

# When the baby might feel blue, but isn't: Congenital Methemoglobinemia

Angelica Ostrowski, DO, Steven Marek, MD, Marie-Ellen Sarvida, MD

Advocate Children's Hospital Department of Pediatrics



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## Patient Background

- 2-week term female
- normal maternal serologies
- newborn screen positive for galactosemia
- previously feeding well and gaining weight appropriately

## Presentation in Emergency Department

- 3 days of diarrhea, poor feeding, lethargy
- afebrile with normal vital signs/physical exam
- started on a soy formula and intramuscular antibiotics due to difficulty maintaining intravenous access
- During hospitalization, her gastrointestinal symptoms resolved, however she developed hypoxemia with pulse oximetry in the high 80s.

## Physical Exam

At the time of desaturation, the patient remained overall well appearing. Pulmonary exam was significant only for mildly labored breathing with subcostal retractions, lungs were otherwise clear to auscultation. **Integumentary exam showed no cyanosis of lips, skin, or mucous membranes.** Neurologic and cardiac exams were age appropriate and she appeared to be well hydrated.

## Diagnostic Evaluation

A full septic workup, including complete blood count, complete metabolic panel, inflammatory markers, cerebral spinal fluid studies, blood cultures and a chest xray revealed no bacterial or viral infection as a source for this patient's symptoms. **Arterial blood gas revealed a methemoglobin level of > 28 (Fig 2.), which was initially attributed to lidocaine within intramuscular antibiotics.** Despite discontinuation of antibiotics and initiation of IV methylene blue, rebound methemoglobin levels remained elevated.

# CASE

## Diagnosis and Treatment

Further genetic testing revealed diagnoses of congenital methemoglobinemia, glucose-6-phosphate-dehydrogenase (G6PD) deficiency carrier state and galactosemia carrier state. Methylene blue is contraindicated, and was thus discontinued. Transition to daily ascorbic acid provided normalization of methemoglobinemia.

**Important Note:** Methylene blue is contraindicated in patients with G6PD deficiency. Because methylene blue requires G6PD to function, it will be ineffective in patients suffering from a G6PD-deficiency. In some cases, methylene blue administration may even cause hemolysis in these patients.

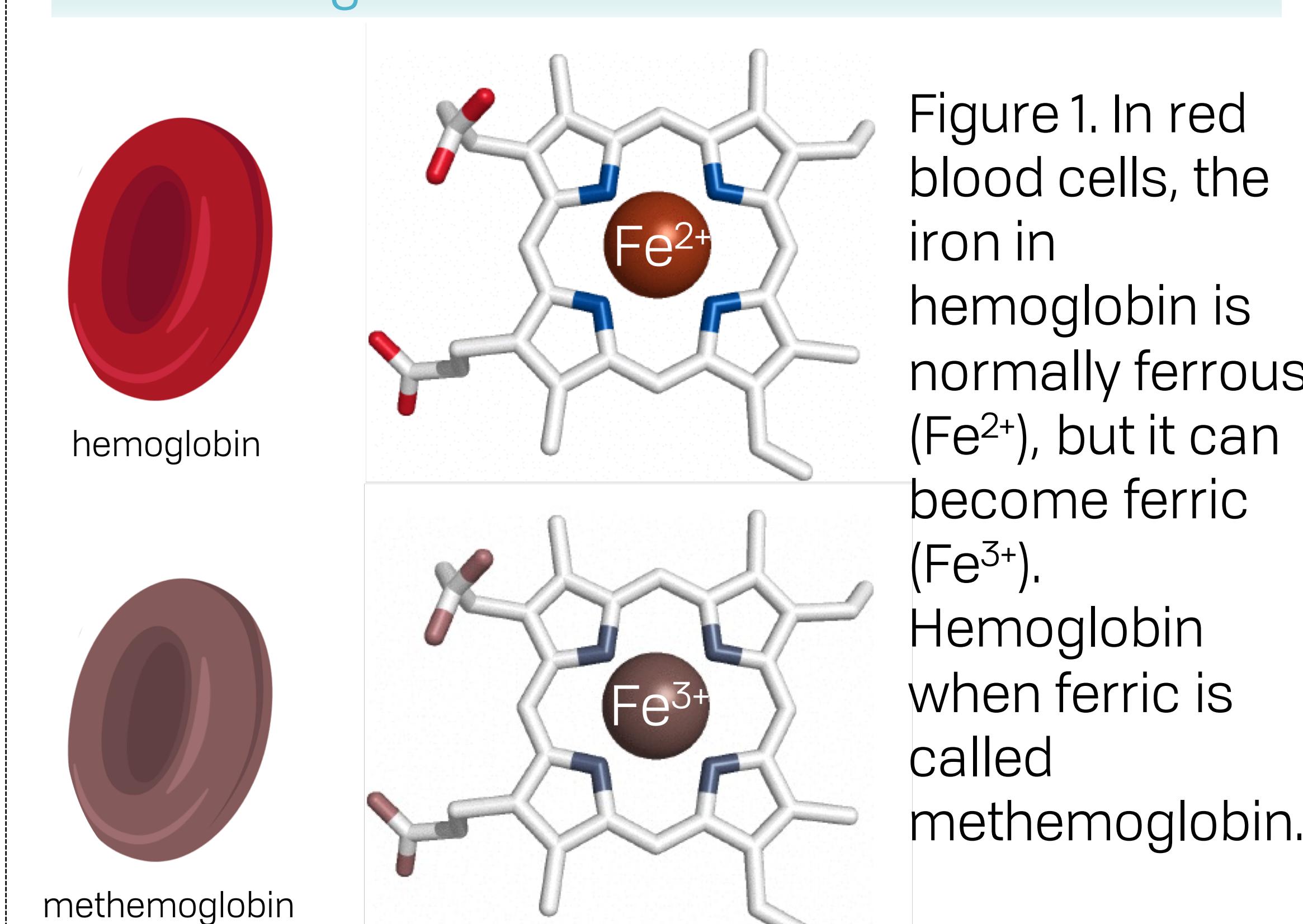
## Discussion

Congenital methemoglobinemia is rare enough that no incidence or prevalence has been reported. Most of the literature consists of case reports in which a patient initially presents with cyanosis, usually secondary to an inciting medication such as lidocaine, nitrates or antibiotic, and is subsequently found to have methemoglobinemia. The case above highlights the importance of maintaining a broad differential for infants presenting with respiratory distress and hypoxemia without cyanosis, symptoms frequently observed during acute illness of the pediatric patient.

Once methemoglobinemia is part of the differential, it is especially important to consider the congenital variant for patients' who have refractory methemoglobinemia despite treatment with IV methylene blue.

# BACKGROUND

## Methemoglobin



## Typical Clinical Manifestation (fMetHb)

fMetHb (%)	Signs and Symptoms
< 3 (normal)	None
3 - 15	Frequently none; greyish skin
15-30	Occasionally none; cyanosis, chocolate-brown blood
30-50	Dyspnea, fatigue, $\text{SpO}_2 \sim 85\%$

50+	Tachypnea, metabolic acidosis, seizure, CNS depression, coma, death
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Figure 2. Potentially non-cyanotic methemoglobinemia levels are shown in blue.

## References

- 1 "Clinical Features, Diagnosis, and Treatment of Methemoglobinemia." UpToDate, 2018,
- 2 Theodore, A. (2018). Oxygenation and mechanisms of hypoxemia.
- 3 Prchal, Josef T. "Clinical Features, Diagnosis, and Treatment of Methemoglobinemia." UpToDate, 2018