

Manuscript no. HAEMATOL.2010.029389 entitled "Atypical familial hemophagocytic lymphohistiocytosis due to mutations in UNC13D and STXBP2 overlaps with primary immunodeficiency diseases"

Authors Jan Rohr, Karin Beutel, Andrea Maul-Pavicic, Thomas Vraetz, Jens Thiel, Klaus Warnatz, Ilka Bondzio, Ute Gross-Wieltsch, Michael Schündeln, Barbara Schütz, Wilhelm Woessmann, Andreas H. Groll, Brigitte Strahm, Julia Pagel, Carsten Speckmann, Gritta Janka, Gilian Griffiths, Klaus Schwarz, Udo zur Stadt, and Stephan Ehl

Information about the contributions of each person named as having participated in the study

1) Guarantor(s), i.e., person(s) who is (are) responsible for the integrity of the work as a whole:
• Ehl. Stephan, *Centre of Chronic Immunodeficiency (CCI), University Medical Centre, Freiburg, Germany, stephan.ehl@uniklinik-freiburg.de*

According to the International Committee of Medical Journal Editors (ICMJE) (http://www.icmje.org/ethical_1author.html): "Authorship credit should be based on: 1) substantial contributions to conception and design, acquisition of data, or analysis and interpretation of data; 2) drafting the article or revising it critically for important intellectual content; and 3) final approval of the version to be published. Authors should meet conditions 1, 2, and 3 Acquisition of funding, collection of data, or general supervision of the research group alone does not constitute authorship".

The guarantors of this manuscript confirm that all persons designated as authors qualify for authorship, and that each author has participated sufficiently in the work to take public responsibility for appropriate portions of the content.

2) Authors who participated in the **conception of the study**: Stephan Ehl, Jan Rohr

3) **Design & Methods**. The following authors were responsible for specific investigations (please detail):

- Jan Rohr, Andrea Maul-Pavicic, Ilka Bondzio and Stephan Ehl were responsible for the functional immunological investigations as well as the flow cytometric immunophenotyping.
- Gilian Griffiths was responsible for the protein expression analysis of Munc13-4.
- Klaus Schwarz, Julia Pagel and Udo zur Stadt were responsible for the genetic analyses.
- Thomas Vraetz, Jens Thiel, Klaus Warnatz, Ute Gross-Wieltsch, Michael Schuendeln, Barbara Schuetz, Wilhelm Woessmann, Andreas H. Groll, Brigitte Strahm, Carsten Speckmann, Julia Pagel, Gritta Janka and Karin Beutel were responsible for investigating the clinical courses of the patients.

4) **Results**. The following authors were responsible for specific portions of the results, including figures and tables (please indicate the person responsible for each figure and each table):

- Jan Rohr, Andrea Maul-Pavicic and Stephan Ehl were responsible for the figures 1-4.
- Jan Rohr, Karin Beutel, Andrea Maul-Pavicic, Thomas Vraetz, Jens Thiel, Klaus Warnatz, Ilka Bondzio, Ute Gross-Wieltsch, Michael Schuendeln, Barbara Schuetz, Wilhelm Woessmann, Andreas H. Groll, Brigitte Strahm, Julia Pagel, Carsten Speckmann, Gritta Janka, Gilian Griffiths, Klaus Schwarz, Udo zur Stadt and Stephan Ehl were responsible for table 1.
- Gilian Griffiths was responsible for supplemental figure 1.

5) **Writing the manuscript**. The following authors were responsible for writing the manuscript:

- Jan Rohr and Stephan Ehl were responsible for writing the manuscript.

6) Contributors Listed in Acknowledgments: (Examples of those who might be acknowledged include a person who provided purely technical help, writing assistance, or a department chair who provided only general support. Corresponding authors must declare whether they had assistance with the study design, data collection, data analysis, or manuscript preparation. If such assistance was available, the authors must disclose the identity of the individuals who provided this assistance and the entity that supported it. Financial and material support should also be acknowledged.)

We are grateful to the CCI Advanced Diagnostic team and to Sam Grieve for excellent technical assistance.