

Department/Division: Department of Pediatrics

Theme of research:

Molecular studies of autism spectrum disorders; phenotype analyses, genetic variation analyses and identification of sensitive genes.

Name of main researcher, title, and e-mail address:

Mariko Y. Momoi (Professor), Takanori Yamagata (Associate Professor), Masato Mori (Assistant Professor), Naomi Nakashima (Assistant Professor), Mari Saito (Graduate Student)

Brief explanation of research activity:

Autism spectrum disorder (ASD) is a developmental disorder characterized by the affected social interaction including verbal development and stereotypic behavior. To reveal molecular pathology of ASD, we have been working on the following projects.

(1) Candidate gene analyses for the disease-sensitive mutations

Both functional and positional candidate genes have been analyzed for their disease-sensitive mutations on the patients with and without familial predisposition.

(2) Genomic microarray studies, comparative genomic hybridization (CGH) and copy number variation (CNV), are also in progress.

(3) Phenotype – genotype interaction

Phenotype – genotype interaction is analyzed with association of Project (1).

(4) Animal model research

By using knockout mice for secretin receptor, Fmr2 and other genes in which disease –sensitive mutation was detected in Project (1), several projects are in progress including behavior analysis, neuropathology and molecular studies.

Department/Division: Department of Pediatrics

Theme of research:

Analysis for the pathophysiology of mitochondrial disorders

Name of main researcher, title, and e-mail address:

Mariko Y. Momoi (Professor), Masato Mori (Assistant Professor), Takanori Yamagata (Associate Professor)

Brief explanation of research activity:

Mitochondrial disorders are multi-systemic disorders with relatively high incidence in childhood. Since the identification of 3243 mutations of MELAS in our laboratory in 1989, clinical, molecular and cytological studies have been in progress mainly being focused on the following issues:

1. Screening of mutations in mitochondrial and also in nuclear genomes.
2. Molecular studies on apoptotic processes in the diseases to search the possible strategy for the treatment.

Department/Division: Department of pediatrics

Theme of research:

The role of circulating endothelial progenitor cells (EPC) and VEGF-C and VEGF-D in the pathogenesis of pediatric hemangioma.

Name of main researcher, title, and e-mail address:

Yuji Gunji (Associate Professor, gunji@jichi.ac.jp)

Brief explanation of research activity:

We have recently reported the expression of vascular endothelial growth factor receptor (VEGFR)-2 and VEGFR-3 in vascular endothelial cells of kaposiform hemangioendothelioma patient (J Pediatr Hematol Oncol ,2009). In addition, circulating endothelial progenitor cells (EPC) have been reported to be associated with the angiogenesis and the pathogenesis of various disease including malignancy. VEGFR2 and VEGFR3 have also been reported to have pivotal roles for the function of EPC and vascular endothelial cells. So, we speculate that the EPC and VEGFRs including their ligands could be associated with the pathogenesis and progression of hemangioma.

In this context, we are now evaluating the EPC and VEGF-C and VEGF-D in addition to VEGF-A and VEGF-B in the pediatric hemangioma patients by FACS analysis and EPC detection culture system.

Department/Division: Department of Pediatrics

Theme of research:

Chromosomal genomic hybridization (CGH) and candidate gene analysis for the patients with multiple anomaly

Name of main researcher, title, and e-mail address:

Mariko Y. Momoi (Professor), Takanori Yamagata (Associate Professor), Masato Mori (Assistant Professor), Mari Saito (Graduate Student)

Brief explanation of research activity:

Chromosomal micro-rearrangements such as micro-deletion or duplication have been reported to be the cause of many diseases including mental retardation and multiple anomaly syndromes. To detect the responsible genes for developmental disorders and/or multiple anomalies including cardiac anomaly, we are analyzing the patients for micro-rearrangement by chromosomal genomic hybridization (CGH) method. And also we analyze the candidate genes for mutations by direct sequencing.