第502回難研セミナー
第75回難治疾患共同研究拠点セミナー

【日　時】 平成25年8月8日（木）13:00 ～ 15:00

【場　所】 M&Dタワー22階　難治疾患研究所セミナー室

【演　者】 Dr. Enkhsaikhan Purevjav

Research Assistant Professor, The Heart Institute, Cincinnati Children' s Hospital Medical Center, Cincinnati, OH, USA

【演　題】 GENETIC SCREENING OF CARDIOVASCULAR DISEASES: CINCINNATI CHILDREN' S EXPERIENCE

Abstract: Cardiac disorders in childhood are the devastating clinical diseases, and cardiomyopathy and heart failure are the most common diagnoses for hospital admissions and costs billions in healthcare dollars yearly. Chromosomal abnormalities, copy number variations (CNV)-deletions or duplications and single gene mutations are common causes of cardiac disorders. Over the past three decades, molecular genetic analysis of genetic diseases has made a significant impact upon the diagnosis and possible therapeutic options of cardiomyopathies. Genomic technologies used for clinical testing are rapidly improving due to implication of massively parallel or next generation sequencing. Candidate gene analysis based on a physiologic, mechanistic, or genetic understanding of the disease is the main approach of cardiovascular genetic screening. As the leader of the Clinical Core and Genetic Core, Dr. Towbin and colleagues have enrolled more than 3,000 patients in Pediatric Cardiomyopathy Registry (PCMR) and have collected the Pediatric Cardiomyopathy Specimen Repository (PCSR), a multicenter North American repository, to study the basis of pediatric cardiomyopathies. Cincinnati Children's Hospital Medical Center (CCHMC) serves as the site for sample processing and storage. Dr. Towbin's laboratory at the CCHMC has identified many novel disease-causing genes, launched the next generation sequencing of cardiac disorders in children and has over 350 publications, the majority of publications focusing on cardiomyopathy. The role of genetic testing for inherited cardiac conditions including cardiomyopathy, cardiac arrhythmia disorders and congenital heart disease is becoming important part of diagnostic and clinical evaluation that may potentially life-saving for patients and their relatives.

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