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Discovery of a new bone marrow disease

Care-for-Rare Foundation
For children with rare diseases

PRESS RELEASE

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Concerted international collaboration between scientists from USA, Israel and Germany leads to the discovery of a new bone marrow disease

Munich, 2013/06/07. Children with rare diseases, also known as orphans of medicine, are disadvantaged in many ways. Long odysseys from doctor to doctor and ill-founded hope for a cure define the lives of affected families. However, children with rare diseases can escape from this shadow and contribute to the advancement of modern medicine. The scientific examination of children with rare diseases can give insights into fundamental principles of biology.

An international group of researchers jointly led by Christoph Klein, director of Dr. von Hauner University Children's Hospital, William Gahl, NIH, Bethesda, USA, and Raz Somech, Children's Hospital Sheba Medical Center, Tel Aviv, Israel, was able to identify a novel disease. The affected children suffered from life-threatening infections during the first years of their young lives due to an unusual bone marrow malfunction. Mutations in a gene named VPS45 (Vacuolar Protein Sorting 45) are the causes of this illness.

The authors of a paper published in today's issue of the renowned New England Journal of Medicine describe that the gene VPS45 has several functions in blood cells – it is responsible for membrane transport, for the vitality of the cells and their migration characteristics. On the one hand, the identification of this disease and the discovery of its molecular mechanism creates a justified hope for affected patients, as young patients can now receive blood stem cell transplantation earlier. On the other hand, this research illustrates the importance of new signalling pathways that can possibly influence the therapy of the future – not only for children with rare diseases, but also for younger and older patients with common diseases.

Further Information:

Beatriz Parente Matschke
Project Manager
Lindwurmstr. 4
80337 München
Tel: +49 89 5160-79 81
Fax:+49 89 5160-77 02
E-Mail: beatriz.parente@med.uni-muenchen.de

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