

Why so blue? A case of neonatal cyanosis due to congenital methaemoglobinaemia (HbM Iwate)

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Accepted 14 November 2016

DESCRIPTION

A 2.18 kg male baby was born by elective caesarean section at 34+3 weeks to a primiparous mother with autosomal dominant congenital methaemoglobinaemia (HbM Iwate) and gestational diabetes. Having been asymptomatic throughout her life, she developed significant respiratory symptoms in the third trimester, possibly due to a superimposed acquired methaemoglobinaemia, which necessitated hospitalisation, red cell exchange and early delivery of her infant.

At birth, the baby remained cyanosed despite good respiratory effort, and congenital methaemoglobinaemia was presumed. However, he quickly developed moderate respiratory distress (presumably unrelated) and was managed with facial continuous positive airway pressure (CPAP) in the delivery room, demonstrating maximal preductal saturations of 73% in 100% oxygen.

In the neonatal unit, he was started on nasal high-flow therapy. Urgent echocardiography excluded heart disease, and chest X-ray was unremarkable. Saturation monitoring was deemed unreliable; therefore, respiratory support was weaned (and stopped within 48 hours) based on regular normal capillary or arterial PaO₂ values and improving respiratory parameters. A co-oximeter, which uses multiple-wavelength spectrophotometry to measure methaemoglobin non-invasively, would

have been helpful. He remained clinically well despite a persistently dusky complexion and characteristically thick, chocolate-coloured blood (figure 1).

Feeds were established, and his care was continued as standard for a 34-week gestation infant, with outpatient paediatric haematology follow-up. DNA sequencing confirmed mutation in the α 2globin chain (α (F8)His->Tyr), diagnostic of HbM Iwate. Minimal clinical sequelae are anticipated.

Congenital methaemoglobinaemia is a rare but recognised cause of neonatal pseudocyanosis¹⁻³ requiring prompt consideration and a pragmatic approach to management when monitoring challenges and genuine respiratory disease coexist.

Learning points

- ▶ Congenital methaemoglobinaemia is a rare cause of visible neonatal cyanosis with low saturations on pulse oximetry but normal PaO₂ on blood gas analysis.
- ▶ The clinical picture can be complicated by premature delivery and/or respiratory disease.
- ▶ Access to a co-oximeter would allow accurate non-invasive monitoring in these infants and avoid unnecessary investigation and intervention.

Contributors JSM planned and drafted this case report. SD and FO'B edited the report. The literature review was performed by SD and JSM. JSM and FO'B revised the manuscript.

Competing interests None declared.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

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Figure 1 Image of a well but cyanosed preterm infant with monitor illustrating his cardiac and respiratory parameters.



To cite: McGrath JS, Datir S, O'Brien F. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2016-216805

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