Congenital Neutropenia Research
Genetic Analysis

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Patient: _____ **Date of Birth:** _____

Clinical Diagnosis and Therapy:

_____

_____

_____

Sample: ☐ Bone Marrow ☐ Peripheral Blood

Date Drawn: _____

Sender: (please print or stamp)

_____	_____	_____
Location, date	Signature	Telephone

Please send 1 – 5 ml heparinized bone marrow aspirate and/or 20 ml heparinized peripheral blood to the address indicated on top of the form.

Samples should be taken at the beginning of the week (Monday or Tuesday) and sent off immediately by overnight express!

Please notify Dr. Germeshausen prior to sending the sample:

phone: +49 511 532 - 9013, - 9036 fax: - 9783

Survey of inherited bone marrow failure syndromes

Medical School Hannover - Pediatric Haematology and oncology
M. Ballmaier, Ph.D. / M. Germeshausen, Ph.D. / C.P. Kratz, M.D.

We would like to invite you to participate in a research study with the goal to investigate the causes of congenital diseases affecting the formation of blood cells (hematopoiesis) and the function of the bone marrow.

Preliminary Note:
The participation in this study is voluntarily. You can decline to participate without any disadvantage for you or your child. You can withdraw your consent at any time.

What We Do:
We are investigating the causes of congenital disorders of hematopoiesis, especially congenital neutropenias and thrombocytopenias. These rare diseases might be of genetic origin, caused by acquired or inherited modifications in the genome.

The goals of our study are
- the identification of changes in the genetic material and the investigations of mechanisms which lead to impaired hematopoiesis,
- the analysis of correlations between the type of genetic modifications and the severity of disease (genotype-phenotype correlations),
- improvement of diagnosis and therapy for this group of rare diseases
- a better understanding of the functions of the hematopoietic system

To achieve this goals we will perform a set of laboratory analyses – depending on the specific disease and previous results – with blood and bone marrow samples and correlate the results with the patient's clinical data.

Your Contribution:
You agree that your physician send us a sample of blood or bone marrow for our investigations. Normally additional vein or bone marrow punctures are not necessary. In most of the cases an extra sample of blood or bone marrow cells can be taken during a regular control puncture.
You also agree with the collection and analysis of personal (name, date of birth) and clinical data (specific medical data related to your disease).

Advantages for You:
Our investigations are mainly designed to increase our understanding of the molecular basis of congenital diseases of hematopoiesis. Our results might help your physician in finding a diagnosis. We will inform your physician about important results of our investigations which can facilitate decisions on diagnosis or therapy.
You/ your child might not have any direct benefit of the study. However, the results of our study might help to improve diagnosis and therapy and thereby of benefit for future patients.

Risks and Potential Consequences of the Study
There is no direct risk of our study for you or your child since blood or bone marrow is taken during routine diagnostic procedures.
In this study we hope to obtain knowledge about the genetic origin of the disease and about potential inheritance of predispositons. You can decide whether you would like to know any genetic information - we will inform you about the results only if desired. The information about genetic results by your treating physician will include a genetic counseling.
Our study will reveal information about parenthood. This might affect aspects of fatherhood and adoption. We generally do not share any of this information.

Storage of Material
Blood or bone marrow from you or your child will be processed in our lab and separated in different components (e.g. Plasma, different types of blood cells). A part will be used for immediate analyses. Processed material, including DNA, from you can be stored for later analyses. In case you withdraw your consent to our study, all material will be discarded.

Confidentially / Data Privacy
All researchers involved in our study are obliged to keep all information about you / your child strictly confidential. Your specific results can only be shared with your treating physician – providing you agree. Research results will be shared with other scientists or published in scientific journals only in anonymous form. In case you withdraw your consent to our study, all files and electronic data will be deleted.

Informed Consent

I have read the information about the study and have had the opportunity to ask questions. I herewith declare my consent for **me / my child***

_____ DOB: _____
Patient's Name, First Name Date of Birth

to participate in the study, in particular (delete as appropriate):

- genetic analyses for identification of genetic causes of bone marrow failure syndromes and their possible involvement in leukemogenesis,
- analysis and storage of biological specimens for research on congenital bone marrow failure syndromes,
- the storage and analysis of personal and clinical data (name, date of birth, diagnosis, information regarding therapy and course of the disease) and of research results,
- sharing and publishing of research results in anonymous form.

You can decline to participate without any disadvantage for you or your child. You can withdraw your consent at any time. In case you withdraw your consent, all material will be discarded and and data will be deleted.

☐ Yes, I would like to be informed about relevant results concerning me / my child.* I know that the information about genetic results will include a genetic counseling.
☐ No, I would not like to know any results concerning me / my child.*

Patient

_____ _____
Patient's Signature Date

Parents of Minors

_____ _____
Parent's Signature Date

Physician
I have explained the study to the above signed person and declare that in my view the participant understood goals and consequences of the project.

_____ _____
attending physician Date

* delete as appropriate