

An incidental finding of congenital methaemoglobinaemia in a patient with an allergy

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Case report

A 23-year old unmarried woman who had been apparently well until three days prior to admission had developed urticarial rash, cough and difficulty in breathing after eating beef. On admission, she was comfortable at rest, not pale, and had central cyanosis. There was no finger clubbing or ankle oedema. She was haemodynamically stable, heart sounds were normal and there were no murmurs. Respiratory system examination was unremarkable except for occasional ronchi on auscultation. Her oxygen saturation on room air was 85% and it did not improve with additional oxygen supplementation via face mask.

She had a good clinical recovery from the allergic reaction to beef and became asymptomatic in two days. However the central cyanosis was persistent and her oxygen saturation on room air was persistently low and remained around 85% despite clear lung fields. This warranted further evaluation.

ECG, chest radiograph and other initial blood investigations including full blood count and ESR were normal. Blood gas analysis was performed while her saturation was 85% on room air. It showed, pH 7.46, pCO₂ 36 mmHg, pO₂ 97 mmHg, SpO₂ 97%, HCO₃ 25 mmHg. The 2D echocardiogram and contrast echocardiogram with agitated saline were normal and this excluded intracardiac and pulmonary A-V shunts.

She was the third child of a consanguineous marriage. Her sister and brother who were asymptomatic had saturations of 88% and 92% respectively on pulse oximetry. Photospectrometry for methaemoglobin was positive. Haemoglobin electrophoresis excluded haemoglobin M disease. A diagnosis of congenital methaemoglobinaemia was made based on clinical and laboratory evaluation. She was counselled about the importance of avoiding certain medications and substances.

Discussion

Methaemoglobin is the oxidised form of haemoglobin

and it can be formed due to congenital enzymatic defects, alterations in the hemoglobin molecule, or as a result of medications or toxins. Co-oximetry is the gold standard for diagnosis.

Congenital methaemoglobinaemia due to NADH-cytochrome b5 reductase 3 deficiency is an autosomal recessive disorder. It present as two distinct phenotypes, type I and type II. In type I, the enzyme deficiency is restricted to erythrocytes and associated with benign cyanosis while the less common type II is associated with generalised enzyme deficiency affecting all cells and is lethal in early infancy [2].

The SpO₂, when measured in arterial blood gas analysis, is calculated from blood pH, the SpO₂ and the standard haemoglobin dissociation curve. Methaemoglobin can falsely elevate the calculated SpO₂. CO oximeter measures light absorbance at four different wavelengths that correspond to the absorbance characteristics of deoxy-haemoglobin, oxy-hemoglobin, carboxy-hemoglobin and methaemoglobin. Therefore CO oximeter can distinguish between these and can give a more accurate measurement of SpO₂ [3]. The difference between the calculated oxygen saturation from a standard blood gas machine and the reading from the pulse oximeter is the "saturation gap". If it is more than 5%, it may be due to carbon monoxide poisoning, methaemoglobinaemia or sulfhaemoglobinaemia [4].

Symptoms correlate with the level of methaemoglobin in the blood and the treatment with methylene blue is warranted for high levels of methaemoglobin. If the patient has got a comorbid condition which impaired oxygen saturation, even with low levels of methaemoglobin, patient may become symptomatic and may warrant treatment [5]. When the methaemoglobin levels below 3%, there are usually no symptoms. Cyanosis starts when it increases to 15%. Dyspnoea usually sets in between the 30%-50%. When severe patients can develop metabolic acidosis, cardiac arrhythmia, seizures, central nervous system depression and coma. Death usually occur with levels about 70% [6].

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Acquired methaemoglobinaemia due to beef allergy or a toxin is also a possibility in this patient as it is much more common than the congenital form. However persistently low O₂ saturation during the follow up period together with affected family members make congenital methaemoglobinaemia the most likely diagnosis.

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