SLOS Natural History Project

A new project for persons with Smith-Lemli-Opitz syndrome and their families

Dear family with a child (of any age) with Smith-Lemli-Opitz syndrome,
We are happy to inform you about a new project for persons with Smith-Lemli-Opitz syndrome (SLOS), ***the SLOS Natural History project.*** We are patient representatives in MetabERN[1] for persons with SLOS. MetabERN is a European Reference Network on Hereditary Metabolic Disorders, established by the EU.

Improve counselling and develop therapies
The new project aims to improve the counselling of families and establish strategies for therapeutic interventions and long-term healthcare. General symptoms and rare conditions will be described, as will the frequency of sleep disturbances, autism and complex behaviour affecting the families.

We hope you will answer a short survey about which symptoms and aspects/features of SLOS you find essential and challenging and if you are interested in getting more information about the project or joining a family network.
Answers from families who have lost one or more children due to SLOS are also very important to us and we appreciate any input.

SLOS is a metabolic disease
Since the discovery in 1993, we have known that SLOS is an inherited metabolic disease. This is vital knowledge to find the right treatments. The cause of SLOS is an error in the synthesis of cholesterol in the body due to genetic changes (DNA variants) in the so-called DHCR7 gene. There are more than 200 different types of such variants causing SLOS.

Several biochemical processes are affected
The defective cholesterol synthesis affects several biochemical processes in the body in a complex way, resulting in varying signs and symptoms. Mildly affected persons may have no or minor inborn signs, near-normal cognitive function but often behavioural problems. A more severe impact of SLOS can be life threatening and involve malformations of the heart, lungs, kidneys, gastrointestinal tract and genitalia.

[11] More information: https://metab.ern-net.eu/

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Scarce knowledge about SLOS
There is still too little knowledge about SLOS. More clinical studies and research are needed to understand the complexity and variability of symptoms and courses of SLOS and for designing healthcare and therapy for persons with varying severity of SLOS. As patient representatives, we, therefore, welcome this project**.**

More data will be collected
The project leader is Dr Dorothea Haas at Heidelberg University Hospital, a member of MetabERN and of the official registry of MetabERN, the Unified European Registry for Inherited Metabolic Disorders[2] **(**U-IMD) that will be the platform of the study.

Dorothea Haas has invited health care providers and centres in MetabERN to participate in the study. Several SLOs patients are already included in U-IMD. More data will be collected and analysed to increase the understanding of this complex metabolic disease.

A survey for families
The project must get information from the families about symptoms and aspects of SLOS that you find essential and challenging. What affects the health of your child and everyday life, where more knowledge could make a difference?

Please answer the questions about that in our survey (https://www.surveymonkey.com/r/7DGQPL6). The results will be presented as statistics without names.

We also ask if you want to have more information at a digital meeting about the project SLOS Natural History and if you are interested in joining a family network connected to the project.

We are looking forward to being in contact!
E-mail address to us: parentsSLOSNH@metab.ern-net.eu

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[2] More information: https://u-imd.org/