

March 2015

Dear 'Chromosome 6' parents,

Here's a Spring update for you on our chromosome 6 project, with news about the start of the multilingual web portal project and the current status of our chromosome 6 research.

Multilingual web portal

We have been awarded a grant for the development of a multilingual web portal. We started working on it in January and it will be used to collect data on patients with chromosome 6 deletions and duplications. This portal is the first step in the process of creating an interactive website that will enable parents and professionals to find integrated and up-to-date information on these rare chromosome aberrations.

We will create a new webpage and an online questionnaire that will enable parents to easily upload information on their child's case and progress. Information can be saved during this process and completed at a later stage if necessary. In the future it will also be possible for parents to modify or add follow-up information on their own child.

The first test version of the website will be in English, but the interactive questionnaire will be translated into as many languages as possible, as soon as it has passed the test stage. Translation into other languages is possible because the questionnaire will have no "open questions". All answers can be chosen from a list of options and all the options are linked to a code. So that if, let's say, a parent fills in the Japanese version of the questionnaire, we can still interpret his or her answers.

We are currently working on transforming the paper questionnaire into a translatable web version. We are making steady progress, but it will take a few more months before the first test version is ready. We will ask some parents to join our panel to test this first version. We will ask the test panel to fill in the questionnaire as well as possible and to mark anything that is unclear, missing, or needs adapting. The remarks of the test panel will be used to optimize the questionnaire and the information on the website. When the webpage and online questionnaire are ready, we will invite all of you to fill in the questionnaire.

Current status of the chromosome 6 research

We are also working on the interpretation of the chromosome 6 data we had already collected with the paper questionnaires. The first group to be analyzed concerns the most proximal 6q deletions. In our next update, we will give you some information about our findings, similar to the 'sneak-previews' that we have previously put on Facebook.

Number of participants

The number of participants is still rising! Over 230 parents have shown interest in our study by sending us an email. About half of these parents have returned the questionnaire and consent forms and we now have 76 complete 6q files and 14 complete 6p files (i.e.

patient files with the questionnaire responses, medical records, and the array results). Of these complete files 7 are 6q duplications and 2 are 6p duplications.

Chromosome 6 funding

We are sincerely grateful to all the parents who have donated or raised money by organizing all kinds of fund-raising events. We are truly impressed by your creativity and dedication. In 2014 parents, grandparents, family members, neighbors, colleagues and others raised the amazing amount of €6,261. You can find a list of sponsors on our webpage (http://www.rug.nl/research/genetics/research/chromosome_6/thank-you).

Finally

Prof. Conny van Ravenswaaij will be in Chicago at a conference for a few days this summer. She will stay at the Renaissance Schaumburg Convention Center Hotel and she is happy to meet you there in the afternoon of August 2nd. If you are interested, please send an e-mail including your name, the name and birth-date of your C6 child and with how many people you will come to Pauline paulinebouman@live.nl or to us chromosome6@umcg.nl

This is a short, spontaneous and informal event and there will be no facilities for children. Conny will explain about the ongoing research and demonstrate the web site and on-line questionnaire. She is happy to answer all your questions.

Further details will follow as soon as we know how many people are interested in this informal meeting.

Our study is still open to new patients, so if you are interested, do please let us know by sending an email to chromosome6@umcg.nl. And please email us if you have any questions or ideas for topics to be addressed in our next Facebook update.

Best wishes,

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