

## Questions for 2009 ISLH Hemoglobinopathy Workshop.

Question Case 1. Single best answer.

- A. Hb Korle Bu and delta chain variant.
- B. G-Philadelphia trait.
- C. Heterozygous G-Philadelphia and homozygous  $\alpha^+$  thalassemia.
- D. Double heterozygous G-Philadelphia and beta thalassemia.
- E. Double heterozygous G-Philadelphia and heterozygous  $\alpha^+$  thalassemia.

Question Case 2. Single best answer.

- A. Homozygous D-Punjab (D-Los Angeles).
- B. Compound heterozygous D-Punjab (D-Los Angeles) and beta zero thalassemia.
- C. Compound heterozygous D-Punjab (D-Los Angeles) and beta plus thalassemia.
- D. Compound heterozygous Hb S and D-Punjab (D-Los Angeles).
- E. Double heterozygous D-Punjab (D-Los Angeles) and alpha thalassemia.

Question Case 3. Single best answer.

- A. Beta thalassemia trait. Promoter mutation.
- B. Hemoglobin E trait
- C. Hemoglobin C trait
- D. Alpha globin variant co-eluting with A2 on HPLC
- E. Gamma chain variant.

Question Case 4. Single best answer.

- A. The patient is lying about her transfusion history
- B. Double heterozygous for HbC and alpha thalassemia.
- C. Double heterozygous HbC and deletional hereditary persistence of fetal hemoglobin (HPFH).
- D. Double heterozygous HbC and nondeletional hereditary persistence of fetal hemoglobin (HPFH).
- E. Double heterozygous HbC and delta beta thalassemia

Question Case 5. Single best answer.

- A. Heterozygous  $\beta$ -globin variant (trait)
- B. Homozygous Hb G-Philadelphia
- C. Homozygous single alpha gene deletion and heterozygous Hb G-Philadelphia
- D. Double heterozygous single alpha gene deletion and Hb G-Philadelphia
- E. Hb AD (trait)

Question Case 6. Single best answer.

- A. Homozygous for gamma chain variant.
- B. Beta thalassemia major
- C. HbH disease in a newborn
- D. Barts Hydrops fetalis
- E. Both parents carry alpha+ thalassemia

Question Case 7. Single best answer.

- A. Alpha thalassemia trait
- B. Delta thalassemia trait
- C. Beta thalassemia trait
- D. Delta beta thalassemia
- E. Double heterozygous for beta thalassemia and delta thalassemia

Question Case 8. Single best answer.

- A. Beta thalassemia trait and unstable alpha globin variant
- B. Beta thalassemia trait and gamma chain variant
- C. Beta thalassemia trait and passively acquired HbE through transfusion.
- D. Beta thalassemia trait and delta chain variant
- E. None of the above.

Question Case 9. Single best answer.

- A. Baby is not at risk for a hemoglobinopathy.
- B. Elevated A2 is borderline and of no significance as there is no microcytosis.
- C. A2 is elevated because the patient also has alpha thalassemia.
- D. Elevated A2 is an artifact.
- E. Baby is at risk for beta thalassemia intermedia.

Question Case 10. Single best answer.

- A. Homozygous delta beta thalassemia.
- B. Double heterozygous for beta thalassemia and delta beta thalassemia.
- C. Double heterozygous for beta zero thalassemia and deletional hereditary persistence of fetal hemoglobin.
- D. Double heterozygous for delta beta thalassemia and deletional hereditary persistence of fetal hemoglobin.
- E. Homozygous deletional hereditary persistence of fetal hemoglobin.