In 1990, Dr. Chin published the first paper describing left ventricular noncompaction (LVNC), reporting that it occurred mostly in young patients who experienced poor prognosis during a short-term follow-up. In 1999, we published a longer-term study in which we reported that older LVNC patients showed a better clinical prognosis than young children, with a gradual decrease in cardiac function. Further, we found that the age of onset was less than 1 year in more than half of cases. Therefore, we divided patients into 2 groups by age of onset: infantile and juvenile. For the infantile type, all neonatal cases were diagnosed by fetal echocardiography and confirmed by echocardiography performed postnatally.

Although neuromuscular and chromosomal abnormalities have been reported to be associated with LVNC, we didn’t include patients with neuromuscular disorders (NMDs) or other systemic syndromes. The study was started 20 years ago, and we focused on patients with isolated LVNC. All the patients were diagnosed by a cardiologist and a family history was considered when the patient’s first- or second-degree relatives presented with a cardiomyopathy, not only noncompaction but also dilated cardiomyopathy or other type of cardiomyopathy. Approximately half of the LVNC patients reported a family history of cardiomyopathy, but none of the parents presented with NMDs and during the follow-up, none of the patients presented with NMDs. In total, 9 patients showed a dysmorphic facial appearance characterized by a prominent forehead, strabismus, low-set ears and a high-arched palate and micrognathia: next-generation DNA sequencing is being undertaken to try to identify variants that may be associated with these features.

When diagnosed, 36% of the patients were asymptomatic, the majority diagnosed as juveniles. They were typically diagnosed during school screening examinations or family screenings as relatives of patients. Almost all the asymptomatic infantile patients developed cardiac symptoms during follow-up, whereas less than one-third of the asymptomatic juvenile patients developed cardiac dysfunction during follow-up. In addition, the infantile cases presented earlier onset heart failure than the juvenile cases. As shown in Figure 1, the age of onset distribution revealed two small peaks at 6 and 12 years old. In Japan, school screening examinations, including ECGs, are performed on all children during the first grade of primary school and then again during the first year of high school. If an abnormal ECG is found, they are encouraged to go to hospital for further examinations, including echocardiogram. Our study showed no significant difference between the 2 types of LVNC in overall morbidity after 2 decades of follow-up, although late deaths occurred from fatal arrhythmias and thromboembolisms in the juvenile type, in addition to progressive congestive heart failure.

Echocardiography video data were only available for 89 cases, therefore the noncompaction score was only reported for these. We excluded the remaining patients, including those who only had echocardiographic reports and photos, because noncompaction scores could not be accurately calculated. ICDs were implanted in LVNC patients who experienced severe ventricular arrhythmias that could not be well controlled by full doses of medication. Appropriate ICD discharge occurred in 25% and 50% of the patients in whom a defibrillator was implanted for primary and secondary prevention, respectively. Two patients with the infantile type and cardiac dysfunction died of ventricular arrhythmia; 3 patients with the juvenile type had sudden death resulting from fatal arrhythmias. In the study, none of the patients who reached an endpoint suffered from a NMD.

Cardiac resynchronization therapy (CRT) was performed if patients showed left bundle branch block. One patient who had multiple hospitalizations despite full doses of several medications underwent CRT at 4 years of age, and cardiac function improved soon after, but worsened again 3 years later, and she underwent transplantation at 7 years old.

**References**