

ECAST Syndrome(Exercise collapse associated with Sickle Cell Trait): first knock of sickle cell in a Young Bodybuilder

Abstract

ECAST Or exercise collapse associated with sickle cell trait is a rare phenomenon associated with sickle cell trait and is an important presentation of sickle cell disease in sports medicine. Collapse is seen following vigorous physical activity, which is due to excessive heat, dehydration and other factors associated with physical exercise. This rare syndrome is often missed by the treating physicians as a result of a lack of knowledge about this rare entity leading to massive underreporting. It is important to identify ECAST as a cause of the collapse in young athletes to prevent mortality and morbidity and in order to provide prompt treatment. We report a case of a 25-year-old young male who was a bodybuilder and reported to the gym after a one-year-long break due to lockdown restrictions of COVID19. After a vigorous exercise session, he collapsed in the gym and was brought to the emergency department. After proper history taking and examination, he was suspected to be a case of ECAST due to a history of a similar episode three years back which was treated as a case of exertional syncope with intravenous fluid therapy and a family history of Sickle cell trait with his mother and father both having sickle cell AS Pattern. Ultimately our patient turned out to be a case of Sickle Cell Trait with evidence of AS pattern on Hb electrophoresis and a small-sized spleen visualized on CT Scan of the abdomen. The patient was managed successfully with intravenous fluids and blood transfusion and was discharged in a stable condition. He was counseled about moderating his exercise and is doing well on follow-up.

Keywords: ECAST,Sickle Cell Trait,Sports Medicine

Introduction

Hemoglobin is a tetrameric protein which is formed with combination of globin subunits where each of the subunit of globin is linked to a co factor known as heme that has the ability to carry oxygen molecule [1].Haemoglobin is expressed by both types of red blood cells,the immature red blood cells known as reticulocytes and erythrocytes which are the mature red blood cells.The different globin proteins are in turn coded by different genes. Various genes have the ability to code for different types of globin proteins as we their tetrameric combinations. This leads to the formation of different types of hemoglobin which are expressed during different phases of life namely embryonic followed by foetal and then adulthood.Haemoglobin A is the most abundant hemoglobin which is comprised of two alpha globin subunits which are coded by HBA1 and HBA2 genes and two beta globin subunits.A mutation in the HBB gene leads to the substitution in beta globin protein that results in formation of sickle haemoglobin (HbS) .Whenever there is deoxygenation and the

oxygen molecule is unbound to the hemoglobin, haemoglobin tetramers comprising of two of these mutant beta globin subunits polymerize and direct the erythrocytes to form a crescent shape [1]. Hemoglobin tetramers comprising of only one of the HbS subunit can also lead to polymerization however not very effectively. Recurrent vaso-occlusive crisis may result from sickling of the red blood cells which are characteristic feature of sickle cell disease.

Sickle cell disease is inherited as an autosomal codominant trait and the individuals who are homozygous for the sickle cell allele have sickle cell anemia whereas the individuals who are heterozygous for the sickle cell allele carry the sickle cell trait [1].

Normally individuals with sickle cell trait can participate in athletic events; however, in rare circumstances, exercise can have a negative impact on their health. This includes pain, weakness of the muscle and collapse. If collapse occurs in an athlete with AS Pattern following exercise, it is referred to as ECAST. It is important to identify and treat ECAST as it causes the risk of death to increase by 40 times in athletes with sickle cell trait when compared to other athletes [2]. Early diagnosis and treatment of ECAST prevents mortality and reduces the complications associated with this rare syndrome [2].

CASE PRESENTATION

A 25 year old young male was brought to the emergency department following an episode of collapse after an intensive work out session in the gym. The patient was doing running on the treadmill followed by heavy weight lifting on a hot day in Wardha, Maharashtra with 44.4 degree Celsius temperature. While carrying out his work out the patient felt extreme pain in the legs and the back followed by collapse. The patient had previous history of similar episode three years back for which he was given intravenous fluids and was diagnosed as exertional syncope. There was no history of any bleeding tendencies in the past. There was no history of fever, cough, cold, chest pain, palpitations or orthopnea. There was no history of hypertension, diabetes mellitus or bronchial asthma in the past. On family history both his parents were revealed to have sickle cell AS pattern.

On general examination patient was afebrile, pulse was 88 beats per minute, regular in rhythm, normal in volume, all peripheral pulses were well felt, blood pressure was 120/80 mm hg in right arm supine position, orthostatic hypotension was ruled out, Jugular venous pressure was not raised with absent abdomino-jugular eflux, pallor was present and spo₂ was 97 percent on room air. On systemic examination chest was bilaterally clear with equal air entry, Heart sounds were normal with no murmurs, abdomen was soft and non tender with no hepatosplenomegaly and patient was conscious, oriented to time, place and person, deep tendon reflexes were normal, bilateral plantar were flexor and there was no neurological deficit. Muscle tenderness was present in the upper and lower limbs, the muscles of the fore arm and thigh were soft and tender with a power of 4/5.

