



## **Koichi Ichimura**

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Prof Koichi Ichimura's research focuses on molecular analysis and development of novel therapy for adult and pediatric malignant brain tumors. He holds the current position as Specially Appointed Professor at Department of Pathology, Kyorin University Faculty of Medicine since April 2024. His current research interests include genome analysis, molecular diagnosis and development of novel therapy for all types of malignant brain tumors in adults and children. His specialty in pediatric neuro-oncology is central nervous system germ cell tumors (CNS GCT). He organized a nationwide consortium for CNS GCT genome analysis (iGCT Consortium) to collect over hundreds of tumor tissue samples and patients' information and published several key papers in this field. He is in charge of providing molecular diagnosis as a part of the central diagnosis of pediatric brain tumors in Japan Children's Cancer Group (JCCG).

### **Topic: All About Genetics in CNSGCT**

Central nervous system germ cell tumors (CNSGCTs) mostly arise in children and young adults. The incidence is higher in East Asia than Europe or North America, suggesting the presence of genetic predisposition. CNSGCTs may be divided into two groups, namely germinomas and non-germinomatous germ cell tumors (NGGCTs). Through the integrated genomic and epigenomic analysis on more than a hundred tumor specimen obtained from the Intracranial Germ Cell Tumor Genome Analysis Consortium, we found that half of CNSGCTs regardless of histology harbored mutations in the MAPK/PI3K pathways, most typically in KIT. Germinomas were characterized by global hypomethylation, presumably reflecting their cell of origin, primordial germ cells (PGCs). Integrated mutational, transcriptional and methylation analysis suggested that germinoma and NGGCT may develop from a common ancestral PGC. A genome-wide association study showed that a risk allele within the enhancer of BAK1 was significantly more common among CNSGCTs than a control Japanese cohort.