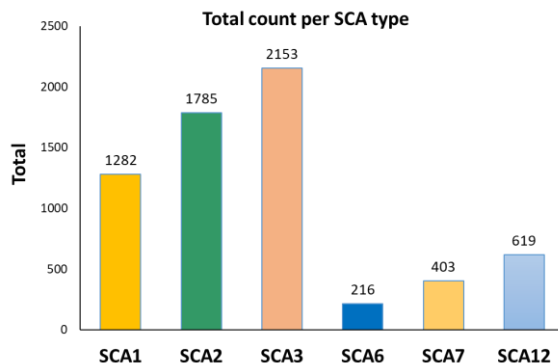
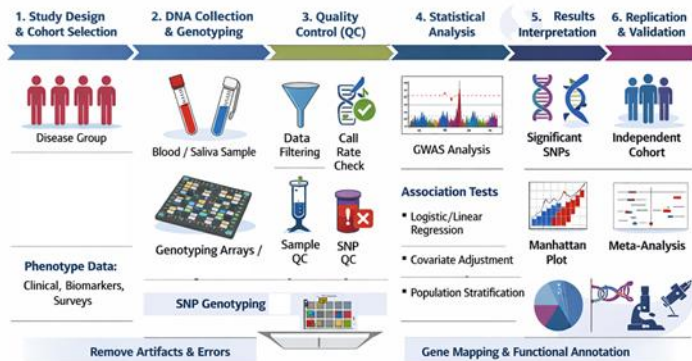
Research Question: What are the genetic modifiers of repeat expansion spinocerebellar ataxias (SCAs) and are they the same or different from other expansion disorders?

Main Objective: To identify genetic variation that modifies clinical phenotype in SCAs.

Aim 1. Identifying novel genetic *loci* that modify age at onset (AAO), and other clinical phenotypes, such as progression, in SCAs.

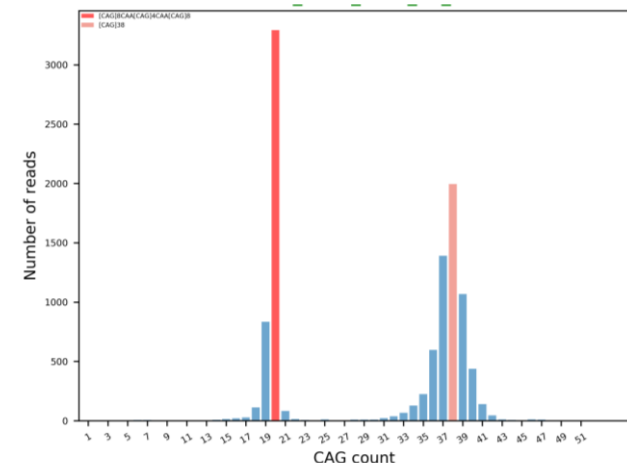
Aim 2. Determining the mechanism whereby genetic *loci* modify AAO and other clinical phenotypes.

Stages of a GWA Study



- Ultra-deep amplicon targeted DNA sequencing

- ✓ Highly sensitive NGS technology
- ✓ Exceptionally high coverage



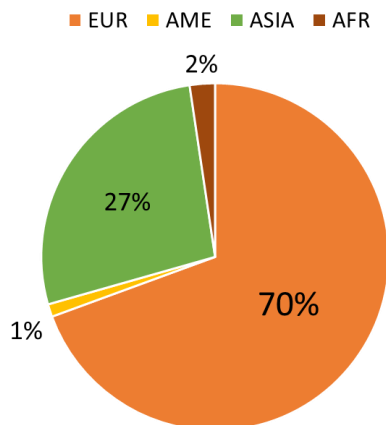
DRPLA Natural History and Biomarkers Study (DRPLA-NHBS)

Research Question: What are the milestones in DRPLA progression and the more informative disease biomarkers?

Main Objective: To characterize the natural history of DRPLA in both juvenile- and adult-onset patients and study different modalities of biomarkers in this condition.

Clinical, Imaging and Fluid Biomarkers

3T SIEMENS PRISMA MRI Scanner Quanterix Simoa HD-X Analyzer



Genetic Biomarkers

ABI 3730xl



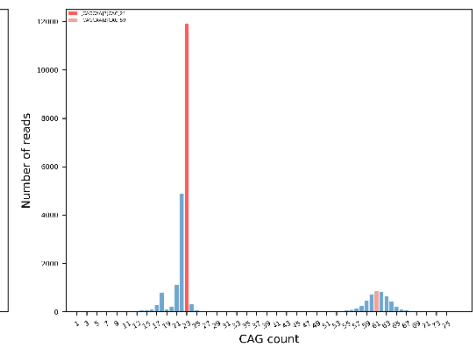
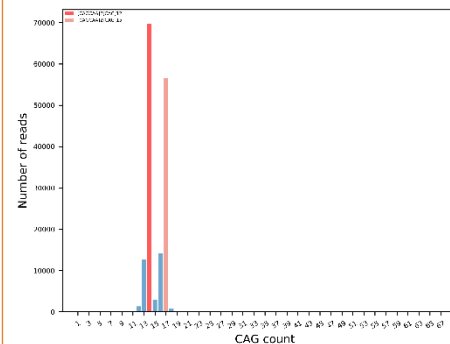
QuantStudio 7 Flex



Illumina MiSeq i100



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DRPLA Natural History and Biomarkers Study (DRPLA-NHBS)

Take-home messages

We need to **increase representation** of patients from Latin America

- ✓ different genetic backgrounds and environmental exposures than those of Europeans, Asians, and Africans.
- ✓ to provide access to Latin American patients to these platforms in support of the design and conduct of clinical trials.

Thank you all!