A Case Report of Protein Kinase Amp Activated Non Catalytic Subunit Gamma 2 Syndrome (Prkag2 Syndrome)

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Abstract
Background: PRKAG2 syndrome is a rare, early onset autosomal dominant inherited disease, characterized by ventricular pre-excitation, supraventricular arrhythmias, and cardiac hypertrophy. In cardiomyocytes the enzyme regulates the glucose and fatty acids uptake, storage and utilization.1

Case report: We present a 44 years old female patient, who came to Wuhan Union hospital with the following presenting complains, chest pain, awareness of heart beat and dizziness for 3 weeks, she denies history of hypertension and diabetes mellitus, she has a strong family history of heart diseases where two nephews were diagnosed of Hypertrophic Cardiomyopathy and one niece was diagnosed of having PSVT-WPW and a cousin with bradycardia. Clinical history and ECG and ECHO findings are suggestive of PRKAG2.

Conclusion: Even though PRKAG2 syndrome has an early onset, our patient presented with symptoms at 44 years making it important because even though its rare may also present at late stage of life.

Keywords: PRKAG2- protein kinase amp activated non catalytic subunit gamma 2, HCM- hypertrophic Cardiomyopathy, ECG-electrocardiogram, ECHO-Echocardiogram, PSVT-paroxysmal Supraventricular tachycardia, WPW- wolf Parkinson white syndrome, RBBB-right bundle branch block

I. Introduction
PRKAG2 syndrome is a rare, early onset autosomal dominant inherited disease, characterized by ventricular pre-excitation, supraventricular arrhythmias, and cardiac hypertrophy. It is frequently accompanied by chronotropic incompetence and advanced heart blocks, leading to premature pacemaker implantation.1 The prevalence of PS is currently unknown. AMPK is a highly conserved serine/threonine protein kinase responsible for cellular energetic homeostasis control. Stimulated by high AMP concentration and AMPK kinase activity, the enzyme counterbalances ATP depletion. It is composed of a catalytic α subunit and 2 non catalytic, but regulatory subunits β and γ. γ subunit of AMPK (PRKAG2) binds AMP, enhancing the activation of α-subunit. AMPK is highly expressed in Cardiac tissue, skeletal muscle, brain, placenta, liver, kidneys and pancreas. In cardiomyocytes the enzyme regulates the glucose and fatty acids uptake, storage and utilization.1, 2

The most common electrocardiographic features of PRKAG2 syndrome are; short PR interval, tachycardia, supraventricular arrhythmias, and cardiac hypertrophy. In cardiomyocytes the enzyme regulates the glucose and fatty acids uptake, storage and utilization.1, 2

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II. Case Presentation
We present a 44 years old female patient, who came to Wuhan Union hospital with the following presenting complains, chest pain, awareness of heart beat and dizziness for 3 weeks, she denies history of hypertension and diabetes mellitus, she has a strong family history of heart diseases(4 family members) where two nephews were diagnosed of Hypertrophic Cardiomyopathy and one niece was diagnosed of having PSVT,WPW-had a radio frequency ablation and a cousin with bradycardia on physical examination she was conscious, not pale, not jaundiced, not dyspneic, vital signs BP=115/80mmhg, HR= 200b/min regular, full volume, strong character. Several investigations were done, Thyroid function test, renal function test, liver function test, CBC, cardiac profile, PT, PT, INR lipid profile were normal, BNP is slightly elevated 107.8pg/ml.
Chest X-ray was normal, Echo revealed interventricular septum thickening. ECG revealed atrial flutter with Right bundle branch block and high voltage QRS, genetic testing was done and confirmed the diagnosis.

**Figure: 1 ECG showing atrial flutter with Right bundle branch block and high voltage QRS**

**III. Discussion**

According to our patient she presented with ECG characteristics of atrial flutter and RBBB, echo revealing Thickened IVS also with a positive family history and positive genetic testing for PRKAG2 syndrome, even though PRKAG2 has early onset but our patient was asymptomatic for most of her life but did present with all classical features of PRKAG2, whereby our patient presented with ECG characteristics of atrial flutter, RBBB and high voltage QRS complexes, Thickened IVS on Echo and a strong family history of heart diseases with 2 nephews with HCM, a niece with PSVT and a cousin with bradycardia. Our patient had EPS and radiofrequency ablation was done to cardiovert and was successful, and kept on Diltiazem and Metoprolol currently our patient has no complains.

**References**