

Dear parent/guardian,

We are writing to invite you and your child to take part in a new research study led by Center Medical Genetics Antwerp at the University of Antwerp Belgium. This research study focuses on individuals with Helsmoortel-Van Der Aa syndrome (HVDAS) and aims to explore the relationship between brain development and clinical outcomes by comparing early-life and later-life MRI-images and linking these findings to clinical information. Given the rarity of Helsmoortel-Van Der Aa syndrome, this study holds significant potential to advance our understanding of how the condition affects brain development over time.

Your involvement could greatly enhance our understanding of how HVDAS progresses and help shape future diagnostic and therapeutic approaches. Please rest assured that all participant information will be handled confidentially and in accordance with ethical guidelines (**see information section in the informed consent form**).

Participation will involve the following three components:

- 1) Access to **all previously conducted brain MRI scans**:

Send all available MRI scans on a CD by post. If you do not have a CD, please provide us with the contact details of your neurologist/radiologist in order for us to request the necessary files.

Please note: Screenshots/PNGs/JPGs are not sufficient, as we need full access to the complete MRI scan.

- 2) Contributing **clinical data** such as relevant medical history and genetic information relevant to HVDAS:

You will receive a link to a questionnaire (RedCap) to fill in all necessary clinical parameters. You are welcome to complete the survey yourself or ask your child's physician or clinical geneticist to do so. The survey should take approximately 30-45 minutes to complete, and your progress can be saved, so you may return later to finish at any time.

- 3) Delivery of **blood sample** of your child

For genetic research, we require 5-10 ml heparin blood (no EDTA!). If you live within a 200 km radius of Antwerp, one of our researchers can pick up the sample within 12 hours of collection. Please contact us for further arrangements.

Alternatively, you may send the sample by post (DHL; room temperature) to the following address:

Claudio D'Incal
Center Medical Genetics Antwerp
Prins Boudewijnlaan 43/6
2650 Edegem
Belgium

If you decide to take part in this research study, it is important that you sign the **informed consent form** before we can officially include you and your child in the study. We have provided this form in the attachment of this email, and we are happy to go over any questions you may have before you sign.

If you have any questions or would like more details, please do not hesitate to reach out to us at

adnp-nd@uantwerpen.be.

Kind regards,

Center Medical Genetics

University Antwerp