UNIVERSITY HOSPITAL
UNIKLINIKUM
SALZBURG

General Hospital Salzburg
Department of Dermatology of the Paracelsus Medical University Salzburg
Head: Univ.-Prof. Dr. Johann W. Bauer, MBA

eb Haus Austria.
debra Hilfe für die Schmetterlingskinder.

EB House Austria
EB Outpatient Unit
Müllner Hauptstrasse 48 |5020 Salzburg |Austria

# PATIENT INFORMATION SHEET
# FOR THE IMPLEMENTATION OF A GENETIC ANALYSIS

**Dear advice-seeker,**

for clarification/diagnosis of certain pathologies, analyses of the genetic material, so called genetic analyses, are necessary. **For those analyses genes (DNA) and, where required, gene products (RNA, proteins, metabolic products) from biological samples (primarily blood or other bodily fluids, cells or tissue samples) of you/your child/the person you are trustee for will be analyzed, to look for the genetic cause and risk factors of the disease.**
The most important examinations, and the ones provided by us or our external contract laboratories, are the so called

- **molecular genetic analysis**
- **genome-wide genetic methods** (exome sequencing, microarray)

Using different molecular genetic analyses, the respective **candidate genes** can be examined. An important part is

- the **analysis of deletions** (loss) and **duplications** (gain) of small or smallest fragments of genetic material
- the **analysis of mutations** (changes) of a single or multiple DNA-building blocks

Your doctor will inform you about the analysis that is planned in your case. We want to point out the following points, which apply for all genome-wide analyses:

- **The evaluation of the analysis focusses on the questioning.** Therefore, not all possible genetic factors in the genetic material, which (could) hold a risk of disease, are gathered and/or evaluated. Inconspicuous findings do neither preclude a genetic cause of the pathology, nor reveal anything about the disposition for other genetic diseases.
- **Auxiliary findings**, which point to risks of diseases that have nothing to do with the actual questioning, can occur. Since there is not always an effective treatment or prophylaxis for the respective disease, you will only be informed about such findings, if the **medical meaning is clear** and you confirm in writing on the **consent form**, that you want to be informed about them. Auxiliary findings, which only hold a slightly increased risk of disease for that person himself/herself (**genetic risk factors**) or possibly their descendants (**genetic carrier** of recessive diseases) are usually **not communicated**.
- Sometimes genetic changes, whose meanings are unclear, are detected. In these cases, this will be stated in the report and discussed with you. In these cases, an **examination of relatives** (e.g. parents, grandparents, siblings of the affected person) can make sense and may lead to clarification.
- **If multiple family members are examined, the accuracy of the stated family relationship is crucial for the correct interpretation of the results**. In the event, that the analysis creates any doubt about the stated family relationship, you will only be informed about this if it is crucial to meeting the requirements of our investigation.

If you still have questions concerning the points explained above, you can arrange (another) **appointment for a human genetic councelling**, in consultation with your attending physician**, at the EB-house Austria by calling the telephone number below.**