CONGENITAL METHEMOGLOBINEMIA: A CASE OF 14-YEAR-OLD MALE

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PRESENTATION OF CASE

A 14-year-old male was presented to our cardiology department with complaints of bluish discolouration of both hands (Figure 1) and feet and easy fatigability for past 2 years. This peripheral cyanosis was first perceived by his parents 2 years ago and it did not advance since then.

A brief history elicited no other symptoms like shortness of breath, chest pain, fever, cough, generalized swelling of body, numbness or paraesthesia, decreased level of consciousness, abnormality in body movement, visual or auditory deficits, gastrointestinal complaints, bone and joint pain or genitourinary symptoms. There was no history of weight loss or diminished appetite. His birth history was also unremarkable and had achieved all the developmental milestones timely. There was no history of any failure to thrive, feeding difficulties or recurrent chest infections in his childhood. He was a student and did not report any exposure to chemicals or drugs prior to the onset of symptoms. He did not use any tobacco, illicit drugs or alcohol. There was no significant medical history in the past. He lived in the city and never travelled outside the state. A general physical examination revealed no abnormality other than the cyanosis of the extremities and the lips. His blood pressure was normal (110/70 mmHg) and pulse rate was 75 beats per minute.

Oxygen saturation level of arterial blood was 96% in both the hands and feet as measured by pulse oximeter. Cardiovascular examination was unremarkable. A review of the other systems did not show any abnormality. Laboratory investigations of blood revealed normal parameters including a normal haemoglobin level, a normal reticulocyte count, normal peripheral blood smear, normal liver function tests. The chest X-Ray, electrocardiogram (Figure 2) and echocardiogram of the patient were also normal. On basis of his history and laboratory investigation, no cardiac cause could be determined.

Cyanosis should be approached with the brief history, physical and laboratory examination of patient to identify and eradicate any underlying cause. This patient came to us with complaints of cyanosis and fatigue; and there can be many possible justifications like decreased arterial oxygen saturation, haemoglobin abnormalities, reduced cardiac output, exposure to cold environment, impaired pulmonary function, anatomic shunts, high altitude (low atmospheric pressure), arterial venous obstruction, and redistribution of blood flow from extremities. Several questions can be helpful in diagnosis of methemoglobinemia: whether the cyanosis is central or peripheral, sometimes exposure to cold weather can also cause peripheral cyanosis (pseudocyanosis); when inception of cyanosis was, may provide allusion of congenital or acquired methemoglobinemia; clubbing associated with cyanosis like congenital heart disease or pulmonary disease.[1]

Patients with cyanotic heart disease do have a prolonged history of cyanosis and fatigue as our patient had for past 2 years. However, this patient did not show any other symptoms or did not give any history suggestive of congenital heart disease such as frequent respiratory infections, failure to thrive or feeding difficulties. His general physical examination and review of the system were also normal. His blood pressure, pulse rate and oxygen saturation were normal and was also breathing comfortably on room air. The cardiac apex was normally positioned, and heart sounds were also heard normally without any murmur or any additional sound on auscultation. Finally, echocardiogram of the patient confirmed no possibility of structural heart disease. If not congenital, patient can also acquire methemoglobinemia from exogenous substances like nitric oxide, nitroprusside, sulphonamides, lidocaine, prilocaine, minocycline, arsenic, chloroquine. Primaquine.[2-5] Other than these substances, poisoning due to herbs (containing ergot alkaloids) can also lead to bluish pigmentation of skin.[6] Intake of such herbs is common practice in Indian households. However, suspicion of ingestion of any such substances or herbs was ruled out by patient. After ruling out the possibility of acquired methemoglobinemia, the patient most likely had congenital methemoglobinemia. Congenital methemoglobinemia occurs either due to blood enzymopathies (cytochrome b5 reductase deficiency) or due to haemoglobin M (single globin gene mutation). However, we could not perform enzymatic tests on patient due to financial constricts. Even though normal functioning of haemoglobin is impaired, the patient doesn't show any signs of cardio-pulmonary dysfunction. Congenital methemoglobinemia is quite rare and for type 1 disease reported median onset of age was 31 years.[7]

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Figure 1. Cyanosis Observed during Initial Inspection in Patient

CLINICAL DIAGNOSIS

Methemoglobinemia

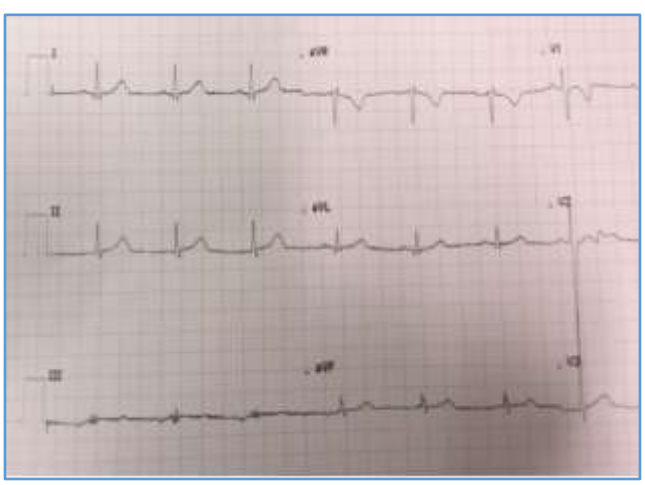

Figure 2. Electrocardiogram of Patient

PATHOLOGICAL DISCUSSION

Investigations revealed the level of methaemoglobin to be 22.9% (normal: 0-1.5%).

DISCUSSION OF MANAGEMENT

The patient was prescribed ascorbic acid tablets for 1 month. He returned for follow up after 1 month and reported neither improvement nor deterioration than the previous ailment. However, treatment with ascorbic acid is not indicated in congenital chronic methemoglobinemia except for cosmetic reasons. Congenital methemoglobinemia lacks systemic epidemiological studies as the disease is rare and under diagnosed when presented in adulthood. No offending agent or herb was found in the presented case and patient did not act in response to the available medication for treatment of congenital methemoglobinemia.

FINAL DIAGNOSIS

Congenital Methemoglobinemia

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