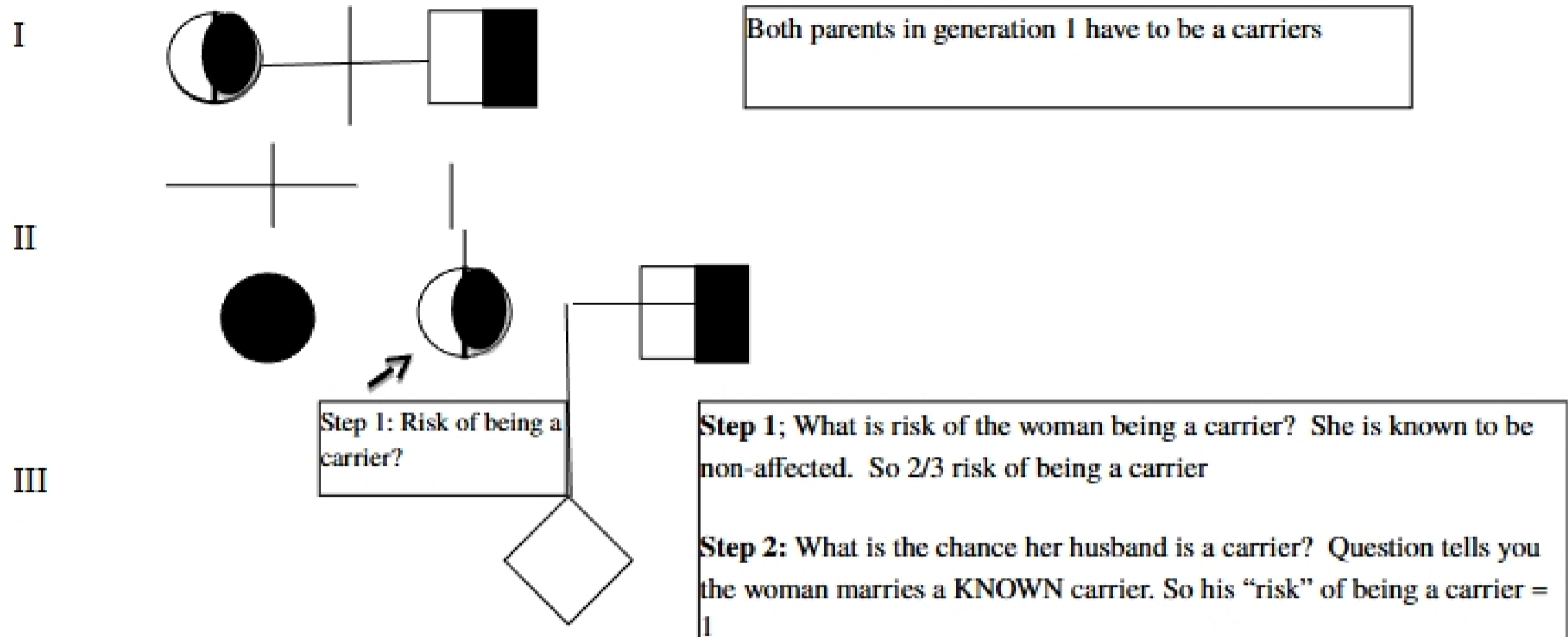


ANSWERS to Practice problems 2013:

1. e - autosomal dominant as it involves affecteds in 3 generations with transmission thru males and females (too easy for the quiz!)
2. b - head MRI; congenital arthrogryposes such as this child has should make one think of abnormal neurologic system, abnormal muscle system or limited intrauterine space. Of the choices, a brain MRI such as the example in class would show brain dysplasia (maybe a little too hard for the quiz)
3. d. - If you were **not** told the child was unaffected, the answer would be $\frac{1}{2}$ since the probability of two carrier parents having a carrier offspring is 50%: there are four possible offspring: affected, carrier from mom passing gene, carrier from dad passing gene, non-carrier. But in this question we eliminated the option of being affected, so of the remaining 3 types of offspring, 2 are carriers – thus, $\frac{2}{3}$ is the answer. *NOTE: This concept may be incorporated into a question on the quiz or exam that will require you to go beyond this and incorporate this risk further in a pedigree to calculate the risk of having an affected child.*
4. d
5. a
6. d. One of the things we wanted you to get out of the patient session was the patient-doctor relationship. That is hard to test, but so important for you to witness. (You should also know the information about Down's syndrome that Dr. Andersson talked about before Harry and his sister came in.)
7. e – Opps, you have not yet had the lecture on intrauterine infections – you don't need to know this! This got left in from previous years. Sorry!
8. b. FISH. Next generation sequencing could get you the answer, but it is not the logical test to order: why order an expensive test that gives you way more information than you need. You want targeted testing for something in which you have a diagnosis in mind. So FISH is the most logical to order. Microarray would be another decent choice (still more expensive than needed) – but that wasn't one of the choices you had to choose from.
9. d. You should be aware that the other options do not have structural abnormalities.
10. e – too easy, but isn't she cute?
11. e All the alternative answers are good terms to make sure you understand

Additional question:

12. b **See explanation below**



Finally calculation=
 $\frac{2}{3}$ (prob consultant is carrier) x 1 (husband is carrier) x $\frac{1}{4}$ (prob affected child) = $\frac{1}{6}$

Step 2: Risk of having an affected child = risk of woman being a carrier ($\frac{1}{2}$) AND her husband being a carrier (1) and both passing on gene ($\frac{1}{4}$ for AR disorder)
 SO = $\frac{1}{2} \times 1 \times \frac{1}{4} = \frac{1}{8}$