

Newsletter June 2019

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1. Connecting the dots - GNAO1 foundations -

The past six months were mainly dominated by making connections and creating visibility.

As far as we know, there are now 3 foundations active worldwide, all of which have set themselves the (joint) objective of promoting research into GNAO1. These are:

- Stichting GNAO1 NL (the Netherlands)
- The Bow Foundation (America)
- Famiglie GNAO1 (Italy)

There is close contact with the aforementioned foundations and we are working hard together on the common goal and we will increasingly join forces. This is a good development. The rarity of the mutation makes it necessary, together we are stronger!

The Bow Foundation (www.gnao1.org) was established at the same time as our foundation and has since financially supported several scientific studies. In addition, the Bow Foundation is the organizer and driving force behind an annual medical conference on GNAO1 in America. The last conference took place in March this year and was very well attended by families from within and outside America. The video images from the conference are definitely recommended for those interested in GNAO1 and can be viewed via: <https://bit.ly/2IqGBo5>. A conference will also be organized in 2020. As a part of the conference people can also attend sessions in which patients can see and consult the doctors present. We will keep you informed of this on our Facebook page.

The Italian Famiglie GNAO1 (www.gnao1.it) is a foundation in formation with great ambitions. In November 2019, this foundation is organizing a first Italian conference on GNAO1. This conference is in principle organized for Italian families, but several doctors from abroad will speak and we are very pleased that Prof. dr. Dr. Frank Baas has also promised to be present at this conference to explain the research in The Netherlands to those present. In our next newsletter we will report on the conference in Italy.

1. Connecting the dots - connecting the doctors -

Over the past six months, the foundation has worked to connect doctors with GNAO1 patients in the Netherlands as well as to increase the visibility of GNAO1 among neurologists in the Netherlands. A round of visits to the Radboud UMC, the Amsterdam UMC and the UMC Groningen hospitals led to good contacts and hopefully contributed to the doctors pointing new patients to our foundation. Our second goal was for neurologists treating GNAO1 patients to find each other quickly and for optimum knowledge transfer to take place. Because of the rarity of the mutation, it is to be avoided that the wheel will have to be reinvented again and again. Partly the doctors were able to reassure us in this by informing us about the current channels of knowledge and information exchange and partly here is also a role for the foundation (which we already fulfill) by bringing important information to the attention of the various doctors as well as bringing doctors (at home and abroad) into contact with each other. We notice that there is also a positive movement in this and the foundation is actively consulting with doctors to look into the possibility of setting up a knowledge point for GNAO1 within one of the Dutch hospitals so that expertise can be bundled and optimized. Hopefully more about this in the next newsletter.

2. The current state of the research

As indicated in the previous newsletter, the nerve cells developed here in the Netherlands by Prof. dr. Dr. F. Baas are currently in America, in the lab where the research under the direction of Dr. McConnell at the University of Virginia is taking place for RNA sequencing. This takes time. Dr. Baas and Dr. McConnell will most likely meet each other this summer to discuss the continuation of the investigation. The hope is then that this autumn there will be more clarity about the next step in the investigation and the finances required for this. For clarity's sake, the phase that the research is now in, is: characterizing the so-called phenotype i.e. trying to discover the deviating molecular pathways (what works differently for a nerve cell with a GNAO1 mutation compared to a nerve cell without the mutation).

3. International developments

In our newsletter of last December we also mentioned other relevant scientific developments. Dr. research Neubig at Michigan State University where existing drugs are tested on mouse models was one of them. There has been a recent publication about this, see: <https://www.biorxiv.org/content/10.1101/662031v1>.

4. Visibility and Collection Actions

Visiting the hospitals and meeting the doctors also contributed to the visibility of the mutation and the foundation. In April we were also allowed to bring the GNAO1 mutation

and the activities of the foundation to the attention of a full lecture hall with medical students. This happened on the initiative of Dr. Bakker from the Amsterdam UMC, who was recently involved in the first Deep Brain Stimulation (DBS) procedure in a young Dutch patient with GNAO1 (a friend of Max). He invited the mother of this patient and our foundation to share our experiences. This was a special and valuable experience.



Over the past six months, no less than € 6,000 has already been raised for scientific research into GNAO1. We sincerely thank all (regular) donors! This time we would also like to thank the organizers of and participants in the large collection campaign that took place last June; The Ride to the Max. The group of cyclists who participated raised no less than 3,000 Euros in one go! We sincerely hope that we can turn this into an annual event.



The fact that such a nice amount has already been raised in the first half of the year means that there is an upward trend in donations! The foundation has since raised a total amount of € 20,000 in donations. In the fall, we hope to know what amounts are needed for the next steps in the investigation, and to what extent additional funds are needed for this.

The following collection actions are still on the agenda:

- Eva Broekhuizen is swimming the Bosbaanswim on 25 August 2019
- Hanneke Kortmann runs the Tilburg Ten Miles on 1 September 2019
- Patricia van de Ven runs the 20km from Paris on October 13, 2019

Are you also joining us or are you coming to encourage us?

Or keep an eye on our social media and website to find out how you can support the collection campaigns.

5. Website

Behind the scenes we are working hard on a new website where our main goal is to make it as accessible and appealing as possible for all target groups.

6. Call for help

The foundation is looking hard for people who want to help the foundation (once or structurally) by starting an action themselves or by encouraging others to do something for the foundation in their own network!

One-off: this can be anything! You can:

- do something active and get sponsored, or;
- maybe the sports club or school is still looking for a charity for the new year? Or;
- for example, you can also look for a good place for our donation box or hold a garage sale for the benefit of the foundation.

Do you think you have a good plan? Let us know and we will gladly help you further. Also don't let yourself be deterred by doubts about material and the like; we are happy to help you with flyers, t-shirts or other supporting material.

With structural help we think, for example, of people who help the foundation by drawing attention to us and our activities where they can, in their own network or in the local newspaper etc. etc.

Here too applies: together we are stronger!

Until the next newsletter! Do you have questions in between? Feel free to mail us at info@гнао1.nl.

Sincerely,
Niek Urbanus, Ed Broekhuizen, Dorine van Haselen, Hanneke Kortmann and Eva Broekhuizen.