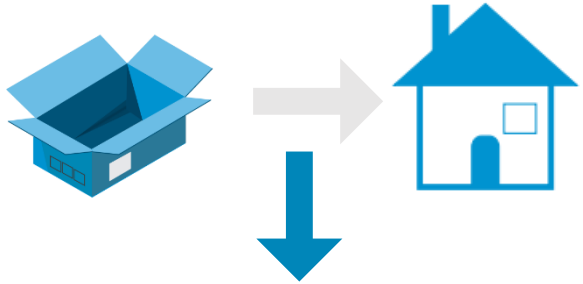
STUDY COURSE

Please call us or write us an e-mail if you want to participate:

Phone: 0341 9711796
Mail: mosaike@medizin.uni-leipzig.de

We will provide you with information about the study and answer your questions.

With your verbal consent, we will send you a small package containing all sample containers, an information sheet about the sample collection and the necessary consent forms.



You collect the samples at home

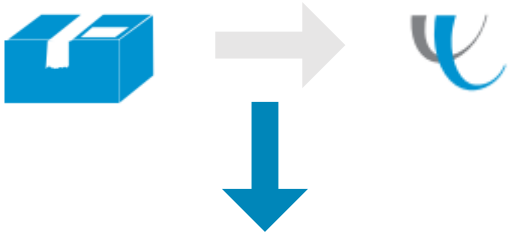
1. buccal swap

2. hair sample (eyebrows, leg-, armpit- or pubic hair)

3. blood sample

4. semen

After sample collection, you send the postpaid package back to us. Please do not forget to add the signed consent forms!



In our laboratory, we will analyse your samples specifically for the genetic variant, which was previously confirmed in your child. We can provide you with individual information about the results of the study during genetic counselling.

Universitätsklinikum Leipzig
Medizin ist unsere Berufung.

LOOKING FOR PARTICIPANTS

Low-Level Mosaicism in Various Tissues of the Parents in Case of Rare Diseases Caused by *de novo* Variants

Research Project:

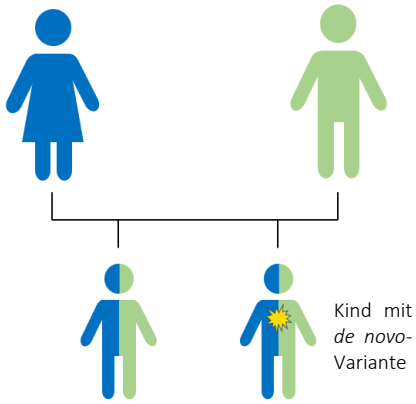
Institute of Human Genetics at
The University of Leipzig
Medical Centre

UNIVERSITÄT LEIPZIG
Medizinische Fakultät

WHAT IS A GENETIC *DE NOVO* VARIANT?

Every child consists half of the mother's and father's genes. A de novo variant is defined as a small change in the genetic information, which cannot be found in the parents DNA, but in the DNA of the child. This change is therefore **completely novel**.

According to the current state of knowledge, there is only a low risk (about 1%) for another child being affected with the same genetic disorder. However, this risk of recurrence can be much higher in individual cases.

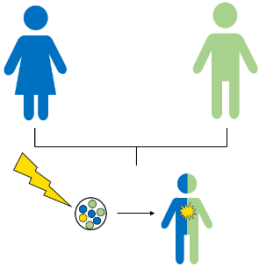


WHAT DOES THE STUDY CONSIST OF?

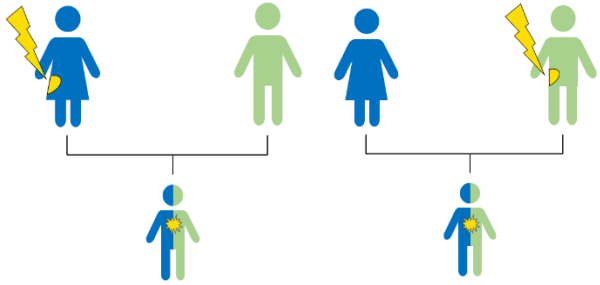
We will examine **various tissue samples** of the parents with very sensitive molecular genetic methods with a focus on the existence of **low-grade mosaicism**. We welcome all parents of a child with a rare genetic disorder **caused by a *de novo* variant** to our study.

HOW DO DE NOVO VARIANTS DEVELOP?

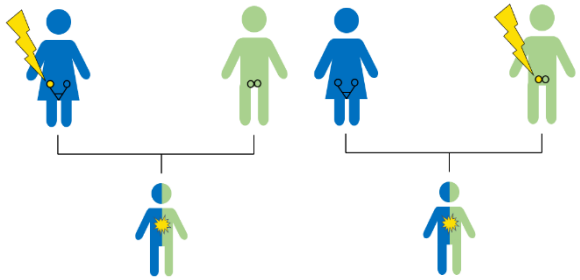
1. The variant develops sporadically **very early during the embryonic development**. The parents carry the genetic variant neither in their body cells nor in the germ cells.



2. A **very small fraction of body cells** of the mother or father carries the genetic variant (mosaicism). However, the parents do not show any symptoms of the genetic disorder, since most of the cells do not carry the genetic variant.



3. A **very small fraction of the male or female germ cells** (egg or sperm cells) carries the genetic variant (germline mosaicism). The parents do not show any symptoms of the genetic disorder, since their body cells do not carry the genetic variant.



WHAT ADVANTAGE DO I HAVE BY PARTICIPATING IN THE STUDY?

Generally, your participation will help us to achieve a better understanding of the human genome and the **origin of genetic *de novo* variants**.

The goal of our study is to, in a more precise way, assess the **recurrence risk** for another child being affected by the same genetic disorder. Should this be relevant for you, we may be able to provide you individual information on the risk of recurrence in the course of the study.

Your contribution to the study will help us to gather **new knowledge about genetic correlations**, which will improve genetic diagnostics and medical therapies in the future.

Hopefully, we could arise your interest to participate in our study. We would highly appreciate your support.

TO PARTICIPATE, PLEASE CALL US OR WRITE AN E-MAIL. IF YOU HAVE ANY QUESTIONS, WE WILL GLADLY ANSWER THEM.

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University of Leipzig Medical Centre
Philipp-Rosenthal-Str. 55
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Principal investigator: Prof. Dr. med. Rami Abou Jamra
Ethical vote: 014/19-ek

Phone: 0341 9711796
(Mo-Fr 8 bis 16 Uhr)

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