

# International chromosome 6 research

A study in collaboration with ECARUCA,  
driven by parents, for parents



## How this project started

The mother of a child with a chromosome 6q deletion, who attends our follow-up clinic for children with rare chromosome disorders, told us about the active international chromosome 6 patient group on Facebook. On request of and in cooperation with this Facebook group we started a study on chromosome 6 deletions and duplications.

## What is our goal?

Our goal is to produce a detailed interactive phenotype-genotype map of chromosome 6. This web-based map can be used to give tailored guidance to patients and their families about the prognosis for their child, ranging from their likely developmental trajectory to additional health problems they may encounter. The ultimate goal is to offer patient-specific guidance and treatment.

## What next?

Together with the help of ECARUCA powered by Cartagena BENCH and the Facebook group we want to involve as many patients as possible in our study. A detailed phenotype-genotype analysis will be performed on the data collected by us and on data already available in the literature and databases. This will enable us to create and further improve the phenotype map of chromosome 6.

## How can you be of help?

Inform your patients with a chromosome 6 aberration or their parents about the 'chromosome 6 Facebook' group and/or tell them about the 'chromosome 6 study'.

Patients can contact us about participating in the study.

Barbara Frentz  
Assistant researcher  
University Medical Centre Groningen

Prof. Conny van Ravenswaaij-Arts  
Clinical geneticist and principal investigator

Email: [chromosome6@umcg.nl](mailto:chromosome6@umcg.nl)

Webpage: [http://www.rug.nl/research/genetics/research/chromosome\\_6/](http://www.rug.nl/research/genetics/research/chromosome_6/)

Twitter: <https://twitter.com/c6study>