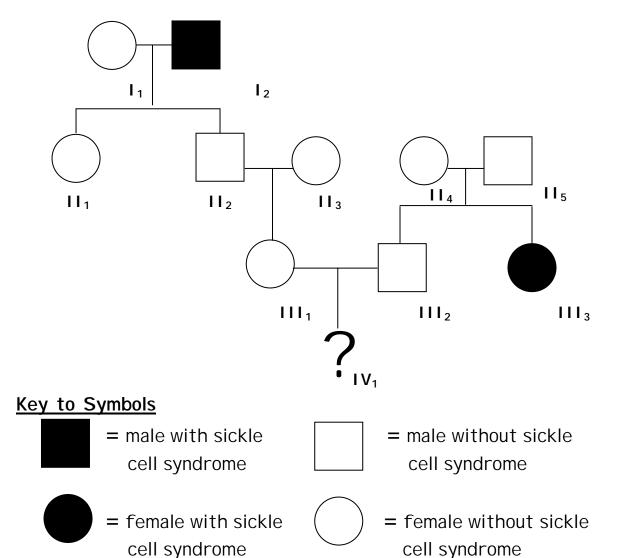
Chances Are? Scenario A: The Family Plan



You are employed as a genetic counselor at a clinic in Boston. Kim and Scott (III_1 and III_2) arrive in your office for a prenatal appointment. They are expecting their first child in 7 months. 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 do a brief 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?

A blood test for sickle cell syndrome based on protein electrophoresis of hemoglobin is available. 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.

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.

Justification 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	
12	
Π_1	
Π_2	
113	
114	
115	
$\Pi\Pi_1$	
1112	
1113	

Genotype Type of Hemoglobin

Hb^A/Hb^A = 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?