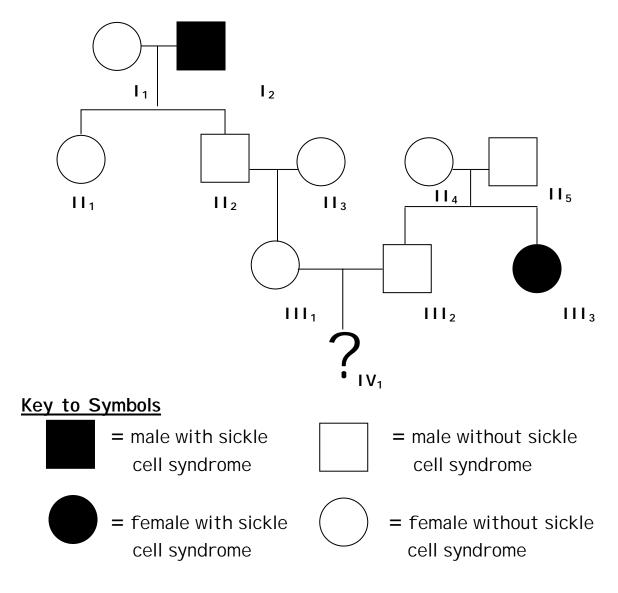
Chances Are? Scenario B: While we're away



You are employed as a genetic counselor at a clinic in Boston. Your clients, Kim and Scott (III $_1$ and III $_2$), have spoken with you about the risks of passing on Sickle Cell Syndrome to their children as both Kim and Scott have a family history of sickle cell syndrome. Even though both of them are perfectly healthy, they are concerned that their child will have the disease. You counseled them on the potential risks and informed them about the possibility of having blood tests to better assess the risk.

Kim and Scott decided to weigh their options while on vacation visiting relatives in Vietnam.

Today you receive a frantic e-mail from the vacationing couple in which they inform you that Kim is two months pregnant. You review the family history to discover which of the couple's blood relatives have sickle cell syndrome. This is shown in the pedigree above.

Given this history <u>only</u>, what are the chances that their child will have sickle cell syndrome? Is there any uncertainty in your prediction?

In Kim and Scott's e-mail, they inquire about the possibility of doing this blood test on one or more of their relatives.

Unfortunately, Kim and Scott spent most of the savings on this once in a lifetime trip to Kim's homeland. Additionally, the insurance company will only cover half the cost of the test.

Which adult or adults from the pedigree would you test? Given the cost of the test, you must justify the necessity of each test that you perform to a Medical Insurance Company (MIC).

The blood test for sickle cell syndrome is based on protein electrophoresis of hemoglobin. This test can be performed at a cost of \$970.00 per test for any adult(s) that you choose from the pedigree above. It is not recommended for fetal blood tests due to high risk to mother and fetus.

Justification to MIC for sickle cell hemoglobin tests:

Gel Electrophoresis:

Use the polyacrylamide gel electrophoresis apparatus and the hemoglobin isolated from the blood of the family members to perform your proposed diagnostic tests. Available to you will be hemoglobin protein samples that serve as controls for your test. List types of control hemoglobin proteins that you wish to run alongside the test samples.

Results from "Possible" Protein Gel Electrophoresis Tests

Adult Tested	Type of Hemoglobin
I ₁	
I ₂	
Π_1	
$ II_2 $	
113	
114	
115	
$\Pi\Pi_1$	
$ III_2 $	
1113	

 $\frac{\text{Genotype}}{\text{Hb}^{A}/\text{Hb}^{A}} = \text{normal adult hemoglobin}$

Hb^A/Hb^s = carrier adult hemoglobin

Hb^s/Hb^s = sickle cell syndrome hemoglobin

Conclusions:

Based on the results of the hemoglobin tests, what are the chances that the child will have the disease sickle cell syndrome? What are the chances that the child will be a carrier of sickle cell syndrome?