Panel Discussion

Fri. Jul 10, 2026 2:00 PM - 3:30 PM JST | Fri. Jul 10, 2026 5:00 AM - 6:30 AM UTC | Room1（Conference Hall A）

[PD6] パネルディスカッション 06（II-PD6）

小児期のブルガダ症候群およびSCN5A遺伝子バリアント関連疾患

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[II-PD6-3] Genetic backgrounds and the usefulness of genetic testing in Brugada syndrome

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Keywords：Brugada syndrome、genetic background、SCN5A

Brugada syndrome (BrS) is an inherited disease that can cause sudden cardiac arrest, mainly in middle-aged men. Although there are very few female patients with BrS in adulthood, there is no gender difference before puberty.
SCN5A, which encodes the cardiac sodium channel, is the only established causative gene for BrS, and loss-of-function (LOF) variants are known to cause BrS. However, the detection rate of pathogenic variants in adult patients with BrS is approximately 10% in Japan. In contrast, the detection rate in young patients is higher, around 50%, suggesting that genetic factors have a stronger influence on disease onset in younger individuals. LOF SCN5A variants are also associated with sinus bradycardia, conduction disturbances, and atrial arrhythmias. In genetic testing in family members of BrS patients, some SCN5A variant carriers do not show BrS phenotype but other electrophysiological abnormalities.
In Japan, most patients with BrS are diagnosed while asymptomatic in a health checkup, across all age groups. Therefore, risk stratification, especially for the decision to implant implantable cardioverter-defibrillators (ICD) to prevent sudden cardiac death, is important. Recently, we reported that patients with BrS carrying LOF SCN5A variants have a worse prognosis than those without such variants. In addition, genetic risk scores for BrS have been developed in Europe and Japan.
In this session, I will discuss the current status of genetic testing for BrS and consider how genetic information should be utilized in the management of young patients with BrS.