The Inozyme Advocate

Your source for news, information, and events on Rare Calcification Disorders

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Welcome to The Inozyme Advocate, a semi-annual communication to bring you news, information, and events from the Patient Advocacy Team at Inozyme!

ENPP1 Deficiency Mini Symposium at the 2019 European Calcified Tissue Society Annual Meeting (ECTS)

Inozyme sponsored an hour long CME symposium on ENPP1 Deficiency at the 2019 European Calcified Tissue Society (ECTS) Annual Meeting. The meeting took place in beautiful Budapest, Hungary from May 11-14. ECTS is an annual meeting for researchers and clinicians in musculoskeletal field and this year it hosted 1,178 delegates from 56 countries. Distinguished lecturers, Prof. Dr. Stefan Schulte-Merker from the University of Muenster, Prof. Dr. Frank Rutsch from the University of Muenster, Prof. Dr. Ralf Oheim from the University Medical Center Hamburg Eppendorf and Dr. Demetrios Braddock from the Yale School of Medicine delivered a series of talks detailing current research efforts to understand the underlying science behind ENPP1 Deficiency and its effects in animal models such as zerbrafish and mice, the consequences of ENNP1 Deficiency and low pyrophosphate (PPi) in human patients and the potential for treatments on the horizon. The mini symposium was moderated by Prof. Dr. Uwe Kornack from the Institute of Medical Genetics and Human Genetics in Berlin and Dr. Leonor Cancela from the University of Algarve. There were significant interactions with the audience showing the growing interest in ENPP1 Deficiency and how new research is helping to create better understanding of this ultra-rare disease.

Pictured left to right: Dr. Ralf Oheim, Dr. Demetrios Braddock, Dr. Stefan Schule-Merker, Dr. Frank Rutsch, Dr. Leonor Cancela and Dr. Uwe Kornack

Pictured left to right: Henric Bjarke, Ruhi Ahmed, Supriya Rao, and Nathan Cullen. Inozyme is now in Twitter!
Like us Follow us @inozymepharma

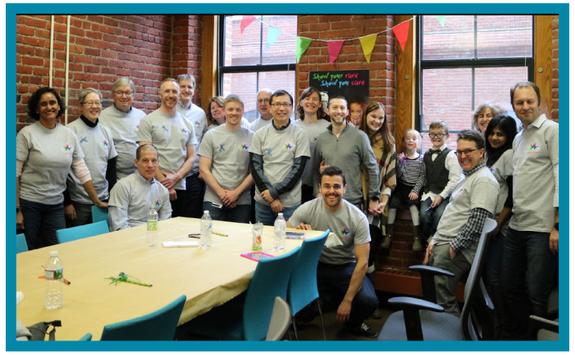
Meet our Team - Prof. Dr. Frank Rutsch, MD

Prof. Dr. Frank Rutsch, MD, is an Associate Professor in Pediatrics at Münster University Children's Hospital, Münster, Germany and a member of Inozyme's Clinical Advisory Board. Dr. Rutsch's main research interests are focused on the discovery of the underlying genetic defects and translational aspects in rare pediatric metabolic and autoimmune disorders. Dr. Rutsch has not only treated patients with Generalized Arterial Calcification of Infancy (GACI) but has been a pioneer in understanding the science underlying ENPP1 Deficiency. He was the lead author of the study which established the link between mutations in the *ENPP1* gene and GACI in 2003 and his group performed the first natural history study of patients with GACI in 2008.

Dr. Rutsch continues to study the underlying disease mechanism in ENPP1 Deficiency and is working with Inozyme by advising the company scientifically and clinically on its mission to bring an enzyme replacement therapy (ERT) for this severe and debilitating disease. According to Dr. Rutsch, "ENPP1 ERT has the potential to address the root deficiency in patients with GACI and could address aspects of the disease, such as arterial stenoses, which are not addressed by current treatments"

Rare Disease Day 2019

Rare Disease Day is observed annually worldwide on the last day of February to raise awareness for rare diseases and what the daily impact of living with a rare disease has on patients and families. This year Inozyme was proud to host Christine O'Brien-Lemanski from GACI Global and her two children, Callum and Nora.



The O'Brien-Lemanski family shared their experience raising two children affected by ENPP1 Deficiency. Over an afternoon of pizza and cake, Inozyme shared their efforts toward helping these patients and their families and Inozyme's goal of developing a safe and effective treatment for this severe and debilitating disease.

RARE DISEASE DAY®

GACI Global

We would like to congratulate GACI Global on the launch of their new website! https://gaciglobal.org

GACI Global
circulating hope

Anyone who has been affected by a rare or ultra-rare disease knows how important it is to find people who have experienced the same situation. The ability to share information and give suggestions on what questions to ask is very important. We are very happy that GACI Global is there for patients affected by ENPP1 Deficiency (GACI & ARHR2).

GACI Global is a nonprofit organization whose mission is to connect families affected by Generalized Arterial Calcification of Infancy or Hypophosphatemic Rickets caused by *ENPP1* or *ABCC6* Deficiencies to each other and to the medical community.

Upcoming Events - 2019

The following meetings maybe of interest to patient, caregivers and HCPs. Visit their websites to find the ones of interest to you.

- **September 3-6:** Society for the Study of Inborn Errors of Metabolism. Rotterdam Netherlands https://ssiem2019.org/
- **September 5:** Rare Disease Scientific Workshop, Washington, DC. https://everylifefoundation.org/scientific-workshop-sponsorship/11th-annual-scientific-workshop/
- **September 18-20:** Global Genes 201 Rare Patient Advocacy Summit https://globalgenes.org/event/patient-summit/
- **September 19-21:** European Society for Paediatric Endocrinology, Vienna, Austria https://www.eurospe.org/meetings/2019/espe2019/
- **September 20-23:** American Society for Bone and Mineral Research, Orlando, FL https://www.asbmr.org/
- **October 21-22:** Rare Diseases & Orphan Products Breakthrough Summit, Washington, D.C. https://rarediseases.org/summit-overview/
- **November 12-14:** World Orphan Drug Congress Europe, Barcelona, Spain https://www.terrapinn.com/conference/world-orphan-drug-congress/index.stm

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