e:Medium Flash NEWSLETTER

May 2025

Dear Reader,

In the May edition of e:Medium Flash, we present new highlights from the e:Med research network. Discover the latest publications and insights that push the boundaries of systems medicine. Our updated format continues to support visibility, exchange, and collaboration across the e:Med community. Please feel free to draw our attention to your new publications via e:Med website.

Enjoy reading!

Your e:Med Management Office



BREAKTHROUGH IN CANAVAN DISEASE DIAGNOSIS

Scientists from the e:Med junior research alliance PeriNAA have identified a previously undetected genetic variant in five Canavan disease patients who lacked a genetic diagnosis. Dr. Annette Bley and colleagues discovered the variant using targeted long-read sequencing, revealing a retrotransposable element in the ASPA gene that standard diagnostic procedures had missed. This discovery enhances genetic diagnostic precision for this rare autosomal recessive leukodystrophy and offers<u>a</u> potential intervention strategy for affected families. **read more**





PROTECTIVE ROLE OF AFAMIN IN KIDNEY DISEASE

New results from e:Med junior research alliance CKDNapp headed by Prof. Helena Zacharias and GCKD scientists suggest that increased serum afamin concentration is linked with better kidney function. In a cohort of 5000 patients with chronic kidney disease, Dr. Ulla Schultheiss and her colleagues show that higher afamin levels are linked to higher eGFR, less albuminuria and therefore better filtration. Their results reveal that the vitamin E-binding glycoprotein afamin can serve as a biomarker for kidney health and disease progression. **read more**







Federal Ministry of Research, Technology and Space



Digger 2.0 UNEARTHS IMPACT OF ALTERNATIVE SPLICING

Current data analysis methods neglect how alternative splicing affects protein structure and consequently larger interaction networks. Dr. Olga Tsoy and colleagues from eMed alliance Sys_CARE led by Prof. Jan Baumbach integrated protein-protein interactions with domain-level data, supporting both human and mouse studies. The addition of NEASE splicing analysis enables comprehensive RNA-seq enrichment analysis online. Demonstrated through multiple sclerosis and cardiac disease studies, DIGGER 2.0 reveals hidden functional impacts of splicing changes.

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UNLOCKING THE PSYCHOLOGY OF **FEAR**

This review from e:Med demonstrator Target-OXY (head Prof. Rainer Spanagel) explains fear conditioning as a method to understand learning, memory, and extinction. Prof. Christian P. Müller and colleagues discuss the molecular mechanisms underlying fear processing, including the amygdala and hippocampus, and evaluate the limitations and advancements in fear conditioning protocols. The findings from rodent studies have the potential to provide therapeutic strategies for fear-related pathologies in humans such as anxiety and PTSD and psychiatric disorders such as schizophrenia. read more



A NEW PROGNOSTIC MARKER FOR CHILDHOOD CANCER?

Latest findings by e:Med junior research alliance MelBrainSys, headed by Dr. Michael Seifert and Dr. Dana Westphal, reveal tertiary lymphoid structures (TLS) to serve as a promising prognostic marker in childhood peripheral neuroblastic tumors. Based on experiences with melanoma brain metastasis, Dr. Rebekka Wehner and colleagues used advanced techniques and identified TLS in nearly half of patients examined, with their presence significantly correlating with prolonged progression-free survival regardless of other immunological features. Therefore, TLS could serve as a valuable prognostic marker for predicting outcomes in children with these challenging tumors. read more



ONLINE SEMINAR SERIES Modelling approaches for disease processes

Tue June 3, 2025, 4pm Simplifying inflammation and fibrosis Prof. Dr. Uri Alon, Weizman Institute, Israel

Location: Zoom, 4 p.m. CEST read more

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