

## ADDITIONAL INFORMATION ON THE DECLARATION OF CONSENT AND REQUESTS FOR ANALYSES

### Current research activities of the SCNIR

Our research laboratory, which is affiliated with the GPOH long-term observational study on neutropenia and the Severe Chronic Neutropenia International Registry (SCNIR), is located at the University Hospital of Tübingen, Department Internal Medicine II.

The current research areas of the laboratory are:

1. To identify **genetic causes of neutropenia, predisposition to leukemia and full-blown leukemia** in children and adults with neutropenia-associated bone marrow failure syndromes.
2. To identify **the processes that control normal hematopoiesis** and to study **the pathological processes that cause bone marrow failure and leukemia** in children and adults with neutropenia-associated bone marrow failure syndromes.
3. To develop **new treatment approaches** for neutropenia-associated bone marrow failure syndromes, in the neutropenia and leukemia stage including:
  - new drugs based on chemical and protein-based substances,
  - gene therapies based on CRISPR/Cas-mediated gene editing of inherited neutropenia-associated and somatic leukemia-associated gene mutations.
4. To establish **experimental models of neutropenia and leukemia**:
  - using patient-derived bone marrow or blood cells,
  - applying genome editing of patient-derived bone marrow cells,
  - generation and genome editing of patient-derived inducible pluripotent stem cells,
  - transplantation of bone marrow cells in mice or zebrafish,
  - comprehensive examination of patient-derived primary cells and experimentally modified cells using state-of-the-art methods and techniques in cell- and molecular biology, biochemistry, and genetics.
5. To establish state-of-the-art approaches for **early diagnosis of neutropenia and leukemia** in samples from patients with neutropenia-associated bone marrow failure syndromes.

By providing blood or bone marrow, you can make a valuable contribution to improving the diagnosis, management, treatment and quality of life of patients with bone marrow failure syndromes, particularly in patients with neutropenia.

We thank you in advance for your support!

Best regards,



Prof. Dr. med. Karl Welte, Prof. Dr. Med. Julia Skokowa, Dr. med. Cornelia Zeidler