**A close up of a person

Description automatically generatedThe Happy Blue Baby**

By: Shuchismita Dutta1 and Kasandra Riley2

1Institute of Quantitative Biomedicine, Rutgers University, Piscataway, NJ 08854

2Department of Chemistry Rollins College, Winter Park, FL 32789

**Part 1: *A “happy blue baby”?***

A full-term female infant was born to a 20-year-old woman in New Jersey in 2008. The infant was described as a “happy blue baby” — that is, cyanotic but well appearing. Cyanosis is the result of poor circulation or inadequate oxygenation of the blood caused by one of many conditions. The patient’s initial hemoglobin oxygen saturation, measured in ambient air with the use of pulse oximetry, was 30 to 50% (normal >95%). After intubation and delivery of 100% oxygen, hemoglobin saturation fluctuated around 85%. The physical examination revealed only cyanosis and moderate hepatomegaly. The infant was extubated, with a transition to oxygen delivery by means of nasal cannula. She was clinically well, although hemoglobin oxygen saturation remained below normal, at 80 to 90%, despite the absence of evidence of arterial hypoxia. Laboratory data were notable only for moderate anemia with reticulocytosis, an elevation in red blood cell counts commonly seen in anemic patients. The results of chest radiography and echocardiography were normal.

1. How does hemoglobin delivery of oxygen work in a healthy newborn? Summarize your findings in 3-4 sentences.

|  |
| --- |
|  |

1. Sketch a single graph with possible oxygen-hemoglobin dissociation curves representing three conditions: (1) a red line for a healthy infant, (2) a blue line for the happy blue baby’s initial hemoglobin oxygen concentration, (3) a black line for the baby’s hemoglobin saturation after intubation. Paste a sketch or JPG of your image here:

**Part 2: *Does the happy blue baby express fetal hemoglobin?***

On the first day of life, electrophoresis performed with cellulose acetate and agarose gel showed that total hemoglobin consisted of approximately 90% hemoglobin F and 10% hemoglobin A, with no bands of varying molecular weight.

1. Thinking carefully about how oxygen delivery by hemoglobin works in a newborn, develop unrelated, testable hypotheses that might explain this patient’s condition if (A) she is cyanotic throughout her entire life, (B) she recovers completely from cyanosis in 1-2 months.

|  |
| --- |
| (A) |
| (B) |

1. If you were the baby’s clinician, how would you explain to this patient’s family the alternate possibilities that their baby’s cyanosis could either clear up in a couple of months or be with her the rest of her life?

|  |
| --- |
|  |

1. If the family asked about genetic testing, who would you take samples from? What sequences would you test? What assumptions are you making in your approach?

|  |
| --- |
|  |