

# Newsletter AHDS

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

Hereby the second newsletter about the Allan-Herndon-Dudley syndrome and the Triac Trial. A few weeks ago we officially started the Triac Trial outside of the Netherlands. South Africa is the second country where a participant has started with Triac. In several other countries we are almost ready to start.

In this newsletter you can also read about our plans to organize a meeting about the AHDS. We would like to hear if you are interested to attend.

Furthermore we will give you, as promised, information about the Allan-Herndon-Dudley syndrome. We start this information at the very beginning, with an explanation about the thyroid hormones and their function. If you have any further questions about this, please don't hesitate to contact us.

## Meeting AHDS

In October 2015 it has been a year since the Triac Trial started in the Netherlands. Therefore, we would like to organize a meeting about the Allan-Herndon-Dudley syndrome (AHDS) for parents, doctors and other interested parties.

The goal is to present the latest results on the Triac Trial and to share experiences between parents on Triac treatment as well as on the AHDS in general. As we have expanded the Triac Trial to other countries, this meeting will be international.

This meeting will take place in the first week of December 2015, the exact date will follow. We would like to know if you are interested to come. We would also like to receive input for the program. What is important for you to hear about? If you consider to attend, please send an e-mail before the 10th of October 2015 to: [a.aleman@erasmusmc.nl](mailto:a.aleman@erasmusmc.nl).

## Thyroid hormones and their function

To understand what goes wrong in Allan-Herndon-Dudley syndrome (AHDS), it is important to have some knowledge about thyroid hormones and their function. In this first part of a series about the AHDS you will find an explanation about thyroid hormones and the function of the hypothalamus, the pituitary gland and the thyroid gland.

In our body there are three glands involved in the regulation of thyroid hormone. The hypothalamus and the pituitary gland, next to each other in the brain, and the thyroid, an organ in the form of a butterfly in the neck.

The hypothalamus produces TRH (thyrotropin releasing hormone) which stimulates the pituitary gland to produce TSH (thyroid stimulating hormone; other name: thyrotropin). TSH which stimulates the thyroid gland to make thyroid hormones: thyroxine (T4) and to a lesser extent also triiodothyronine (T3).

T4 is the inactive precursor of T3 and is only effective when converted to the active hormone T3.

This process takes place in different tissues and organs.

The hypothalamus and pituitary both sense the thyroid hormone levels in the blood. Excess of thyroid hormones in the blood and tissues decreases the production of TRH by the hypothalamus and TSH by the pituitary. As a consequence, the thyroid gland decreases the production of T4 and T3. And the other way around: shortage of thyroid hormone results in increased production of TRH and TSH by the hypothalamus and pituitary, respectively. With this we have come full circle.

When this all goes well, the body has sufficient energy, just the right temperature, and a stable heartbeat and weight. During pregnancy and the first years after birth, thyroid hormones also play an important role in the development of the brain.

Thyroid hormone is transported via blood to the different tissues of the body that need the hormone, such as the brain. To enter tissues and cells, thyroid hormone requires specific transporter proteins in the membrane of cells. These transporters function as "gates" to facilitate cellular entry of thyroid hormone. MCT8 is one of the most important "gates". MCT8 is required for T3 to enter the brain. Children with the Allan-Herndon-Dudley syndrome have a defect in MCT8, preventing T3 access to the brain. This has severe consequences.

In the next newsletter we will provide more information about those consequences.

