UNIQUE patients and families:

Cognition, behaviour and psychiatric comorbidity in intellectual disabled syndromes

Centre for Rare Diseases
Karolinska University Hospital
Karolinska Institutet

UNIQUE – the people behind the project

The project will be carried out at Centre for rare diseases (CSD) and be lead by:

- Ann Nordgren – Professor, senior consultant at clinical genetics
- Charlotte Willfors – Project coordinator, PhD, psychologist

In collaboration with:

- Britt-Marie Anderlid – PhD, Child neurologist/Geneticists
- MaiBritt Giacobini – PhD, Psychiatrist
- Louise Frisén – PhD, Psychiatrist

- Ågrenska foundation in Gothenburg

UNIQUE – the aim of the project

To increase the knowledge of cognition, behaviour, and psychiatric comorbidity in rare diseases

Spread the knowledge families, clinicians and decision makers via e.g. clinical guidlines

Improve the care and treatment options in rare diseases

The literature on cognition, behaviour and comorbidity in Williams syndrome

- Mild to moderat intellectual disability with a mean total IQ of 55 (40-100)
- Verbal abilities usually a strength (concrete language) and visuo-spatial ablities a weakness
- Mixed results on delayed and atypical language development, typically impairments in social aspects of the language
- Mixed results regarding how IQ affects other behavioral problems
- Great musical interest and talent
- High comorbidity with ADHD (65%) and specific phobia (54%)

Martens, Wilson & Reutens, 2008, Mervis & John, 2010, Rossi & Giacheti, 2017

UNIQUE – what we want to study

- We want to study what is specific for the different syndromes regarding:
  - cognitive profiles (verbal ability, perceptual ability, working memory, executive functions)
  - behaviour (language, motoric skills, social abilities, adaptive functioning)
  - neuropsychiatric comorbidity (ASD, ADHD, specific learning disabilities)
  - other psychiatric comorbidity (anxiety, depression, sleeping disorders, OCD)

- Williams syndrome will be one of the first syndromes that will study, thereafter we plan to study:
  - Fragile X syndrome
  - 22q11 deletions syndrome
  - Noonan syndrome
  - Neurofibrometosis type 1
  - KBG syndrome

- Longitudinel 3 years follow-up

Who can participate in the study?

Inclusion criteria:

- A molecular confirmed diagnosis of Williams syndrome, Fragile X syndrome, 22q11 deletions syndrome, Noonan syndrome, Neurofibrometosis type 1, or KGB syndrome
- Age of 6 years or older (no upper limit)
- Ability to travel to Karolinska hospital in Solna for assessments (or possibly other university hospitals such as Lund, Linköping, Gothenburg, Örebro, Umeå)

How is the assessment done?

Before the visit:

- Information is sent to the family
- A researcher will call the family to answer any questions and plan a date for the visit
- Consent and questionnaires are sent to the family

During the visit:

- One day of psychological testing and interviews (in total 7h including breaks)
- Adaptations are done to each individual such as breaks, company by parents during testing

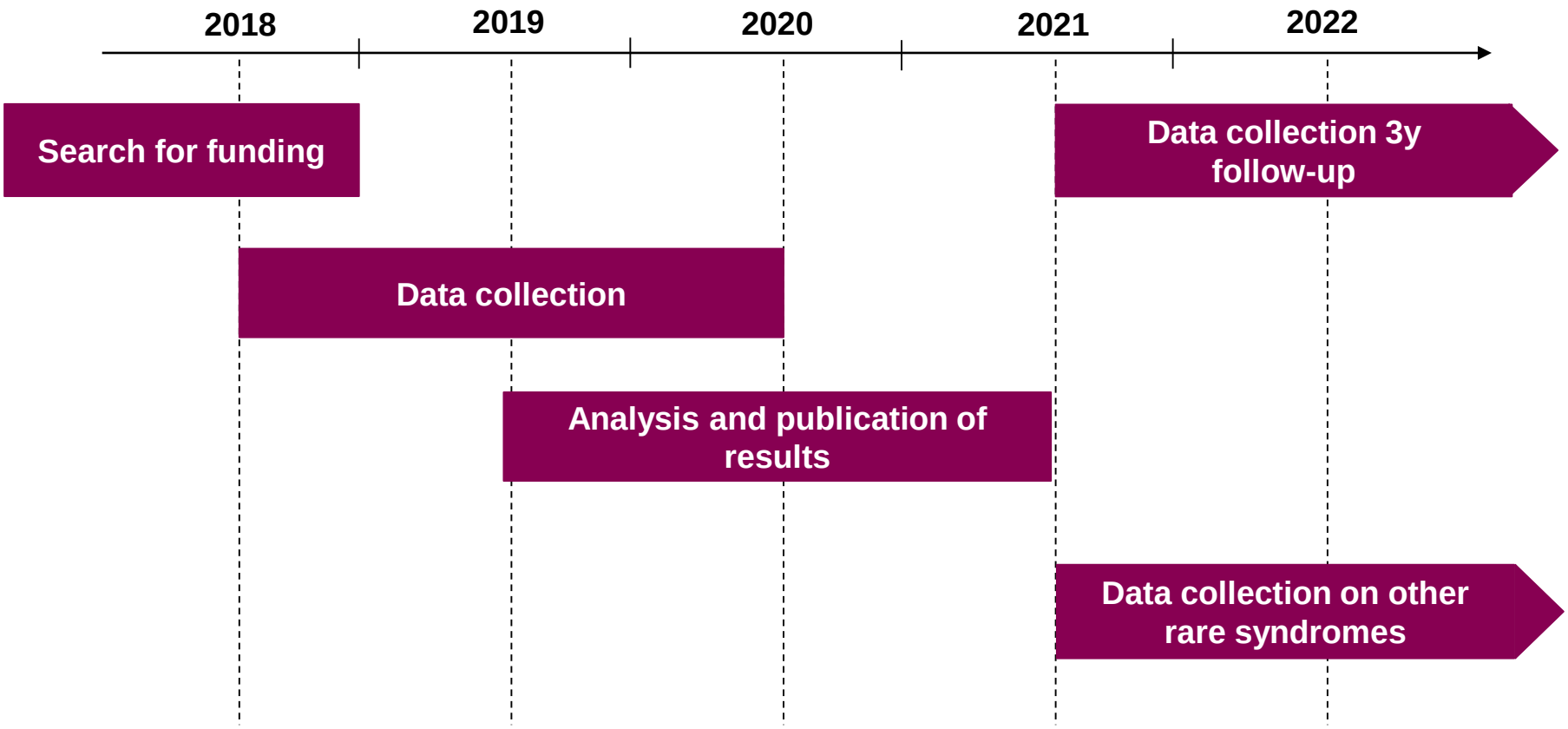
After the visit:

- Feed-back on the results of the assessment if the family request
- Remitation for furhter assessment and/or treatment if adequate

Risk/benefits with participating

- There are no risks in participating. The participation is completely volontary and can be interupted at any point.
- There are no concrete benefits of participating.
- The participants can get a in-depth description of there functioning.
- If any not previous diagnoses or treated conditions are discovered the participant will be refered for further clincial assment and treatment.

Time plan


2018
2019
2020
2021
2022
Search for funding
Data collection 3y follow-up
Data collection
Analysis and publication of results
Data collection on other rare syndromes

Intressted in the project?

http://www.karolinska.se/sallsyntadiagnoser

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