Sixth edition, October 2018

Newsletter AHDS/ MCT8 Deficiency

Dear parents, doctors and all who care for people with MCT8 deficiency,

this is the sixth newsletter in our series of newsletters on the AHDS or MCT8 deficiency. This special edition focusses on the recent MCT8 symposium, the MCT8 patient registry and the upcoming international MCT8 day.

Progress of Triac Trial I

The Triac Trial is currently ongoing. Inclusion of new patients in the trial is not possible.

8th MCT8 deficiency symposium

On 10 and 11 September, the 8th MCT8 Deficiency symposium was held in Doorn, The Netherlands. The meeting was attended by parents, researchers and physicians from all over the world. During the meeting, ongoing research in different animal models and several potential therapeutic approaches were discussed in a constructive way. Also, new data of the Triac Trial I was presented. Importantly, the clinicians, researchers and patient representatives (parents) agreed to avoid the term 'psychomotor retardation', since parents can experience these words in an offensive way. Instead, 'intellectual and motor disability' is the preferred term when one refers to the neurocognitive part of MCT8 deficiency.

As concluded during the meeting, research will continue to be focussed on understanding different aspects of the disease and on exploring several potential therapeutic approaches.

International Patient Registry launched!

As already mentioned in our fourth newsletter (November 2017), we have been working on an international database/registry for MCT8 deficiency patients. Recently, this international patient registry has been officially launched! The registry adheres to all legal, ethical and security requirements and therefore is a safe place to collect information about patients with MCT8 deficiency. We strongly believe that your participation will be of great help to increase the understanding of MCT8 deficiency and to improve the care for each individual patient with MCT8 deficiency. If you are interested in participating in the registry, please visit our website mct8registry.erasmusmc.nl to find out more and join!

MCT8 deficiency
International registry for rare thyroid disorders

WORLD MCT8-AHDS DAY

On October 8th this year, the first World MCT8-AHDS Day will be held. This event is initiated by the MCT8-AHDS Foundation, the first world-wide patient led organization, in order to raise awareness for the disease. Also, the MCT8 website (mct8.info) will be updated, there will be several patient movies and a social media frame (Facebook) in order to celebrate the event. We spoke to Veronica Popa, one of the initiators of the first World MCT8-AHDS Day.

Veronica, how did you come up with the idea of an international MCT8 Deficiency day?

Thank you for asking Ferdy! The idea of having a World MCT8-AHDS Day has been with the community for a while, it just never seemed the right time. However, once we have created the MCT8-AHDS Foundation, with the help and support of the Sherman Foundation, things have changed. We started dreaming a bit bigger and that became the right time.

What is the main goal of this day?

The main purpose of the World MCT8-AHDS Day is to raise awareness. To let people know we exist, that our challenges are real and sometimes severe; to send a message of urgency as there is no cure or treatment for MCT8 deficiency and last but not least, to have a voice. On the longer run however our goal is to change perception and behaviour. People acknowledging the existence and severity of this syndrome is desirable but people wanting to get involved and help, people changing their perception and behaviour towards our children, that is better. And that is what our final goal is.

What is your reason to pick the 8th of October?

Well, there was a lot of debate regarding the date in which to celebrate World MCT8-AHDS Day. But we all agreed that October 2003 was a turning point in MCT8-AHDS research. This major breakthrough in the research of AHDS was the publishing of a paper connecting the Allan-Herndon-Dudley syndrome phenotype with the Monocarboxylate Transporter 8 (MCT8). Finally, the day was given by the transporter, the MCT8, therefore the 8th of the month. Choosing this date is also our way of showing respect and gratitude to the research and memory of Prof. Dr. Theo Visser.

What does this day mean to you and the MCT8 community?

The World MCT8-AHDS Day represents our birthday as a community with all the hopes and expectations that birthdays bring. It's the day in which we forget a bit about our medically complex lives and we gaze towards the future, a day in which we celebrate our children and the bond we have as a MCT8-AHDS worldwide family. It is a celebration of joy and hope but it is also a day of recognition of the efforts, the struggle and challenges that our community faces.

Thank you, Veronica!

MCT8 - AHDS Foundation
Ultra-rare syndrome
Striving for a cure. Empowering families

Renewed website!

As part of the campaign for raising awareness of MCT8 deficiency, the current MCT8 deficiency website (mct8.info) will be renewed. This website is meant to be supportive for parents with children with MCT8 deficiency and also contains background information on the disease. Also, these newsletters are published via these ways. The website is supported by the Sherman foundation and the Erasmus MC. We are looking forward to the renewed website!