AMENDMENT to the DECLARATION FORM

FOR PARTICIPATION IN THE LONG-TERM OBSERVATIONAL STUDY / REGISTRY

Identifying the causes of severe chronic neutropenia, as well as the changes that lead to leukemia, can help us develop new treatments for neutropenia. By studying samples from patients, scientists can gain crucial insights that may lead to breakthroughs in treatment and prevention. The use of human samples, particularly for the generation of inducible pluripotent stem cells, also helps to replace animal studies.

Our SCNIR research laboratory at the University Hospital Tübingen (see address under point 10 of the information sheet) deals with the following main research topics:

1. Identification of genetic predispositions to leukemia in children and adults.
2. Identification of physiological mechanisms that drive normal hematopoiesis and the study of pathomechanisms of bone marrow failure, particularly in neutropenia.
3. Development of new treatment approaches for neutropenia, including new drugs based on chemical and protein-based substances, as well as development of gene therapies based on CRISPR/Cas-mediated insertion or correction of gene mutations in the *ELANE, HAX1, JAGN1, SBDS, SRP54, CLPB* genes. The same applies to so-called candidate genes whose role in neutropenia has not yet been defined.
4. Establishment of experimental models of neutropenia and leukemia, such as the cultivation of bone marrow or blood cells, genome editing of bone marrow cells, generation and genome editing of inducible pluripotent stem cells, transplantation of bone marrow cells in mice or zebrafish.
5. Comprehensive examination of primary and cells modified in the laboratory.
6. Early diagnosis of leukemogenic transformation in bone marrow and blood samples from patients by deep sequencing, compared to routine bone marrow and blood analysis of cell morphology.

With your willingness to participate in the long-term observational study/registry and biobank, you can become a valuable part of our team working to improve therapies and the quality of life of patients with congenital neutropenia.