Erasmus MC
University Medical Center Rotterdam

First edition,
August 2015

Newsletter AHDS

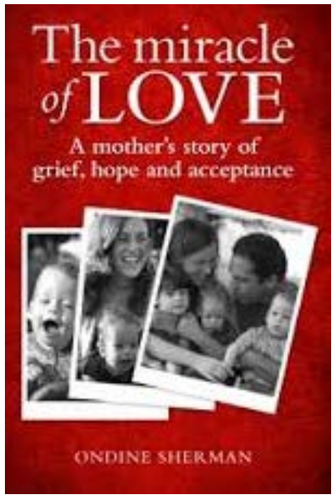
Dear parents, doctors and all who care for people with AHDS,

We are excited to present to you the first newsletter about the Allan-Herndon-Dudley syndrome (AHDS) and the Triac Trial. You have probably noticed there is limited information available on AHDS apart from research papers. At the Erasmus MC we have been studying the AHDS for many years now. We think it is important to share the available information with you through this newsletter.
In this first newsletter we would like to introduce three members of the research team: Edward Visser, Stefan Groeneweg and Arjanne Aleman. We will also give an update about the latest progress of the Triac Trial, the first clinical study in which the effect of a putative therapy with Triac is under investigation.
What can you expect from this newsletter in the future? First, we will provide more information about the AHDS and its research. Second, we would like to keep you updated about the Triac Trial. Furthermore, you can ask any question to the research team, or to a doctor, dietician, physical therapist or a speech therapist that will be answered in the newsletter.
In addition, Arjanne will write about visits from Trial participants to the Erasmus MC. If you would like to write something about your experiences with AHDS we would like to hear it. If you know anyone else who would like to receive this newsletter, please contact us at a.aleman@erasmusmc.nl.
Enjoy reading this first newsletter!

Going abroad

Because AHDS is such a rare disease, and there are little alternative treatment options, we have expanded our trial abroad.
We are now working on the trial in twelve different countries. As this is an official trial, this brings about a lot of work. This includes obtaining consent from each medical ethical committee, ensuring proper safety measures, insurance issues and so on. In some countries we are now ready to start with the trial. We will keep you informed of further progress.

Suggestion to read



Ondine Sherman, daughter of the founders of the Sherman foundation, wrote a book about her life. Central in the book are her twin sons, both born with AHDS.
In The miracle of Love Ondine Sherman shows the reader an insight in her, at first, perfect life. She grows up in a rich family, is ambitious and marries the love of her life. The birth of her first born makes her life even more complete. A few years later she gets pregnant with twins and a few months after the birth the worries begin. Dov and Lev's development is slower than usual. The search for a diagnosis leads them to many doctors and hospitals, and the desperate parents also visit religious figures to get answers. After a long search the boys get diagnosed with AHDS. While her husband is focused on research, Ondine tries to stimulate her sons as much as possible in their development, with the help of many therapists.
A heartbreaking story, which is without a doubt very easy to identify with for parents and caregivers of children with AHDS. The book is available on paper and as ebook on amazon.com.

Stefan Groeneweg

In 2007 I started my study medicine at the Erasmus MC. Not only did I want to become a doctor, I especially wanted to contribute to the research on (rare) diseases.
That's why I decided to start with a master on Molecular Medicine in 2009. Then I contacted the head of the thyroid laboratory Prof. Theo Visser. He rapidly transferred his enthusiasm for thyroid hormones to me. Since then (early 2010) I`m involved in the research on the Allan-Herndon-Dudley syndrome and the function of the thyroid hormone transporter MCT8 with a lot of gratification and dedication. Since the discovery that defects in MCT8 are the cause of the Allan-Herndon-Dudley syndrome in 2004, we started our search for putative treatment options for AHDS, which eventually resulted last year in the start of the Triac Trial. Currently, I am one of the coordinators of the Triac Trial. It is an unique experience to see that all families, doctors and researches pursue the same objective: finding solutions to improve the care for these boys! I am very grateful to be able to contribute to this work and be part of the growing global MCT8 network.

Edward Visser

In 2000 I started my study medicine at the Erasmus MC. Since 2010 I started my specialization in internal medicine and since this year I am specializing in endocrinology in the Erasmus MC. Since my PhD research (supervised by prof. Theo Visser), where I studied MCT8 and the AHDS, I became fascinated with the regulation of thyroid hormones on cellular level by thyroid hormone transporters, deiodinases and nuclear receptors and their importance in health and disease. This means in plain English: how does thyroid hormone get to the right place at the right time? It is of the utmost importance to bundle all expertise and knowledge of rare diseases. Specifically, this means that we will bring together all information on patients with MCT8 mutations worldwide. The Triac Trial is an example how patients with the same rare condition are now treated in the same way throughout the world. This is crucial to improve the health of every individual patient and of the group at the same time. Apart from that we hope to contribute by bringing parents, patients, doctors and researchers from different countries together to improve the exchange of information.

Arjanne Aleman

In March 2015 I started as a research nurse to work together with Edward and Stefan on the Triac Trial. Because the trial was expanding to other countries, help was needed urgently. I have been a nurse for many years and I just finished my study journalism. The combination of these two studies is very helpful with this job. Previously I worked as a nurse on different wards in the Erasmus MC wich was completely different from what I do now. It took some time getting used to, to not be running on the ward all day and mainly care for patients but mostly do my work on a computer. That's why I still work a shift in the hospital from time to time. That way I also do not forget how to draw blood, and other technical procedures.
Meanwhile I have, apart from all the paperwork, also met a few patients for baseline and control visits. For me it was very motivating to see a face behind the syndrome. I am really looking forward to contribute to this research that will hopefully improve the quality of life of all patients with the Allan-Herndon-Dudley syndrome. Hopefully we will all get to meet one day!