The Vienna Center for Rare and Undiagnosed Diseases (CeRUD)/Ludwig Boltzmann Institute for Rare and Undiagnosed Diseases (LBI-RUD)
(Vienna, Austria)

Brief description of the "Clinical Center" or "Services on undiagnosed rare diseases"
CeRUD/LBI-RUD, directed by Prof. Kaan Boztug, is Austria's first and leading clinical and research institute dedicated to rare and undiagnosed diseases research. CeRUD/LBI-RUD with its clinical partners established well-studied patient cohorts of rare and ultra-rare diseases receiving around 250 cases/year. It has built an effective infrastructure for generating, processing, interpreting, and securely sharing genome/exome/panel sequencing data. LBI-RUD scientists have a strong track record in the functional dissection of rare disease mechanisms especially in the area of hematopoietic, immune, and neurological disorders. LBI-RUD developed and applied advanced multi-omics technology including epigenome mapping, proteomics, metabolomics, and single-cell sequencing and also leads early drug development projects.

Past (six months) and future/upcoming events related to undiagnosed rare disease. Please indicate date and venue.
Rare Diseases and Big Data – Hopes, Opportunities and Challenges, 22nd – 23rd February 2018, Josephinum, Collections of the Medical University of Vienna, Austria

Relevant scientific publications (selected and / or published by the center)
Van Rijn JM et al. (2018). Intestinal failure and aberrant lipid metabolism in patients with DGAT1 deficiency. Gastroenterology, doi: 10.1053/j.gastro.2018.03.040.
Pfajfer L et al. (2017). WIP deficiency severely affects human lymphocyte architecture during migration and synapse assembly. Blood 130(17):1949-53.
Ozen A et al. (2017). CD55 Deficiency, Early-Onset Protein-Losing Enteropathy, and Thrombosis. N Engl J Med 377(1):52-61.
Müller H et al. (2017). VCF.Filter: interactive prioritization of disease-linked genetic variants from sequencing data. Nucleic Acids Res 45(W1):W567-72.
Sheffield NC et al. DNA methylation heterogeneity defines a disease spectrum in Ewing sarcoma. Nat Med 23(3):386-95.
Salzer E et al. (2016). RASGRP1 deficiency causes immunodeficiency with impaired cytoskeletal dynamics responsive to lenalidomide. Nat Immunol 17(12):1352-60.
Farlik M et al. (2017). DNA methylation dynamics of human hematopoietic stem cell differentiation. Cell Stem Cell 19(6):808-22.

Grants. a) grant award to announce funding for projects; b) Received grants from other organizations/institutions for undiagnosed rare diseases research.
- LBI-RUD core funding, Ludwig Boltzmann Gesellschaft GmbH, CeMM, MedUni Vienna, CCRI EUR 8,300,000 04/2016 – 03/2023
- Systems precision medicine of inborn errors of the immune system (PrecisePID) Vienna Science and Technology Fund (WWTF), EUR 418,800, 03/2017 – 02/2020
- Novel immunodeficiency unravels immune homeostasis mechanism, Austrian Science Fund (FWF), EUR 351,272, 05/2017 – 04/2020
- New insights into DNA repair disorders: integrating genomics and functional studies for developing diagnostic and therapeutic approaches, Jeffrey Modell Foundation, US$ 62,500, 09/2016 – 08/2018 - High-resolution imaging to unravel the molecular etiology of disturbed T-cell antigen recognition, Vienna Science and Technology Fund (WWTF), EUR 291,040, 03/2015 – 02/2018
- Inborn errors of innate immunity: systems genomics route to the core of the immune system (ImmunoCore), European Research Council (ERC), EUR 1,498,950; 11/2013 – 10/2018

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