Name of Trainee: Jodi Forward (jodi.forward@yale.edu)
Name of Mentor: Mary Jane Hogan (maryjane.hogan@yale.edu)
Type of Trainee: Pediatric Resident

Title: A Perplexing Case of Hypoxia.
Forward, JA\(^1\), Hogan, MJ\(^1\).
\(^1\) Yale New Haven Children’s Hospital

Background: Hypoxia in children may result from acute or chronic respiratory, cardiovascular, or hematologic conditions. We report on a healthy child who received an extensive evaluation for low oxygen saturation by pulse oximetry. A multidisciplinary, patient-centered approach to family history taking may have expedited his diagnosis due to genetic testing delays.

Case Description: A 4-year-old boy with a history of allergies presented to the emergency department after possible exposure to peanut butter. He had facial swelling, no apparent respiratory distress, and a pulse oximetry reading (SpO\(_2\)) of 85%. He received intramuscular epinephrine, intravenous dexamethasone, and was placed on 10 L/min of oxygen via non-rebreather without improvement in SpO\(_2\). Inpatient evaluation included normal EKG, normal echocardiogram, and a chest computed tomography that revealed mild bronchiectasis in the right upper lobe. Gastric lavage was negative for tuberculosis. An arterial blood gas in room air was normal. Normal complete blood count, carboxyhemoglobin, hemoglobin electrophoresis, and oxygen dissociation curve revealed no evidence of a low oxygen affinity hemoglobin variant. Inpatient genetic testing was not covered by the child’s health insurance. Outpatient results 2 months after discharge revealed Hemoglobin Lansing, a congenital hemoglobin variant that may interfere with standard pulse oximetry by having different spectral properties. Home oxygen was discontinued and a medical alert card was given to the family. Mother reported that father also has low SpO\(_2\) pulse oximetry readings.

Conclusion: The limitations of pulse oximetry need to be recognized by clinicians when confronted with a well-appearing child who may have an undiagnosed inherited hemoglobin variant. While hemoglobin electrophoresis is a necessary test in the evaluation of discrepant pulse oximetry readings, a negative result does not disprove a hemoglobin variant. Genetic testing should ideally be available while inpatient to expedite diagnosis and prevent unnecessary interventions.

Word Count: 291