Case Report

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# Familial dilated cardiomyopathy linked with hearing loss in brothers: Case Report

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**Abstract**
Dilated cardiomyopathy (DCM) is the third leading cause of severe heart failure and the most common cause of heart transplantation. Many cases (25–30%) of DCM are familial, indicating a genetic contribution to the etiology. The diagnosis of Familial dilated cardiomyopathy (FDC) is clinically based on the clinical manifestation, with at least two affected members from the same family. More than 30 genes associated with FDC have been identified, but still theses explain only a minority of the etiology of FDC. Here we present a strange case of FDC accompanied by hearing loss and rapid progressive course. The manifestations of FDC in this family was really rare and it is anticipated that more susceptibility genes may be discovered.
Keywords: familial dilated cardiomyopathy; hearing loss; rapid progressive course

## Introduction

Dilated cardiomyopathy (DCM) is a cardiac muscle disease, characterized by dilatation and impaired contraction of the left ventricle or both ventricles, and leads to progressive heart failure and sudden or heart failure-related death [1]. The life expectancy is limited and varies according to the underlying etiology with a median survival time of about 5 years after diagnosis [2]. Although the pathogenesis of this disease has been extensively studied for decades, it remains ambiguous. Currently, myocarditis, immunological abnormalities, toxic myocardial damage, and persistent cardio-tropic viral infection are all assumed to be causes of DCM [3]. Dilated cardiomyopathy occurring in families, or the familial dilated cardiomyopathy (FDC) may occur in 25% to 35% of DCM cases, implicating a genetic contribution to the etiology [4-7]. More than 30 susceptibility genes have been shown to be associated with an increased risk of developing a DCM. Here we report three strange cases of FDC accompanied by hearing loss and rapid progressive course in brothers from Sichuan Province of China. The presentation of the family was really rare and it is anticipated that more susceptibility genes may be discovered.

## Case report

The patient was a boy from Sichuan Province, and had lost his hearing when he was five years old. At the age of eight, the boy presented with cough and acute onset breathlessness. On examination, he had blood pressure (BP) of 90/60mmHg, heart rate (HR) 105/min, raised jugular venous pressure (JVP), crackles over the lung bases and a pansystolic murmur at the apex. A huge cardiomegaly was seen on chest X-ray (CXR), and the cardiothoracic ratio (CT ratio) was 0.721. ECG revealed primary atrioventricular block and left ventricular hypertrophy (LVH). Echocardiography (Echo) showed enlargement of both ventricles of the heart, a decreased left ventricular ejection fraction (LVEF), and severe mitral regurgitation (MR). The patient was treated in line with congestive cardiac failure (CCF). However, he died three months after the acute onset of breathlessness.

Surprisingly, the progression was nearly the same as two of his older brothers. Both of them also lost hearings at the age of five. Then presented with acute onset breathlessness and they were diagnosed with DCM aged seven to eight years. They also died three months later after the acute onset of breathlessness. Because of the terrible experience of his older brothers, the boys' parents took him to hospital every year to be examined. ECG and Echo images were normal 6 months before the onset of breathlessness. Moreover, the boy had no symptoms 1 month before his presentation.

## Discussion

The definition of FDC is clinically based on manifestation with at least two affected members from the same family [5]. The most common mode of inheritance is the autosomal dominant type, while X-linked, autosomal recessive and mitochondrial forms are less common [8, 9]. Although most people affected die in early adulthood, the age of onset, rate of progression, disease complications, as well as overall prognosis and outcome vary within families [5, 10]. Nevertheless, the age of onsets in this family were similar and with a rapid progressive course. All of the sons in the family suffered from DCM as well as hearing loss. The manifestation of the brothers hasn't been reported

before. We traced back three generations of this family finding no other affected members. As all the patients were male, we speculated that the possible mode of inheritance in this family is X-linked. Regrettably, the parents had no daughters and we were not able to investigate the possible association between gender and FDC of this kind. Because of the rapid progressive course, we hypothesize that autoimmune abnormalities might be the pathogenic factors for this disease, but we do not have any solid evidence yet. Fortunately, we were able to get the blood samples from the patient and the relatives. Further studies are needed to explore new susceptibility genes as well as the molecular mechanisms that are involved in the disease.

**Competing Interests**
None declared

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**REFERENCES**

1. Richardson P, McKenna W, Bristow M, Maisch B, Mautner B, O'Connell J, Olsen E, Thiene G, Goodwin J, Gyarfas I, Martin I, Nordet P: Report of the 1995 World Health Organization/International Society and Federation of Cardiology Task Force on the Definition and Classification of cardiomyopathies. Circulation 1996;93:841-842.
2. Osterziel KJ, Hassfeld S, Geier C, Perrot A: [Familial dilated cardiomyopathy]. Herz 2005;30:529-534.
3. Chen Y, Peng Y, Zhou B, Wang Y, Zhou C, Song Y, Li C, Zhang J, Rao L: Analysis of adiponectin gene polymorphisms in dilated cardiomyopathy in a Han Chinese population. DNA Cell Biol 2010;29:313-317.
4. Ghosh N, Haddad H: Recent progress in the genetics of cardiomyopathy and its role in the clinical evaluation of patients with cardiomyopathy. Curr Opin Cardiol 2011;26:155-164.
5. Pasotti M, Repetto A, Pisani A, Arbustini E: [Genetic diagnosis of familial dilated cardiomyopathy]. Ital Heart J Suppl 2002;3:386-393.
6. Burkett EL, Hershberger RE: Clinical and genetic issues in familial dilated cardiomyopathy. J Am Coll Cardiol 2005;45:969-981.
7. Hershberger RE, Siegfried JD: Update 2011: clinical and genetic issues in familial dilated cardiomyopathy. J Am Coll Cardiol 2011;57:1641-1649.
8. Martins E, Cardoso JS, Abreu-Lima C: Familial dilated cardiomyopathy. Rev Port Cardiol 2002;21:1487-1503.
9. Zheng DD, Yang JH, Tao Q, Geng M, Lin J, Yang XJ, Song JP, Li HX, Han LH, Jiang WP: Mutations in the beta-myosin heavy chain gene in southern Chinese families with hypertrophic cardiomyopathy. J Int Med Res 2010;38:810-820.
10. Serio A, Narula N, Kodama T, Favalli V, Arbustini E: Familial dilated cardiomyopathy. Clinical and genetic characteristics. Herz 2012;37:822-829.