

Introductory Remarks to Symposium 28

Translational science in pediatric neurology – what we can learn!

Florian Heinen, Munich

In pediatric neurology there is a growing knowledge about molecular mechanisms and pathophysiology in known diseases leading to new therapeutic options. Additionally, in the last years new phenotypes or diseases could be identified coming from clinical characterization and identification of new genes. Using this knowledge more individually addressed therapies are and will be developed so that in patients with neuropediatric, even lethal disorders life expectancy and quality of life dramatically improved and will be improved. Following aspects will be addressed in this symposium:

Katharina Vill, (AG Vill, LMU, Munich) will describe the journey for the autosomal recessive spinal muscular atrophy from new molecular findings to gene therapy leading to a change in treatment and an implementation in the newborn screening program, which causes a completely new situation for these patients and families: "Molecular neuroscience in neuropaediatrics - from Screening to intervention". Julia Wallmeier (AG Omas, Uniklinik, Münster) will present new genetic aspects in hydrocephalus, a symptom often caused by other different underlying disorders, but new are the genetic aspects, especially for the diagnostic-work-up and treatment in these patient group: Genetic mechanisms involved in hydrocephalus. Markus Schülke (Charité Berlin) will present a translational project, in which a pharmacological therapy was developed using iPSC-derived neuronal precursor cells of a patient with Leigh syndrome to identify a compound, which was already on the market for another purpose. Repurposing of EMA-approved drugs for rare neurogenetic disorders will greatly accelerate the journey from molecular diagnosis to custom tailored therapies. Niccolò Milani (AG Bader, MDC, Berlin) will present the project (EU Horizon 2020) with the aim to evaluate the contribution of extraembryonic sources of serotonin – via placenta – for embryonic brain development.

Symposium 28

Friday, March 24, 2023
08:30 - 10:30, Lecture Hall 103

Chair: Florian Heinen, Munich

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| 08:30 | Opening Remarks |
| 08:35 | Katharina Vill, Munich
MOLECULAR NEUROSCIENCE IN NEUROPAEDIATRICS - FROM INTERVENTION TO SCREENING (S28-1) |
| 09:00 | Julia Wallmeier, Münster
GENETIC MECHANISMS INVOLVED IN HYDROCEPHALUS FORMATION (S28-2) |
| 09:25 | Markus Schülke, Berlin
REPURPOSING FDA APPROVED DRUGS FOR MATERNALLY INHERITED LEIGH SYNDROME USING HUMAN IPSC DERIVED DISEASE MODELS (S28-3) |
| 09:50 | Niccolò Milani, Berlin
EXTRAEMBRYONIC SOURCE OF SEROTONIN INVOLVED IN NEURODEVELOPMENT (S28-4) |
| 10:05 | Discussion / Concluding Remarks |