

OBITUARY

Obituary Norio Niikawa, M.D., Ph.D., 1942–2022

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Professor Norio Niikawa, an honorary member of the Japan Society of Human Genetics, Emeritus Professor of Nagasaki University, Emeritus Professor of the Health Science University of Hokkaido, and former President of Health Sciences University of Hokkaido, sadly passed away on 4 April 2022, at the age of 79 years. On behalf of his students, I would like to express my deepest condolences, and my sincere appreciation for his tremendous academic contributions and dedication to the Japan Society of Human Genetics.

Professor Niikawa was born in Hokkaido on 8 May 1942. He graduated from Hokkaido Sapporo Nishi High School in 1961 and Hokkaido University School of Medicine in 1967, and went on to join the Department of Pediatrics at Hokkaido University. After clinical training as an intern and a pediatrician, he obtained a

research position in the Chromosome Research Unit at Hokkaido University in 1970 under Professor Sajiro Makino, the first director of the Research Unit founded in 1969. He was promoted to Assistant Professor in the Department of Obstetrics and Gynecology at the University of Geneva, Switzerland, in 1972, and performed chromosomal research under Dr. Tadashi Kajii. He returned to the Department of Pediatrics at Hokkaido University as an Assistant Professor in 1976. He then obtained a full professorship at the Department of Human Genetics, the Atomic Bomb Disease Institute, Nagasaki University School of Medicine in 1984 and remained there until his retirement in 2007. He served as the President of the Japan Society of Human Genetics from 2004 to 2007. After retiring from Nagasaki University, he was awarded the title of Emeritus Professor of Nagasaki University. He then went on to serve as the President of the Health Sciences University of Hokkaido from 2010 to 2016, and was also awarded the title of Emeritus Professor of the Health Sciences University of Hokkaido after his resignation. His awards/honors included the Prize for Science and Technology (Research Category), MEXT, in 2006 and an Academic Award from the Japan Society of Human Genetics in 2008.

He started chromosomal research at the Chromosomal Research Unit at Hokkaido University and subsequently applied chromosomal analysis technologies to medical research at the University of Geneva, with Dr. Kajii. He helped to reveal that the predominantly dispermic fertilization in triploidy; tetraploidy originated from errors in the first mitotic division of zygotes; and also contributed to the finding of the androgenic origin of hydatidiform moles by Drs. Kajii and Ohama. He was also renowned worldwide for establishing Kabuki syndrome, in parallel with Dr. Yoshikazu Kuroki in 1981, which is also accordingly referred to as Niikawa-Kuroki syndrome. Through international collaboration, Professor Niikawa also succeeded in identifying pathogenic *KMT2D* variants in Kabuki syndrome in 2010. After moving to Nagasaki University in 1984, he focused on the application of molecular genetic technologies to medical genetics. By incorporating molecular techniques, he confirmed the maternal origin of submicroscopic deletions in Angelman syndrome using DNA markers, subsequently leading to the establishment of imprinting diseases such as Prader-Willi syndrome and Angelman syndrome. He also developed chromosomal microdissection technology, which can be useful for isolating DNA microclones and developing FISH painting probes. Furthermore, he found disease-causing gene variations in various Mendelian diseases, including Waardenburg syndrome type I (*PAX3*) in 1994, Sotos syndrome (*NSD1*) in 2002, Marfan syndrome type II (*TGFBR2*) in 2004 using chromosomal structural abnormalities, Camurati-

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Engelmann disease (*TGFB1*) in 2000, and earwax (*ABCC11*) in 2006, using linkage analysis of familial pedigrees.

Despite his tremendous achievements, Professor Niikawa remained fair, open, friendly, and cheerful to everyone who approached him, and inspired passion for science and research. When we asked him about possible plans for our projects, he advised us to carry them out right away, highlighting him as a 'man of action', especially in terms of research. However, he also showed broad interests in many other things: he belonged to a rowing club and choir at university, he loved reading scientific books, especially on quantum physics, listening to classical music, and even playing computer-based role-playing games. When I was a graduate student in his laboratory, we had hot long evening discussions about how to proceed with his ongoing role-playing video games, and he readily became enthusiastic about many things. I recall Professor Niikawa's laboratory including a medium-sized room that housed a secretary (Mori-san), several graduate students (including me), a TV, a large central desk, and several

students' desks, all packed into the one room. Although that might not have provided the best environment for graduate students, I recall spending some of my happiest times as a researcher in that room, studying under the guidance of Professor Niikawa. All the graduate students were aiming for the highest quality results and frequently worked well into the midnight (although this style is not recommended in the current work-style reform era!).

I first visited Professor Niikawa's laboratory in 1992, and was then lucky enough to spend time with him over the next 30 years. The Professor was a truly wise man, whose words frequently turned out to be true. His passing is a source of great sadness and personal grief, and represents a huge loss to the human genetics community worldwide.

Professor Niikawa, I hope you can now relax while smoking your favorite cigarette in heaven; may your soul rest joyfully in peace. I would like to express my heartiest condolences to Professor Niikawa's family. We put our hands together in prayer.