



Dr. Patricia K. Duffner

Professor of Neurology

Clinical Center, HJKRI

Dr. Patricia K. Duffner established the Clinical Center of the HJKRI in 2006 and served as Clinical Director of the Institute until January 2012. She is currently a consultant to the Institute.

Dr. Duffner received her undergraduate degree from the University of Rochester in 1968, her medical degree from the University at Buffalo in 1972 and completed her residency in Pediatrics and Neurology at the Children's Hospital of Buffalo/University at Buffalo School of Medicine program in 1978.

She joined the Division of Child Neurology at the Children's Hospital after completing her residency. She was appointed Professor of Neurology and Pediatrics in 1989 and served for 18 months (2000-2001) as interim chair of the Department of Neurology.

Her primary research interests for most of her career were pediatric brain tumors, particularly those occurring in babies and very young children and the long-term effects of CNS therapy on children with brain tumors and leukemia.

In 1997 she diagnosed four-month-old Hunter Kelly with Krabbe Disease, leading eventually to a shift in her research focus. She developed a lasting relationship with the Kelly family and their Foundation, Hunter's Hope, serving as chair of the Scientific Advisory Committee for many years.

In 2006, with the advent of newborn screening for Krabbe Disease in NYS, she focused her efforts on maximizing the effectiveness of the NYS program and became Clinical Director of the Institute. She established the NYS Krabbe Consortium, the NYS Newborn Screening Clinical Database/Registry and the World Wide Registry for Krabbe Disease.

Dr. Duffner has written more than 120 papers and book chapters, has co-authored two books including two editions of Brain Tumors in Children: Principles of Diagnosis and Treatment (1984, 1994) and Weiner and Levitt's Pediatric Neurology-4th edition (2003) and has lectured throughout the US, Europe and South America. She serves on two subcommittees of the National Newborn Screening Translational Network and is a member of the Lysosomal Disease Network core committee. She serves on the editorial board of Pediatric Neurology and is an ad hoc reviewer for more than 20 journals. Her work has been funded by HRSA (maternal and child health), NINDS, Children's Guild of Buffalo and Hunter's Hope Foundation.