JCK Oral

# JCK Oral 1 (II-JCKO1)

## Basic/New Insights

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Sat. Jul 8, 2017 8:30 AM - 9:20 AM ROOM 3 (Exhibition and Event Hall Room 3)

8:30 AM - 9:20 AM

# [II-JCKO1-02]Phenotype-Genotype correlations in the fetal patients with left ventricular noncompaction

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## Background:

Left ventricular noncompaction (LVNC) is a hereditary cardiomyopathy and associated with high morbidity and mortality, but the genetic background has not been fully evaluated. The aim of the present study was to identify the genetic background using next-generation sequencing (NGS) and to identify genotype-phenotype correlations in fetal patients with LVNC.

### Methods:

We screened 73 genes associated with a cardiomyopathy in 20 fetal patients (11 males and 9 females) with LVNC for mutations by next generating sequencing (NGS). We compared the clinical features, anatomical properties and long-term prognosis between fetal patients and 111 other age patients with LVNC.

### Results:

The age at diagnosis ranged at 21 to 36 week's gestation (median: 29 week's gestation). Seven patients had a family history. Seventeen patients had congestive heart failure (CHF) and 5 patients had arrhythmias. Associated congenital heart diseases were identified in 5 patients. Nine patients died and 3 patients had intrauterine death or termination of pregnancy. Fourteen pathogenic mutations were found among 7 genes in 12 patients; 7 mutations in *MYH7* gene and 8 were novel. The *MYH7* group presented with lower age at onset and higher prevalence of congenital heart defects than that without *MYH7* mutations. The fetal patients had more frequency of positive for *MYH7* gene mutations and higher mortality than other age patients. The multivariable proportional hazards model showed that fetal patients and CHF at diagnosis were independent risk factors for death in all LVNC patients. Conclusions:

The present study was the first report focused on genotype-phenotype relationships in fetal patients with LVNC using NGS. MYH7 gene mutations can be used to predict the risk of other congenital heart in fetal LVNC patients and might have a pivotal role during maturation of heart in the fetus.