Phenotype-genotype analysis in chromosome 6 aberrations: a parent-driven study

Re-opening of the study!

We are happy to let you know that we have decided to restart the collection of information. Until now we were focusing on 6q deletions, but from now on 6p and 6q duplications are welcome as well.

I will contact the parents who have already showed interest in the study by email; if you did not express your interest in the study yet, but want to participate please send an email to chromosome6@umcg.nl

What is phenotype-genotype analysis?

The *genotype* is the genetic information of an individual. The *phenotype* is the effect this person's genetic make-up has on what they look like and which clinical features they have.

In this chromosome 6 study we want to compare the exact abnormalities found (the genotype) with their clinical effect (the phenotype). We will try to link different features to specific parts of chromosome 6. In this way, we may be able to predict the clinical effect of certain deletions and duplications. We also hope to gain more insight into the function of the various genes that are located on chromosome 6.

Such a study can only be successful if we have a sufficient number of participants and if their abnormalities have been characterized as precisely as possible, for instance by array analysis.

Why do we want to do this study?

Chromosome 6 deletions and duplications are rare and there have been hardly any systematic studies performed. Little is known about the problems that can develop when there is an aberration in the genetic material on chromosome 6. This means that we are unable to give good answers to many of the questions that parents have. There is clearly a need for the answers this study will provide, because it has been requested by the parents of affected children. We are excited to have the opportunity to perform such a study with the cooperation of the Chromosome 6 Facebook group.

In the future we hope to be able to better inform parents about their child's prospects.

Why is it called “a parent-driven study”?

This study was originally requested by a parent who informed us about the Chromosome 6 Facebook group. The parent launched the idea for the study in the Facebook group and it was enthusiastically welcomed. The parents are therefore the major stakeholders for this study and we, the researchers, are open to their ideas. This study can only succeed through collaboration with the parents.

We have already proved that it works, within several weeks we managed to collect 65 ‘complete’ 6q files and eight for 6p!

Who can participate in this study?

Anyone with a deletion or duplication of chromosome 6 can participate in this study. However, the chromosome aberration has to be confirmed with a detailed chromosome analysis (array CGH, aCGH, micro-array, CMA or SNP array analysis).

Who can NOT participate in this study?

- If no detailed chromosome analysis has been performed, we do not have reliable genotype information of your child. So without array analysis we can not include your child in the study unless an array is going to be performed.
- Some children have a more complex chromosome aberration, for instance the combination of a duplication of 6q with a deletion on another chromosome. Such complex aberrations may hamper the analysis.

Please contact us if you are not sure whether the aberration in you or your child can be used in our study.

What do you need to do for this study?

- To participate in this study, you need to fill in a questionnaire.
- We also ask you to sign consent forms so we can collect additional information from your child’s medical doctors.
- We would very much like to see a face and/or full photograph of your child, but this is not obligatory.

No visits, examinations or blood samples will be required.

All the information you provide will, of course, be treated confidentially and will be used and stored anonymously. The results of the study will be reported anonymously.

What information are we going to collect?

- The questionnaire will ask about the pregnancy, birth, development, behavior and health of both you and your child.
- We will ask the child's doctors to send us clinical information, including the results of any imaging studies (MRI of the brain, ultrasound of the heart, etc.).
- We will also need to have the original result of the detailed chromosome study.

What are we going to do with your information?

With your information, we will try to pinpoint which parts of chromosome 6 are responsible for specific birth defects, developmental delays and learning difficulties.

- We want to combine the collected information with data available in existing databases
- We want to construct a high-resolution genotype-phenotype map, a map indicating in as much detail as possible what the effect of different chromosome 6 alterations is.
- We want to translate this information back to families using an interactive website.
- We will publish our results in an international medical journal so that all doctors and parents can benefit from our findings.

Want to participate?

If you would like to participate in our study or if you have any questions, don't hesitate to contact us at chromosome6@umcg.nl

Keeping you updated on this study

We will ask our contact parent to post information about the data collection and other interesting news on Facebook.
In order to respect your privacy we will not participate in your Facebook group.

Yours sincerely,

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