

Dear 'Chromosome 6' parents,

It's been some time since we updated you about the chromosome 6 project, so we'd like to inform you about the current status of our Chromosome 6 Research and answer some of the questions that were forwarded to us by Pauline. However, we are still gathering more information and have a long way to go.

Number of participants

The number of participants is still rising! Over 220 parents have shown interest in our study by sending an email. About half of these parents have returned the questionnaire and consent forms and at the moment we have 74 'complete' 6q files and 13 'complete' 6p files (i.e. files with the questionnaire responses, medical records, and the array results).

Chromosome 6 funding

We heard from Pauline that some people did the 'ice bucket challenge' not only for ALS but also for C6! How brave and wonderful of them! We sincerely thank all the parents who have donated or raised money by organizing all kinds of fund-raising events. We are very impressed by all your creativity.

The money you have collected for the study is being used for hiring a physician-researcher (Barbara) for 1 day a week. The costs are about €1,150 per month. We realize this is a lot of money, but Barbara is on as low a salary scale as possible for the study and staff costs in a Dutch university medical centre are high due to social and other taxes. There have been no other expenses involved. With the latest budget report of the C6 money, we have calculated that Barbara can spend 1 day a week on research until the end of October.

Of course, we too are also trying hard to obtain more funding. A grant application to the Dutch Association of Science (for €200,000) earlier this year was unfortunately unsuccessful. A recent and more modest proposal (for €50,000) has passed the first round of the review process and on September 26th we heard that we can present our proposal in person to the assessment committee on October 30th.

Pauline asked us to post a list of givers on the C6 research webpage. This is an excellent suggestion and we will draw up a list on the website. If you don't want your name to be listed for any reason, please let us know!

We will also draw up our annual financial report, which will also be posted on the C6 research webpage.

Articles

Our analyses of the chromosome 6 data are progressing steadily. We are now working on the first articles, which we will submit to a scientific journal for publication. The work is proving very exciting since we have been able to localise the genetic cause for some of the clinical features seen in patients with a 6q deletion!

We were making good progress on an article about microcephaly (small head circumference), since it seemed we had found the responsible gene, but unfortunately that has become less likely after further research. We have not yet received information on the head circumference for all the chromosome 6 children. A few weeks ago we asked some of the parents to measure the head circumference of their child and we sent instructions on how to do this. We are still waiting for measurements from some but hope to receive them soon by email.

We are also working on a review in which the various groups of 6q deletions are described. It is similar to the 'sneak-previews' that we have put on Facebook during the Barbara's internship research project, but in far more detail and on a larger number of children. Later, we will analyse 6q duplications and the 6p abnormalities.

We are sorry that we cannot give you more information about the articles at this stage, but as soon as one is accepted for publication we will certainly let you all know.

Interactive webpage

As described above, we applied for a large grant that unfortunately was not awarded, so the interactive website is not yet available. A few weeks ago we applied for a second grant, in which 'e-health' plays an important role. If we are awarded this grant of approximately €50,000, we can create a multilingual website with a portal where parents can easily upload the information on their child. Our aim is for the new information to be linked to a database automatically and incorporated into the information on the website.

Such an interactive website with automatic data analysis needs to be built by IT experts in our university medical centre, but of course, they need to be paid for their services. The good news is that we are hopeful of getting the €50,000 funding. We will know more after October 30th!

Conclusion

Progress is slow but nonetheless, we are moving forward and hope to inform you in the upcoming months on the physical and other effects of deletions and duplications of specific parts of chromosome 6.

Our study is still open to new patients, so if you are interested, please let us know by email (see email address below)! And please e-mail us if you have any questions or ideas for topics to be addressed in our next Facebook update.

In the next update we would like to explain why two people with the same deletion or duplication do not have exactly the same clinical problems.

With best wishes,

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