

Methemoglobinemia (Case report)

AL Kubati AK Sallam

Pediatric, Sana'a University, Yemen

E-mail: draksallam@gmail.com

DOI: <https://doi.org/10.47372/uajnas.2018.n2.a21>

Abstract

Congenital methemoglobinemia is a rare overlooked differential diagnosis in patients presented with generalized (peripheral and central) cyanosis unrelated to cardiopulmonary causes. Here, we report a case of 9-year-old boy who was diagnosed as methemoglobinemia. He was presented with peripheral and central cyanosis, while his cardiovascular and respiratory system were normal. This case stresses the importance of alertness among the treating physicians to this disease occurring in a patient with cyanosis unconnected to cardiopulmonary causes.

Keywords: Methemoglobinemia; Congenital; Cyanosis.

Introduction

Congenital methemoglobinemia is a rare overlooked differential diagnosis in patients presented with cyanosis unconnected to cardiopulmonary causes. Methaemoglobinaemia is an uncommon etiology of cyanosis, but one that need prompt diagnosis and treatment. Methemoglobinemia is usually asymptomatic, even when methemoglobin (metHb) levels are as high as 40% of the total hemoglobin (Hb) value. History, physical examination, bedside diagnostic techniques and laboratory confirmation are all important in the evaluation. [1,2]

Case Report

A9-year-old boy, from Shamir, Taiz Government, was presented with peripheral and central cyanosis. His complaints were prolonged recurrent headache, weakness, occasional, visual disturbances, tinnitus, pruritus after exposure to warm water with burning pain, warmth, of extremities, epigastric discomfort, anorexia and constipation since 4 years. No other symptoms related were to cardiovascular and respiratory system.

On examination, he had bluish discoloration of lips, tongue and skin and had no jaundice. His growth was normal, moderately built, his body weight was 22 kg, and he was not aware of his peripheral cyanosis. His cardiovascular and respiratory systems were normal. Other systems were normal, no hepatosplenomegaly or organomegaly were detected. His Pulse was 85 beats/min, respiratory rate 15 cycle/min and his BP 100/60 mmHg was normal. Once we extract blood from the patient, it was difficult to extract and thick and not easy to extract by 5 cc bore syringe, repeated trial, blood was taken, then placed on a piece of filter paper and when dried it turn into a deep chocolate-brown, Laboratory investigations revealed that hemoglobin was 17.6 g/dL (normal for his age is 11-14), white blood cell count was 4000 cells/mm³ with a differential count of 57% polymorphs (NR - 40-70%), 41% lymphocytes (NR - 20-45%), and 2% monocytes (NR - 2-10%). Erythrocyte sedimentation rate (Westergren) was 20 mm in the first hour, platelet count was 170×10³/μL, and reticulocyte count was 0.5%. RBC-4.5.40X10⁶ Cells/mm³, PCV-42.8%, MCV, MCHC, MCH were normal. Hemoglobin electrophoresis revealed adult hemoglobin of 95% (AA) and no M band hemoglobin (Figure 4 photos), metHb concentration showed 24% while the normal reference value was 0-3%, Erythropoietin level was 137 and the normal range was 3.2-32mIU/ml., the metHb reductase enzyme deficiency was not done in our country. RBC morphology normocytic and normochromic. Renal and liver function tests were normal, except random blood sugar, was 65mg/dl, ECG and ECHO, CXR, abdominal US, brain CT were normal. For the above features, history, investigations and examination was diagnosed as methemoglobinemia.

Discussion

Methaemoglobinaemia can be acquired secondary to exposure to oxidant agents; few such agents commonly used in anesthesia practice include nitroglycerine, local anesthetics, like lidocaine and prilocaine [3,4].

Congenital causes of methaemoglobinaemia include deficiency of enzyme NADH cytochrome-b5 reductase (autosomal recessive), cytochrome-b5 deficiency (autosomal recessive) or hemoglobin M disease due to globin chain mutation (autosomal dominant) [5,6].

Methaemoglobinaemia is a condition in which the iron within hemoglobin is oxidized from the ferrous (Fe^{2+}) state to the ferric (Fe^{3+}) state, resulting in the inability to transport oxygen and carbon dioxide [7-9]. Methaemoglobinaemia occurs when methemoglobin levels is more than 2%. The failure of 100% oxygen to correct cyanosis is suggestive of methaemoglobinaemia [10,11]. Diagnosis is based upon central cyanosis unresponsive to oxygen therapy decreased measured oxygen saturation in presence of a normal PaO_2 . Since methemoglobin has absorption characteristic similar to that of deoxyhaemoglobin, its presence in blood lowers the saturation as read on the pulse oximeter. The saturation reported on the arterial blood gas is based on the partial pressure of dissolved oxygen and assumes no abnormal hemoglobin is present, therefore the reported oxygen saturation in arterial blood gas analysis is higher than that measured with the pulse oximeter [12].

This rare *congenital methemoglobinemia* (10% to 50% of total hemoglobin) may be found in persons with either hemoglobin M disease* or a deficiency of NADH-dependent methemoglobin reductase. *Acquired methemoglobinemia* is caused by exposure to oxidizing substances including nitrates and nitrites. Persons with a deficiency of NADH-dependent reductase may be more susceptible to developing symptomatic methemoglobinemia after exposure to nitrates and nitrites. A disease caused by a group of abnormal hemoglobin in which a single amino acid substitution favors the formation of methemoglobin, in spite of normal quantities of methemoglobin reductase.

In the meantime, in our country, the public water sources are usually not monitored for nitrate concentrations and no screening for rural wells to test for nitrates. Methemoglobinemia must be taken into consideration in the differential diagnosis of peripheral and central cyanosis. This case alerts general practitioners, pediatricians, and health authorities to the possibilities of methemoglobinemia in patients presented with cyanosis in the absence of cardiopulmonary causes.

Acknowledgment

The authors gratefully acknowledges and sincerely thanks all the laboratory technicians and all team working in Shamir, Taiz Government, Yemen section who participated in the study.



Figure – 4 Photographs of a 9-year-old patient 'revealing generalized cyanoses all over the body

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ميثيموغلوبينية الدم (دراسة حالة)

عبد الخالق سلام القباطي

طب أطفال، كلية الطب، جامعة صنعاء

DOI: <https://doi.org/10.47372/uajnas.2018.n2.a21>

المخلص

ميثوغلوبين الدم الوراثي هو تشخيص تفاضلي نادر يتم تجاهله عند المرضى المصابين بأزرقاق عام في الجسم لا علاقة لها بالأمراض القلبية والرئوية. يسعى هذا البحث إلى تقرير حالة طفل يبلغ من العمر 9 سنوات الذي تم تشخيصه بأنه ميثيموغلوبينية الدم. لوحظ أزرقاق كامل في جسمه في حين أن نظام القلب والأوعية الدموية والجهاز التنفسي كانت طبيعية. وتشدد هذه الحالة على أهمية اليقظة بين الأطباء المعالجين لهذا المرض الذي يحدث في مريض مصاب بأزرقاق غير مرتبط بالأمراض القلبية والرئوية.

الكلمات المفتاحية: ميثيموغلوبينية الدم، خلقي، الأزرقاق.