Quality of Life in Female Carriers of X-linked Adrenoleukodystrophy

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What is the aim of the study?

X-linked Adrenoleukodystrophy (X-ALD) is a hereditary disease caused by mutations in the ABCD1 gene leading to disturbances in the metabolism of fatty acids. This results in an accumulation of very long chain fatty acids in the cells of the body causing damage to the central nervous system (white matter of the brain and spinal cord). The most common clinical form of X-ALD is adrenomyeloneuropathy (AMN), in which the symptoms usually first appear in early adulthood. It is often milder than the childhood form. Clinically leading is a slowly progressive spastic paraparesis of the legs. In addition, there are often bladder and bowel dysfunctions.

X-ALD is a rare disease that typically affects men due to the X-linked pattern of inheritance. It is now known that female carriers of X-ALD often develop neurological symptoms of comparable severity to those of male patients. The frequency of symptoms increases with age.

In this study we would therefore like to investigate the influence of neurological symptoms of X-ALD on the quality of life of affected women in various areas (including everyday life, work, social network, sleep quality, sexuality, mood).

What do I have to do in order to participate?

You will be asked to complete questionnaires about your neuropsychological symptoms due to X-ALD and its impact on your psychological well-being and quality of life in various aspects. The electronic questionnaires can be completed online via the Leuconnect platform, which will take approximately 40 minutes of your time.

What is my personal benefit from participating?

With your participation you contribute to gain new insights into the quality of life of female carriers of X-ALD. This will help to emphasize the need for early, profound diagnosis and treatment of neurological symptoms in women with X-ALD. A financial compensation for study participation is not provided. We will be happy to provide you with a summary of the results after the study is completed.

Who can take part in the study?

Female carriers of X-ALD (genetically verified) with or without clinical symptoms aged >=18 years.

What are the risks of participation?

Potential risks arising from participation are believed to be minimal. Short-term fatigue may occur as a result of the above-mentioned procedures, as the procedures make varying demands on the ability to concentrate. Certain diagnostic questions in questionnaires may lead to a slight and temporary emotional response.

Is a withdrawal of study participation possible?

You can withdraw your participation at any time without stating your reasons. Under certain circumstances, the study staff could decide to terminate your participation prematurely due to lack of cooperation or non-compliance (e.g., failing to answer questionnaires).

During the project, personal information will be collected, stored electronically in the testing center. In addition, your name, date of birth, address, and email address will be stored electronically in a separate database. This information is needed and used exclusively for getting in contact with you. Personal data is secured against unauthorized access, only investigators in the research project are granted access. Data which is used for evaluation and analysis within the research project is additionally stored in a pseudonymized form. Pseudonymized means all information containing your name or initials will be replaced with a numerical code. Data is secured against unauthorized access.

For further details, please read the informed consent form.