

UMCG,
Groningen
21 May, 2014

Dear 'Chromosome 6' parents,

This letter is to update you on the progress of the Chromosome 6 study, both what we are working on at the moment and what we intend to do in the future. We want to inform you about the current status of our Chromosome 6 Research Project and answer some of the questions that have been forwarded to us by Pauline Bouman.

Progress

Our work on the chromosome 6 data is progressing steadily. We are currently working on the first paper, which is very exciting because we have been able to identify the genetic cause for one of the clinical features seen in some patients with a 6q deletion. Unfortunately we cannot give you more information at this stage, but as soon as our paper is accepted for publication we will let you know all about it.

As soon as that paper is finished, we will begin working on a second paper giving an overview of all the features seen in 6q deletions. Further work will include analyzing 6q duplications and the 6p abnormalities. There is a long way to go and we are still gathering more information: by sending out questionnaires to new participants and requesting medical information from their doctors.

Question: What do the letters and numbers in an array result mean?

Some of the questions from your group concerned array results and what all the numbers, letters and abbreviations in these results actually mean. Below we explain how to read an array report. The format used is identical for all array reports and follows international guidelines.

The formal results are likely to look something like this:

arr [hg18] 6q25.1q25.3 (152,039,128-158,147,951)x1

We can break down this result and explain what these letters and numbers mean.

arr

The analysis was done by an array (arr) technique. This could be an array CGH or SNP array, for example.

hg18

hg18 refers to 'Human Genome build 18'. This is the genetic 'language' in which the report is written. The number refers to the reference DNA sequence that was used to compare to the DNA of your child. As more information about the human genome becomes available, new 'builds' of the genome are made (the most recent is hg19, while hg20 will be used in the future). In some reports the old name, Genome Reference Consortium (GRCh), is used. For reference, GRCh36 = hg18 and GRCh37 = hg19. All the different genome builds can be translated to the most recent genome build (hg19).

6q25.1q25.3

In this report the chromosome involved is number 6, and the chromosomal bands involved are numbered 25.1 up to 25.3 on the long (q) arm of the chromosome. In a different report the short (p) arm might be used instead.

(152,039,128-158,147,951)x1

DNA is a long string of building blocks called base pairs. All base pairs in a chromosome are numbered from the top to the bottom of the chromosome, so from the top end of the short (p) arm to the end of the long (q) arm. In this example, the DNA between base pairs 152,039,128 and 158,147,951 is only present once (x1) instead of as the normal two copies. This is therefore a *deletion*. In the case of a *duplication*, there would be three copies (x3).

6.1 Mb

To calculate the size of the deletion, the lowest base pair number (see above) is subtracted from the highest base pair number. In this example the size is $152,039,128 - 158,147,951 = 6,108,823$, or approximately 6.1 million base pairs, which is abbreviated as 6.1 Mb. This is the number of base pairs that are deleted (or duplicated if the result reports x3).

We hope this explanation is helpful, but do please contact us if you have any questions about the array result for your child.

Many thanks for your continued support!

We are still waiting to find out whether the research proposal that we submitted to the Dutch funding body *ZonMw* on 5th February will be funded. We expect to hear the answer in June. In the meantime, your group has collected money that has enabled Barbara Frentz to continue working on the Chromosome 6 project for one day a week for the past couple of months. We sincerely thank all the parents who have donated and/or raised money by organizing fundraising events. We are continually impressed by the creativity of your group and we have given an impression of your hard work on our own webpage: http://www.rug.nl/research/genetics/research/chromosome_6/fundraising-for-chromosome-6-research.

Please also check out the Chromosome 6 JustGiving webpage:
<https://justgiving.nl/en/charities/388-umcg-chromosome-6-research>

More information

For more information please visit the website of the Chromosome 6 Research Project at http://www.rug.nl/research/genetics/research/chromosome_6/

This study is still open to new patients, so if you are interested, please do let us know! In addition, please feel free to e-mail us if you have any questions or ideas for topics to be addressed in our next Facebook update.

With best wishes,

Barbara Frentz and Prof. Conny van Ravenswaaij-Arts

Department of Genetics

UMCG Groningen

Email: Chromosome6@umcg.nl Web:

http://www.rug.nl/research/genetics/research/chromosome_6/

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