

## Case Report

### *Friedreich's Ataxia and Hypertrophic Cardiomyopathy: A Case Report and Review*

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#### ABSTRACT

Friedreich's ataxia is an autosomal recessive, spinocerebellar, degenerative disease characterized clinically by the ataxia of the limbs and trunk, dysarthria, loss of deep tendon reflexes, sensory abnormalities, skeletal deformities, diabetes mellitus, and cardiac involvement. Friedreich's ataxia is generally associated with concentric hypertrophic cardiomyopathy. Cardiac death occurs primarily in those developing dilated cardiomyopathy. These patients tend to do poorly with rapid progression to end-stage congestive heart failure. (*Iranian Heart Journal 2015; 16(4): 57-59*)

**Keywords** ■ Friedreich's ataxia ■ Hypertrophic cardiomyopathy ■ Familial neurodegenerative disease

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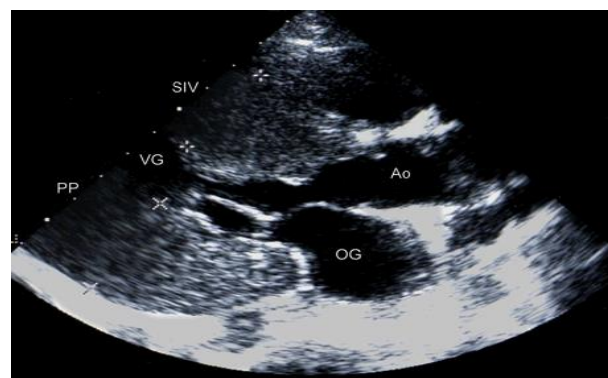
**F**riedreich's ataxia is an autosomal, recessive, multisystem disease that leads to a mitochondrial dysfunction affecting the nerve tissue and heart muscle. According to previous studies, cardiac dysfunction, predisposing to congestive heart failure and supraventricular arrhythmias, is the most frequent cause of death.

#### CASE PRESENTATION

We describe an 19-year-old female with a history of familial neurodegenerative disease, who suffered from a type 1 diabetes mellitus and presented with progressive weakness, vertigo, and loss of lower-limb muscle force of 1 year's duration. The patient had occasional respiratory distress of 6 months' duration, which was exacerbated with activity and improved with rest. She also had atypical chest pain. Physical examination detected truncal ataxia, absent deep tendon reflexes, and dysarthria. The patient's electrocardiographic (ECG) findings were

normal sinus rhythm, left axis deviation, heart rate of 94, and left ventricular hypertrophy voltage criteria.

Echocardiography showed left ventricular ejection fraction of 50%, severe left ventricular hypertrophy, no regional wall motion abnormalities at rest, concentric hypertrophy, no aortic or mitral insufficiency, no aortic stenosis, no coarctation of the aorta, and grade 1 diastolic dysfunction (Figure 1).



**Figure 1.** Echocardiography in the parasternal long-axis view shows the severity of the hypertrophic cardiomyopathy.

Given the patient's neurological findings, familial history of neurodegenerative disease, type 1 diabetes mellitus, cardiac involvement, and echocardiographic findings, Friedreich's ataxia was suggested, which was subsequently confirmed by the neurologist.

## DISCUSSION

We reported the case of a 19-year-old female, who presented with progressive weakness, vertigo, and loss of lower-limb muscle force of 1 year's duration. In her ECG, there were left ventricular hypertrophy voltage criteria. The most prominent point in her echo was concentric hypertrophic cardiomyopathy.

According to her neurological findings, familial history of diabetes mellitus, cardiac involvement, and echocardiographic findings, Friedreich's ataxia was suggested. The neurologist subsequently confirmed the diagnosis.

Friedreich's ataxia is an autosomal, recessive, spinocerebellar, degenerative disease characterized clinically by the ataxia of the limbs and trunk, dysarthria, loss of deep tendon reflexes, sensory abnormalities, skeletal deformities, diabetes mellitus, and cardiac involvement.<sup>1</sup> It is worthy of note that 98% of the patients have an expansion of GAA trinucleotide repeat located within the first intron of the FXN gene.<sup>2</sup> In addition, larger GAA expansions are correlated with earlier age at onset and shorter times to loss of ambulation.<sup>2</sup>

Friedreich's ataxia is the most common inherited spinocerebellar degenerative disease, with a prevalence of 1/50000. Neurological symptoms usually manifest around puberty and almost always before the age of 25 years. Progressive loss of neuromuscular function and neurological symptoms precede cardiac symptoms in most but not all cases. Friedreich's ataxia is associated with a high incidence rate of diabetes mellitus.<sup>3</sup> Clinically apparent diabetes is seen in approximately 18% of the

affected individuals, while impaired glucose tolerance is present in up to 37% of patients with Friedreich's ataxia.<sup>3</sup>

Friedreich's ataxia is generally associated with concentric hypertrophic cardiomyopathy. Less commonly, asymmetric septal hypertrophy is observed. The presence of a left ventricular outflow gradient associated with septal hypertrophy has been reported. Left ventricular hypertrophy is not always present on the ECG despite echocardiographic evidence. Widespread T-wave inversions are common.<sup>4</sup> The prevalence of hypertrophy varies among studies but increases in prevalence with a younger age at diagnosis and with increasing GAA trinucleotide repeat length. Myocardial fiber disarray is not commonly seen in the hypertrophic cardiomyopathy of Friedreich's ataxia.<sup>5</sup> In most patients with Friedreich's ataxia, progressive neurological dysfunction is the norm, with death from respiratory failure or infection in the fourth or fifth decade. Cardiac death occurs primarily in those developing dilated cardiomyopathy. These patients tend to do poorly with rapid progression to end-stage congestive heart failure. It is unclear whether pharmacological or ICD therapy improves outcomes in Friedreich's ataxia and dilated cardiomyopathy.<sup>6</sup>

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