# 肥大型心筋症における心筋トロポニンI遺伝子変異

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# 2001 Fiscal Year Final Research Report Summary

## Cardiac troponin I gene mutation in patients with hypertrophic cardiomyopathy

Research Project

### Project/Area Number

11670665

### Research Category

Grant-in-Aid for Scientific Research (C)

### Allocation Type

Single-year Grants

### Section

一般

### Research Field

Circulatory organs internal medicine

### Research Institution

Kanazawa University

### Principal Investigator

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### Project Period (FY)

**1999 – 2001**

### Keywords

Cardiac troponin I / Mutation / Hypertrophic cardiomyopathy

### Research Abstract

We examined 270 consecutive unrelated probands with hypertrophic cardiomyopathy (HCM) who underwent genetic analysis, a 12-lead electrocardiography (ECG), and echocardiography. Informed consent was obtained from all subjects or from the parents of minors participating in the study.
DNA was isolated from peripheral white blood cells of all subjects. In vitro amplification of genomic DNA was performed via polymerase chain reaction. Single-strand conformational polymorphism (SSCP) analysis of amplified DNA was then performed. For abnormal SSCP pattern, DNA sequences were determined by the Dye Terminator Cycle Sequencing method using an automated fluorescent sequencer. The lysine 183 deletion (K183del) mutation in the cardiac troponin I (cTnI) gene was identified in 10 of 270 probands with HCM. Family members of the affected probands were evaluated similarly after informed consent was obtained.
In the carrier subjects, ECG abnormalities were initially noted during the early teenage years. Abnormal Q waves were found first and were frequently observed in leads II, III, aVF, V5 and V6 in teenage patients. On the other hand, wall hypertrophy became noticeable only in their late teens, and echocardiographic abnormalities appeared later than ECG abnormalities. HCM caused by the K183del mutation in the cTnI gene has a high disease penetrance in subjects over 20 years of age. About 30% of patients with HCM caused by a K183del mutation in the cTnI gene developed systolic dysfunction after 40 years of age. The change in interventricular septal thickness and the change in % fractional shortening were significantly correlated.

# Research Products (8 results)

All Other

All Publications

[Publications] Hiromasa Kokado: "Clinical features of hypertrophic cardiomyopathy caused by a Lys183 deletion mutation in the cardiac troponin I gene"Circulation. 102. 663-669 (2000)

[Publications] Masami Shimizu: "Chronologic electrocardiographic changes in patients with hypertrophic cardiomyopathy associated with cardiac troponin I mutation"American Heart Journal. 143. 289-293 (2002)

[Publications] Masami Shimizu: "Septal wall thinning and systolic dysfunction in patients with hypertrophic cardiomyopathy caused by a cardiac troponin I gene mutation"American Heart Journal. 143(印刷中). (2002)

[Publications] Masami Shimizu: "T-peak to T-end interval is a better predictor of high risk patients with hypertrophic cardiomyopathy associated with a cardiac troponin I mutation than QT dispersion"Clinical Cardiology. 25(印刷中). (2002)

[Publications] Hiromas Kokado: "Clinical features of hypertrophic cardiomyopathy caused by a Lysl83 deletion mutation in the cardiac troponin I gene"Circulation. 102-6. 663-669 (2000)

[Publications] Masami shimizu: "Chronologic electrocardiographic changes in patients with hypertrophic cardiomyopathy associated with cardiac troponin I mutation"American Heart Journal. 143-2. 289-293 (2002)

[Publications] Masami Shimizu: "Septal wall thinning and systolic dysfunction in patients with hypertrophic cardiomyopathy caused by a cardiac troponin I gene mutation"American Heart Journal. 143(in press). (2002)

[Publications] Masami Shimizu: "T-peak to T-end interval is a better predictor of high risk patients with hypertrophic cardiomyopathy associated with a cardiac troponin I mutation than QT dispersion"Clinical Cardiology. 25(in press). (2002)

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