

Dear Colleagues,

It is our pleasure to welcome you to the 4th Freiburg Pediatric Research Day. The conference will highlight and discuss interesting aspects of research in pediatrics. We have put together an exciting program and hope for a productive interdisciplinary exchange between researchers and for new promising scientific projects.

During the coffee and lunch breaks, research group leaders would be glad to talk to anyone interested in joining or working with their group. Feel free to approach them! Several research group leaders have put together short profiles of their work, which you can download here: <https://www.research-for-children.de/event/zkj-research-day>

We wish you all an exciting conference and fruitful discussions!

Yours,

The Research Day Organizing Committee

*Alexej Bobrowski
Roland Elling
Christian Flotho
Anneke Haddad
Anna Lena Klauser*

*Thorsten Langer
Florens Lohrmann
Maria Elena Maccari
Anke Schumann*

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Registration

<https://www.uniklinik-freiburg.de/kinderklinik/forschung-und-klinische-studien/anmeldung-zkj-forschungstag-2022.html>

or via Sandra Schickinger
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Venue:



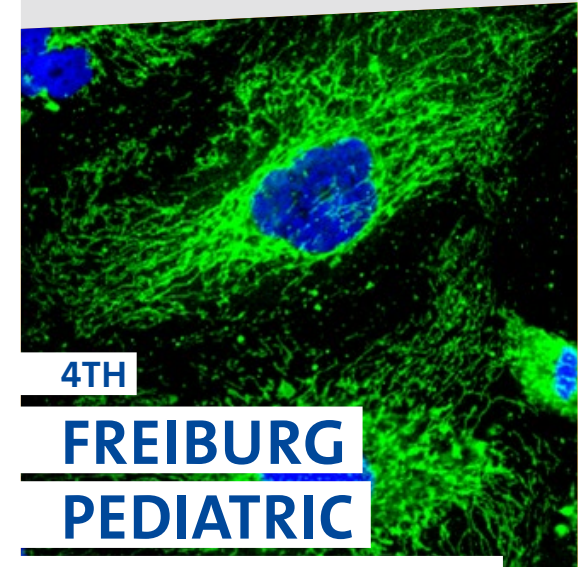
ETAGE Tagungszentrum

Solar Info Center Freiburg, 1. OG
Emmy-Noether-Straße 2
79110 Freiburg im Breisgau

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PROGRAM



4TH

FREIBURG PEDIATRIC RESEARCH DAY

29TH NOVEMBER 2022
08:45 - 17:30 Uhr

Für diese Veranstaltung wurden
8 Fortbildungspunkte bei der LÄK
Baden-Württemberg beantragt.

Program

8:45 - 9:00	Welcome
9:00 - 10:20	SESSION 1: Genetic and molecular aspects of pediatric diseases (Chair: Alexej Bobrowski) <i>Christian Flotho</i> Role of the NF1 tumor suppressor gene in juvenile myelomonocytic leukemia: Genetic evidence of driver function and implications for diagnostic work-up <i>Vera Wimmers</i> Implication of FOXD2 loss of function in syndromic congenital anomalies of the kidney and urinary tract (CAKUT) <i>Anna Wolfers</i> Pathogen control versus hyperinflammation: A STAT1 variant beyond the gain- or loss-of function dualism <i>Vasil Toskov</i> A novel interferonopathy with B cell deficiency associated with PRIM1 primordial dwarfism syndrome <i>Nagi Mahammadzade</i> Using the zebrafish model to characterize novel RIT1 variants in arteriovenous malformations. <i>Katharina Neubauer</i> Impaired platelet function in Sept8-deficient mice
10:20 - 10:50	Coffee break + Odd-numbered posters

10:50 - 12:00	SESSION 2: Session 2: Health, brain and development in children (Chair: Thorsten Langer) <i>Alexandra Klotz</i> Hypothalamic Hamartoma as a model of subcortically generated seizures <i>Johanna Umlauf</i> Valproate-associated cerebral atrophy in children with epilepsy <i>Jan Schönberger</i> How epilepsy disrupts psychomotor development: Are structure-specific sleep mechanisms the missing link? <i>Anneke Haddad</i> Long COVID symptoms in exposed and infected children, adolescents and their parents one year after SARS-CoV-2 infection: A prospective observational cohort study <i>Olimpia Alice Manzardo</i> Human milk pasteurisation: Investigating high-temperature short-time processing as a novel heat treatment protocol
12:00 - 12:45	KEYNOTE LECTURE: Pediatric Neuro-oncology 2.0 – How patients benefit from state-of-the-art molecular diagnostics <i>Keynote Speaker: Prof. Dr. Stefan Pfister, Heidelberg</i>
12:45 - 13:15	Lunch break
13:15 - 13:45	Even-numbered posters
13:45 - 14:15	Odd-numbered posters
14:15 - 15:30	Career paths of a physician scientist (Chair: Anna-Lena Klauser + Stephan Ehl) <i>Anne Geweniger</i> <i>Friedrich Kapp</i> <i>Philipp Henneke</i>

15:30 - 16:00	Coffee break + Even-numbered posters
16:00 - 17:20	SESSION 3: From cellular function to disease pathogenesis (Chair: Roland Elling) <i>Anke Schumann</i> Towards deciphering kidney-disease driving mechanisms in organic acidurias <i>Sheila Bohler</i> RIOK1-a new gene associated with Diamond Blackfan anemia <i>Jovana Rajak</i> Understanding the crosstalk between juvenile myelomonocytic leukemia (JMML) and immune cells <i>Lennart Moritz</i> Targeted metabolomics in phenylketonuria highlights disease clustering even in patients with good metabolic control <i>Hannah Kappler</i> Disease severity and fibrosis are associated with distinct action potential shape in right ventricular myocardium of patients with congenital heart defects <i>Pia Kröning</i> Does [18 F] Fluorodeoxyglucose Positron Emission Tomography (FDGPET)/ CT detect bone marrow involvement in children and adolescents with rhabdomyosarcoma?
17:20 - 17:30	Awards and closing remarks