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CASE REPORT

Yamaguchi Apical Cardiomyopathy: A Rare Presentation of an Autosomal Dominant Cardiac Illness in a Young Patient

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ABSTRACT

Yamaguchi apical cardiomyopathy, also known as Yamaguchi syndrome or apical hypertrophic cardiomyopathy, is a rare subtype of hypertrophic cardiomyopathy (HCM), which is itself a genetic cardiac disorder characterized by thickening of the heart muscle (myocardium). However, unlike the typical form of HCM where the thickening occurs primarily in the interventricular septum (the wall that separates the left and right ventricles), Yamaguchi apical cardiomyopathy predominantly affects the apex (tip) of the heart. The condition is autosomal dominant and presents with symptoms like chest pain, palpitations, dyspnea, syncope and other similar features of an acute coronary syndrome. Majority of mutations occur in genes encoding the sarcomere. The predominant gene mutations occur in "myosin-binding protein C (MYBPC3) and myosin heavy chain (MYH7). Here is a case of a 40-year-old male patient who presented to the emergency with complaints of chest heaviness and diaphoresis since the past one day. Patient had a similar complaint around 3 years back for which he underwent a coronary angiography and was found to have normal coronaries. On examination, patient had normal findings and including the primary and secondary survey. Patient's ECG showed Deep T waves. In view of ECG findings, patient was shifted for a coronary angiography which showed normal epicardial coronaries. 2D echo showed an apical variant of HCM. Patient was discharged in a stable condition.

KEYWORDS

• Cardiomyopathy • Genetic • Apex • Myosin

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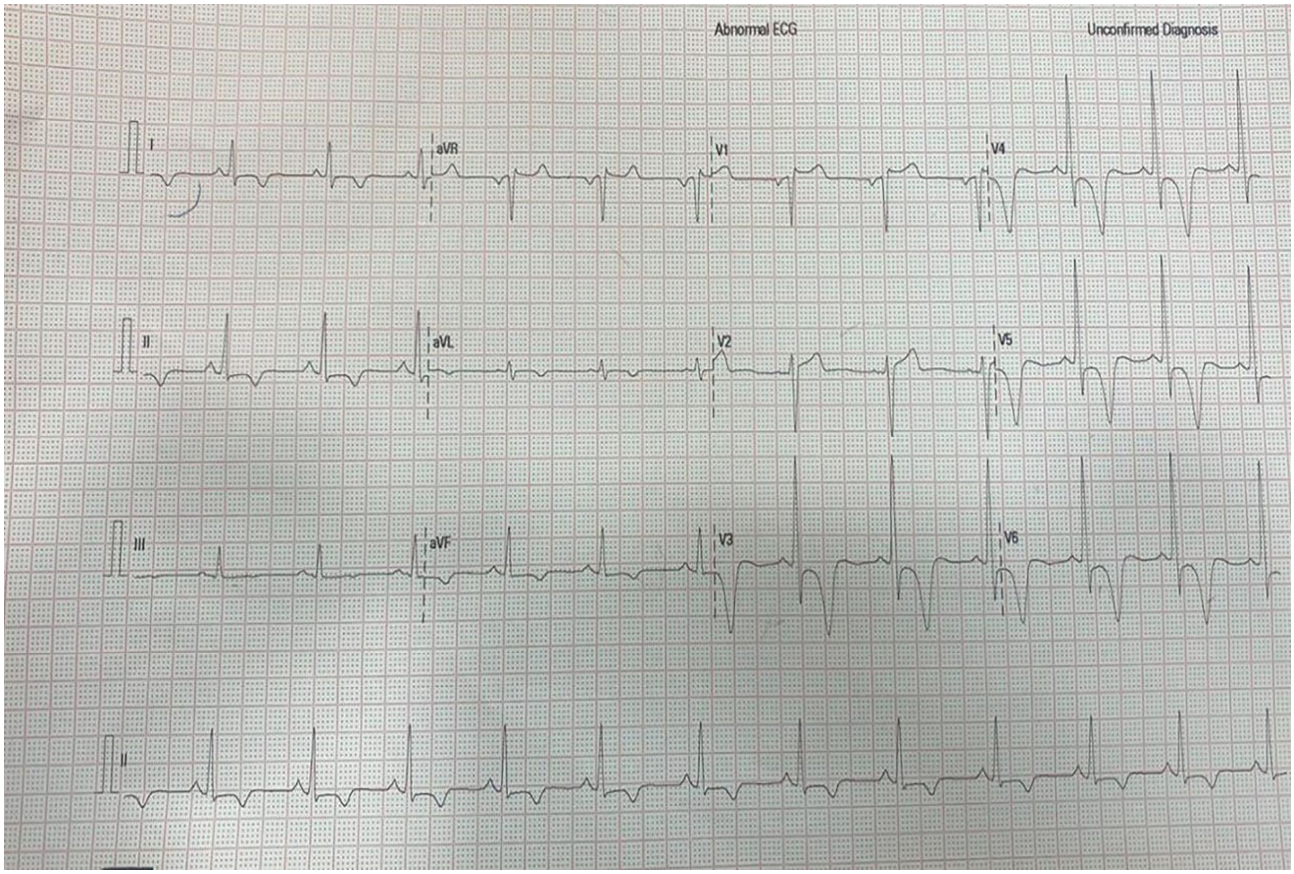
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CASE REPORT

This is a case of a young 40-year-old male patient who presented to the emergency department with complaints of chest heaviness since the past one day which worsened over the night associated with dizziness that had started the same morning of presentation. On physcial examination, the patient looked stated age, comfortable and not in distress with stable vitals on primary survey and normal examination on secondary survey.

On taking detailed history, the patient reported that he underwent coronary angiography around 3 years back which was found to be normal as verbally reported by the patient, however there were no documents availed at the time of presentation. The patient was since then started on Tablet Ecosprin 150 at bedtime.



ECG done showed Deep T wave inversions in leads V3-V6; II, III and aVF

Labs were sent which included complete blood count and cardiac enzymes. TLC was 12.8; TropI - 0.01 and CKMB - 0.76. Patient was taken for a coronary angiography the same day which showed the following results:

1. Access - Right Radial
2. Left main - Normal
3. Left anterior descending - Normal
4. Diagonal 1 - High , major, normal
5. Left circumflex - Non dominant, normal
6. RCA - Dominant, normal
7. Post. Descending - Normal



Final impression: Normal Epicardial Coronaries

Patient was discharged with a final diagnosis of hypertrophic cardiomyopathy (Non obstructive/Apical); Normal coronaries and Good LV function

DISCUSSION

A cardiac condition more prevelant in the Japanese population has slowly found it's way to other countries and is now rarely but seen in countires like India. Early recognition, intervention and accurate diagnosis can help young patients live a healthy and delay mortalility with an expectancy of a normal life.

CONCLUSION

Recognition of Yamaguchi HCM requires combination of clinical suspicion, appropraite diagnostic tests, and expertise in interpreting imaging findings. A rare finding yet significant cardiac condition which could result in serious complications was identified and timely recognition helped in and proper assessment and evaluation in a young patient with an underlying disease.

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