

## INSIGHTS



### OBITUARY

# Claude Bachmann, MD (1941–2022)

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*Pediatric Research* would like to honor Claude Bachmann with this obituary. Claude Bachmann served as European Editor-in-Chief of *Pediatric Research* from 1981 to 1985. Dr. Bachmann's focus was on translational research, especially in the area of biochemical genetics. Claude entered the field of biochemical metabolism during his internship at the University Children's Hospital in Basel, studying tryptophan metabolites in patients with Vitamin B6-dependent seizures. A patient with non-ketotic hyperglycinemia led to his first publication on the potential use of strychnine therapy in this disorder and motivated him to move his family to the University of California, San Diego to become a fellow of Bill Nyhan, with whom he developed a close relationship that he maintained throughout his life. In San Diego, he learned gas chromatography–mass spectrometry techniques for measuring organic acids and identified 3-hydroxyisovalerate in a patient who was later shown to have biotinidase deficiency. He also became interested in urea cycle disorders and developed an high-performance liquid chromatography method to measure orotic acid. He wrote one of the first reviews of these disorders in Nyhan's *Heritable Disorders of Amino Acid Metabolism* (1974). In the area of propionic and methylmalonic acidemia, he formulated the hypothesis of *N*-acetylglutamate synthetase (NAGS) inhibition as the cause of hyperammonemia in these conditions and described the first case of NAGS deficiency in 1981.



After returning to Switzerland and spending an additional 2 years at the University Children's Hospital in Basel completing his residency in pediatrics, Claude became the Vice-Director and Section Head of Special Analyses in the Central Clinical Chemistry Laboratory of Inselspital in Berne. He remained in Berne for 13 years developing novel methodologies and

engaging in international collaborations in the areas of urea cycle disorders and organic acidemias.

In 1988, he was appointed Professor and Director of the Central Clinical Chemistry Laboratory at the University Hospital of Lausanne, where he continued his research interests in urea cycle disorders and developed a specialized diagnostic laboratory for inherited metabolic diseases. He was director for almost 20 years, until his Swiss mandated retirement at age 65 years in 2006.

Claude was also a key player in clinical chemistry. His leadership manifested in particular in his role within the *Swiss Society of Clinical Chemistry* (of which he successively directed several working groups, the scientific commission, and the steering committee), his involvement in the implementation of a post-graduate training program in laboratory medicine (FAMH), and his contribution to the scientific development of this specialty.

Among his many honors and international positions held were his being European Editor-in-Chief of *Pediatric Research*, a corresponding editor for the *Journal of Inherited Metabolic Disease*, and Editor-in-Chief of *Enzyme and Protein*. During his eminent career, Claude published over 300 articles and book chapters.

In writing this obituary, Claude's friends, family, and colleagues noted some of his unique qualities: his great intellectual curiosity and honesty, his capacity for thinking outside the box, and his passion for culture. He was witty but could also be acerbic and was constantly challenging colleagues around scientific hypotheses and data. He did so with a smile and a puff from his ever-present pipe. Claude was always happy to talk to colleagues from all countries of the world, and indeed his modesty attracted young people who found an attentive and empathic listener. He was a great mentor, always focused on the "maturation" of his students and junior faculty. He instilled in them his enthusiasm and passion for scientific discovery and was always available and interested in dialogue, discussion, and exchange of ideas. His contributions to the field of biochemical metabolism and inherited metabolic disorders were many and major.

After retirement, Claude and his wife spent much of their time with their daughter and grandchildren in Greece and indulged his interests in classical music (playing the piano), art, literature, history, politics, gardening, and visiting the zoo. Four years ago, Claude was diagnosed with Alzheimer's disease, and he was cared for by his loving wife of >50 years, Anne. His daughter, Isabelle, noted: "Though it was hard to see him slowly fade away, we will keep in

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memory his brilliant, witty, humorous and deeply humanistic mind and personality." Claude will be remembered not only as a brilliant scientist but also as a kind and generous colleague and friend.

Contributed by Mark L. Batshaw, Olivier Braissant, Carlo Dionisi Vici, Beat Steinmann, and Andrea Superti-Furga.

#### COMPETING INTERESTS

The author declares no competing interests.

#### ADDITIONAL INFORMATION

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