**Happy Blue Baby**

Case Study Pre-Work MCQ for large classes

[multiple answer]

1. Based on the information in the article, the baby girl has a genetic mutation causing the hypoxia. Which family members likely have the gene for this genetic mutation? Select ALL answers that apply.

The infant’s mother

The infant’s father

A paternal grandparent

A maternal grandparent

All future children the infant later gives birth to

[fill in multiple blanks]

2. The doctors found a single nucleotide mutation in the Toms River Baby’s fetal hemoglobin gene. The mutation changed the codon 67 sequence from GTG to ATG. Consult the genetic code (<https://www.genome.gov/genetics-glossary/Genetic-Code>) to describe the mutation:

Give the full name of the amino acid corresponding to the original (wild type) codon

 Valine

Give the full name of the amino acid corresponding to the mutation

 Methionine

By convention for indicating mutations using one-letter amino acid codes, how would this amino acid mutation be abbreviated:

 V67M

[multiple answer]

3. Is the mutated residue side chain similar to or different from that found in the native protein? From the list below, choose all terms that describe the size and physicochemical properties that describe the wild type residue 67:

Polar

Nonpolar

Charged

Beta-branched

Aromatic

Relatively small

Relatively large

[multiple answer]

4. Is the mutated residue side chain similar to or different from that found in the native protein? From the list below, choose all terms that describe the size and physicochemical properties that describe the mutated residue 67:

Polar

Nonpolar

Charged

Beta-branched

Aromatic

Relatively small

Relatively large

*Box 1: Resource*

RCSB Protein Data Bank (**RCSB PDB**, [www.rcsb.org](http://www.rcsb.org)) provides access to 3D structural data of biological macromolecules (proteins, nucleic acids, carbohydrates and their various complexes). In addition, it provides information about the experiment used to derive the data, details about the molecules included in the experiment, and links to various bioinformatics resources that can provide additional information about the protein/molecule of interest. Each structure in the PDB is identified by a unique identifier (called PDB ID). Atomic coordinates form the PDB can be visualized and analyzed using various visualization software (some available from RCSB PDB).

To explore the molecular bases of the newborn’s cyanosis, search the Protein Data Bank (at [www.rcsb.org](http://www.rcsb.org)) for structures of this mutant protein. You can start your search using the protein name or other details that you know. (Hint: search by the name of the mutation, or the mutation itself (e.g. X##Y, where X is the original amino acid, ## is the position of that amino acid in the protein chain, and Y is the mutated amino acid). Examine the search results and refine them as necessary.

5. Provide the 4-digit alphanumerical PDB ID that corresponds to this mutated protein.

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