Children with rare diseases – from therapeutic orphans to pioneers of personalized medicine

Christoph Klein – Bad Gastein
October 2nd 2014
Nosological spectrum of childhood diseases

Early mortality

Sulin 8 years –

Idiopathic liver failure
IL21-Receptor Deficiency

Exome sequencing:

- 25,877 variants
- 25,985 variants
- 25,557 variants

Neutrophil granulocytes - defective maturation and structural defects
JAGN1 interacts with COPI proteins

Boztug K et al. Nat Gen 2014
Wiskott Aldrich Syndrom

Recurrent life-threatening infections

Autoimmunity

Eczema

Bleeding

Leukemia/Lymphoma

Life expectancy <20 years
Clinical gene therapy protocol for WAS

Purification of HSC

Transduction using GALV-retroviral vectors

Busulfan 8 mg/kg
Eczema

Pre GT

Day 157 post GT

Day 283 post GT

Braun C et al. Sci Transl Med 2014
Clinical Medicine
Clinical Medicine

Protecting time for creativity
12 German Networks on Rare Diseases

- **Hannover**: CARPuD
- **Münster**: TranSaRNet, AID-Net
- **Duisburg-Essen**: Imprinting diseases
- **Heidelberg**: GERAMY
- **Tübingen**: HOPE, IonNeurONet
- **Freiburg**: FACE
- **Ulm**: MND.NET
- **München**: mitoNet, PID-NET, GALENUS
Requests for Policy Makers

Re-think medicine and re-define its structural environment

Provide protected time for creativity and innovation

Create better „option spaces“ for interdisciplinary and global investigations aiming to understand diseases and to develop novel and better therapies