Patient was admitted and lab investigations were done and are mentioned in table number 1. Random blood sugar was 152 mg/dl and MRI Brain was done which was normal.

An ultrasonography was performed which showed small spleen which was confirmed on CT Scan of abdomen (Figure 1) and therefore hb electrophoresis was done which revealed AS Pattern. As electrolytes were normal, orthostatic hypotension was ruled out, MRI Brain was normal and heat exhaustion was also ruled out a diagnosis of ECAST was made.

Patient was managed with intravenous fluids, blood transfusion and other supportive therapy. He responded well to treatment and was discharged in stable condition five days after admission .



Figure 1:CT Scan showing small sized spleen suggestive of sickle cell anemia

Lab Parameter	Measured Value
CBC	Hb-6.6 gm/dl MCV-73fl Platelet count-180000/dl WBC Count-8900/dl
LFT	Total Protein-7.2gm/dl, Albumin3.6gm/dl, Globulin3.6gm/dl, aspartate aminotransferase 31 units/l , alanine aminotransferase 27 units/l, AlkanlinePhophatase102IU/l, Total Bilirubin :1.9mg/dl Unconjugated 1.2mg/dl Conjugated 0.7 mg/dl
KFT	Creatinine:1.2 mg/dl, Urea 32mg/dl, Sodium133 mmol/l, Potassium -4.3mmol/l
D-Dimer	0.45

Table 1:Showing lab investigations of the case

Discussion

Patients with sickle cell trait have one gene mutation coding for the two chains of hemoglobin leading to formation of sickle cell red blood cells in conditions of deoxygenation. ECAST associated with sickle cell disease has a complex mechanism. It is unexpected to encounter collapse or death in an individual with sickle cell trait who otherwise have stable course of disease. Extremely low muscle oxygen, raised body temperature and dehydration all may contribute to sickling of red blood cells when an individual with sickle cell trait exercises [3]. Also, when there is physical exertion the level of epinephrine in blood rises rapidly which makes the sickled red blood cells to become sticky and block the blood vessels. It is not clearly understood why only some individuals with sickle cell disease develop ECAST. The risk factors likely to contribute in predisposing to ECAST are intense physical activity over a long period of time, inadequate rest between intense physical activity, return to athletic activity after prolonged period of rest, high altitude and excessive exposure to heat. Other contributing factors such as increased humidity, asthma (exercise induced), fatigue due to illness and stimulants used as dietary supplements may also play crucial role in predisposing an individual to ECAST [4]. Our patient was performing high intensity running followed by heavy weight lifting in a hot environment of central India with a temperature of 44.4 degree Celsius which may have contributed in developing ECAST. Presenting symptoms of ECAST include weakness of muscle, muscular tenderness, dropping to the ground, tachypnoea and fever (usually less than 103 degree Fahrenheit). There are several ways to differentiate ECAST from other causes of collapse. Athletes with muscle cramps usually have tight, hard muscles which is different than soft and weak muscles of ECAST. Athletes with exertional heat stroke will have a body temperature above 104 degree Fahrenheit whereas in ECAST the temperature will be normal or slightly elevated with a conscious and oriented patient. Pain and weakness of the muscles is the most definitive symptom of ECAST [5]. ECAST can lead to serious medical conditions such as explosive rhabdomyolysis and therefore should be diagnosed and treated promptly. Treatment of ECAST includes immediate intravenous fluid transfusion and monitoring of electrolytes as well as heart function. Prevention of ECAST includes screening for Sickle cell trait in the athletes, provision of adequate recovery time during work out session, increasing exercise intensity gradually, carrying out mild physical activity routinely throughout the year, giving time for acclimatisation to altitude or temperature, proper hydration before during and post exercise. If an athlete is known case of sickle cell trait he/she should be counselled about the risk factors and symptoms of ECAST and to report to medical staff immediately in case of appearance of any symptoms. Before an athlete returns to physical activities post an episode of collapse, he/she should be tested for sickle cell trait. Other diseases such as diabetes, electrolyte imbalance and muscle damage should also be ruled out. Training or physical activity should only be resumed slowly and under a doctor's supervision.

Conclusion

ECAST is a rare phenomenon encountered in sickle cell trait individuals carrying out physical activities without proper precautions. The treating clinicians should therefore be made aware of this rare but important consequence of sickle cell trait in order to enable prompt diagnosis and prevention of complications.

Ethical Approval:

As per international standard or university standard written ethical approval has been collected and preserved by the author(s).

Consent

As per international standard or university standard, patients' written consent has been collected and preserved by the author(s).

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