Quantifying and modeling the impact of rare SNVs variants on cognitive and behavioral traits in neurodevelopmental disorders

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Upcoming Master 2 internship

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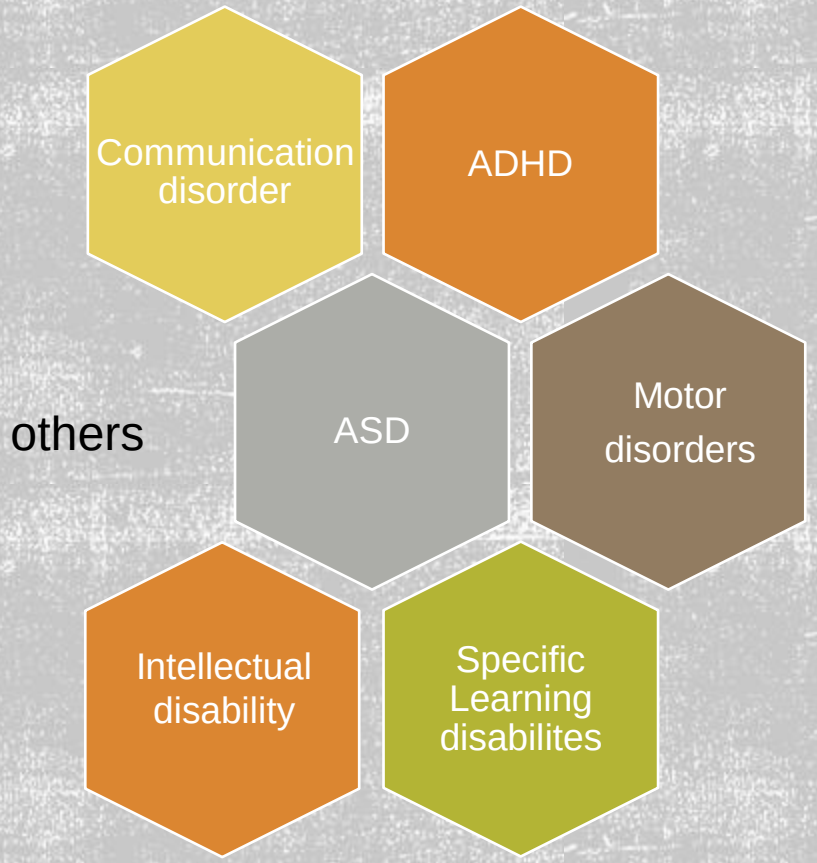
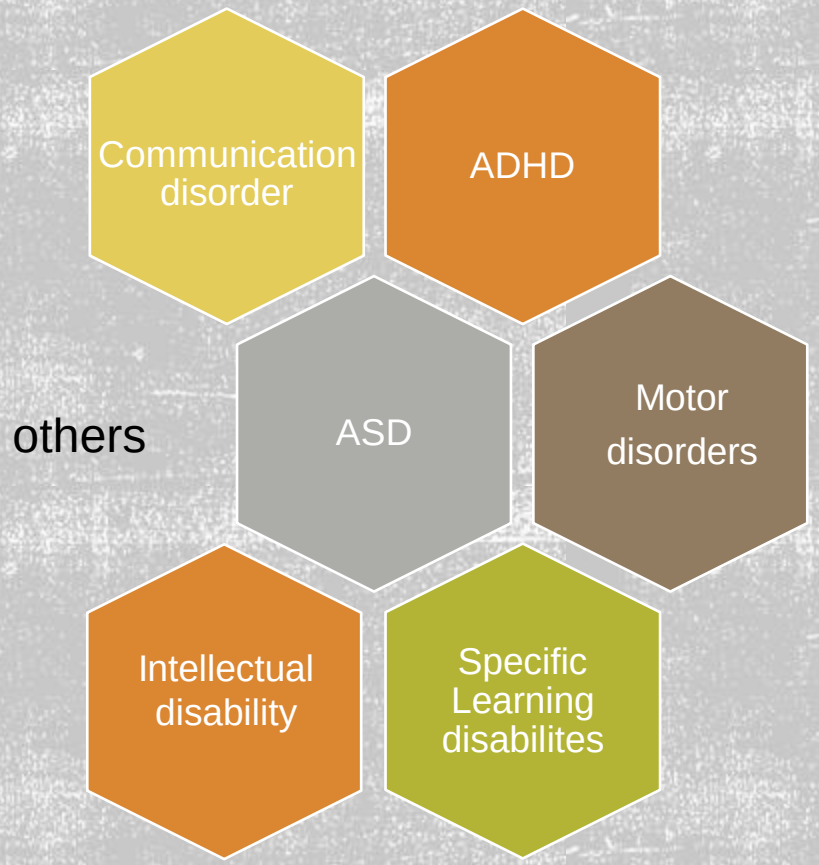
Neurodevelopmental disorders



