

**Manuscript no. HAEMATOL/2012/068791 entitled “Combined immunodeficiency with life-threatening EBV-associated lymphoproliferative disorder in patients lacking functional CD27 “**

**Authors: Elisabeth Salzer, Svenja Daschkey, Sharon Choo, Michael Gombert, Elisangela Santos-Valente, Sebastian Ginzel, Martina Schwendinger, Oskar A. Haas, Gerhard Fritsch, Winfried F. Pickl, Elisabeth Förster-Waldl, Arndt Borkhardt, Kaan Boztug, Kirsten Bienemann, Markus G. Seidel**

**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:**

- Arndt Borkhardt, M.D., Pediatric Oncology, Hematology and Clinical Immunology, Medical Faculty, University Düsseldorf, Moorenstr. 5, 40225 Düsseldorf, Germany, Tel.: 0049 211 8117680, Fax 0049 211 8117607, mail: arndt.borkhardt@med.uni-duesseldorf.de;
- Kirsten Bienemann, M.D., Pediatric Oncology, Hematology and Clinical Immunology, Medical Faculty, University Düsseldorf, Moorenstr. 5, 40225 Düsseldorf, Germany, Tel.: 0049 211 8117680, Fax 0049 211 8117607, mail: Kirsten.Bienemann@med.uni-duesseldorf.de;
- Kaan Boztug, M.D., CeMM Research Center for Molecular Medicine of the Austrian Academy of Sciences, Lazarettgasse 14 AKH BT 25.3, Tel. 0043 1 40160 70069, Fax 0043 1 40160 970 000, mail: kboztug@cemm.oeaw.ac.at ;
- Markus G. Seidel, M.D., Pediatric Hematology-Oncology, Medical University Graz, Auenbruggerpl. 34/2, A-8036 Graz, Austria, Tel. 0043 316 38580215, Fax 0043 316 38513450, mail: markus.seidel@medunigraz.at

According to the International Committee of Medical Journal Editors (ICMJE) ([http://www.icmje.org/ethical\\_1author.html](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:**

Arndt Borkhardt, Kaan Boztug, Kirsten Bienemann, Markus G. Seidel

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

- SC, EFW, KBi, MGS, and AB cared for the patients, had clinical, therapeutic, and/or diagnostic responsibilities
- MGS arranged and ES, ESV, MS and KBo performed homozygosity mapping, whole exome sequencing and bioinformatics analyses in family A
- SD, KBi, and MG performed exome sequencing and bioinformatic analyses in families B & C
- SC and MGS arranged and ESV, SG, OAH, GF, WFP, SD, KBi performed immunophenotypical, functional, and immunogenetic analyses

**4) Results. The following authors were responsible for specific portions of the results, including figures and tables:**

- MGS, SC, and KBi prepared the table
- ES and KBo prepared the figures 1, S4;
- ES, SD, KBi, and KBo prepared figures S1, S2, S3
- see also “3) *Design and Methods*” for specific portions of the experiments/results
- MGS, KBi, AB, and KBo coordinated the multicentre cooperation to finalize comprehensive data analyses and manuscript preparation

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

- MGS, KBo, ES, KBi, SC, and AB prepared the manuscript
- SC edited the manuscript

**6) Contributors Listed in Acknowledgments:** The authors thank the patient families and the staff of the immunology outpatient clinics and hematology-oncology wards for their cooperation, Drs. H. Gadner, A. Heitger, and G. Mann (Vienna) for their support of experimental analyses, clinical advice, and insightful comments, Dr. S. Ehl and his team (Freiburg) for critical review of the manuscript and HLH diagnostics including NK cytotoxicity assays, and Dr. W. Schwinger (Graz) for TCR Vbeta spectratyping. The authors from Vienna thank Drs. M. Eibl and H. Wolf for referral of family-A and constructive discussions. Funding of parts of this work was provided by the Deutsche Forschungs-gemeinschaft (to KBi) and the START Programme of the Austrian Science Fund (FWF): [Y595B13] (to KBo).