DSM-V classification

Neurodevelopmental disorders



DSM-V classification

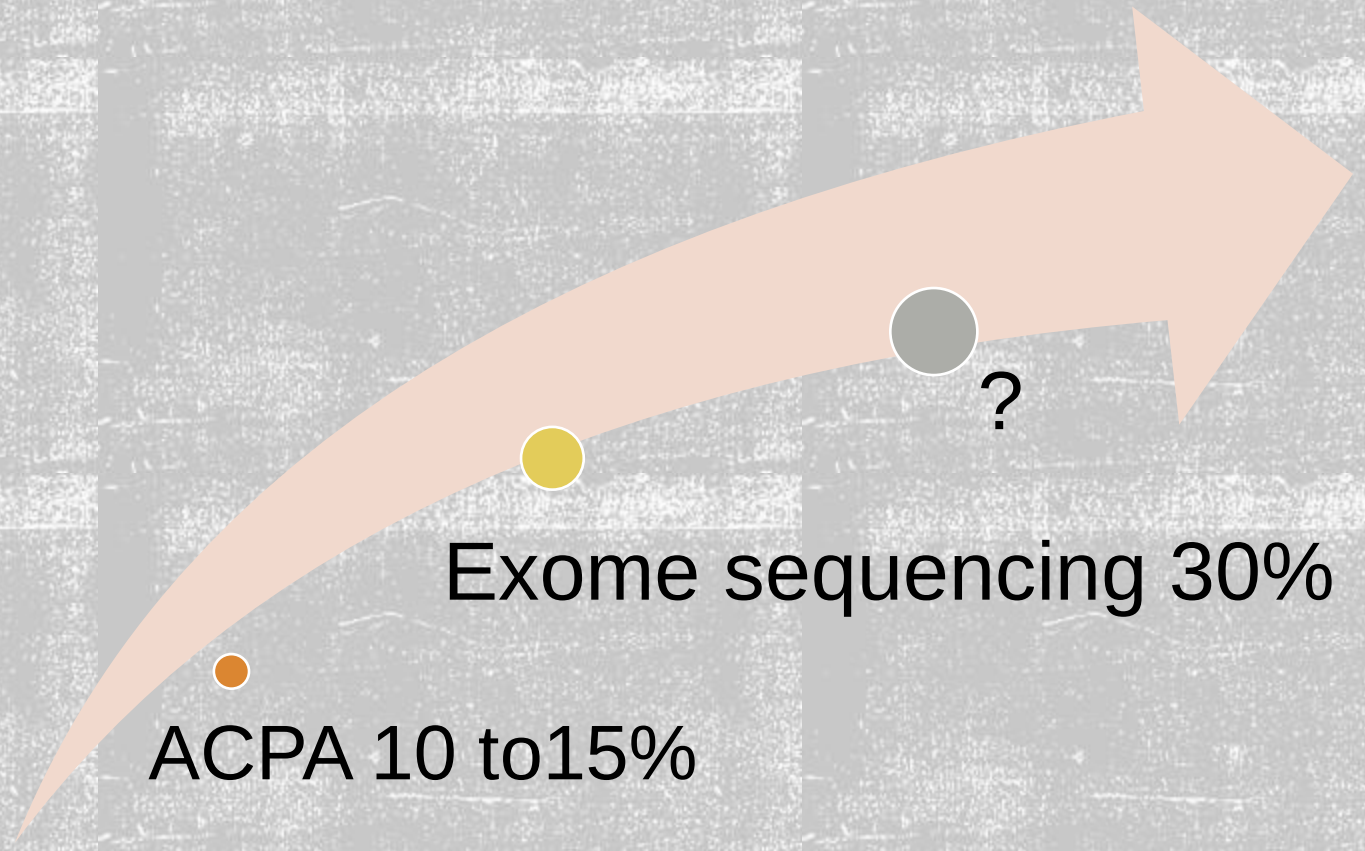
Heterogeneity

- Clinical
- Genetic
- Biological

Major health problem

- Genetic cause in 50 to 80%
- ASD = 1% world population

Genetic diagnosis in NDDs



How to improve genetic diagnosis?

Mapping the effects of rare variants genome-wide onto cognitive and behavioral dimensions

Shown that genomic dosage due to CNVs could modulate brain structure, cognition and behavior

Develop new strategies to identify genetic scores and functional annotations that explain the effect of rare variants on cognitive and behavioral traits

Purpose of the internship

Population

- General population
- Autism cohorts
- Local cohort of >200 families of patients

Clinical data

- Quantitative measures of global cognition (IQ test)
- Verbal (non-word repetition task)
- Behavioral (social communication test)

Tasks

- Create an algorithm to interpret non-recurrent SNVs in NDDs
- Validate this algorithm on a local cohort
- Evaluate the data via statistical linear models between estimations from autism & general population and estimations from the local cohort

Thank you for your attention