

STUDENT PROBLEM-BASED LEARNING SESSION
DEPARTMENT OF OBSTETRICS AND GYNECOLOGY
LOYOLA UNIVERSITY MEDICAL CENTER

TOPIC: Prenatal Genetic Counseling and Diagnosis

RECOMMENDED READING ASSIGNMENT:

Embryology, Anatomy and Reproductive Genetics, Chapter 3 in *Obstetrics and Gynecology for Medical Students*, 3rd Edition, Beckmann et al, pp31-54, 1998.

CASE PRESENTATION:

Ms. Chance Genetics is a 38 y.o. G3 P1011 Caucasian female, from Downs, Illinois. Her LMP was approximately 15 weeks ago. You have seen her for her yearly routine examinations for the last 4 years and this was a planned pregnancy. This is her second prenatal visit; her first was about 5 weeks ago. You inquire about her current pregnancy history and learn that she drinks one glass of wine each day (she switched from one glass of brandy when she learned of her pregnancy). When you take her family history, you learn she has had one abortion 10 years ago and she and her husband have a healthy 7 y.o. son. Her husband, Mr. D.N.A. Risk is 42 years old and in good health. Ms. Genetics has two healthy brothers and a sister who is learning disabled. Her mother's brother is said to be mentally retarded. Mr. Risk's sister died in early childhood from Cystic Fibrosis. Mr. Risk is of Greek, Jewish, and German descent, while Ms. Genetics is of Jewish, Italian, and Danish descent.

- (1) Draw the pedigree.
- (2) A) List the disorders that this couple is at increased risk for.
B) Next to this list, explain what percentages of risk exist for each disorder (at this point).
- (3) While you're working on this, Ms. Genetics mentions that she and her husband terminated her first pregnancy due to the diagnosis of anencephaly in the fetus. Modify the pedigree to incorporate this additional information. Does this change your risk assessment at all, and if so, how?

[Over]

Carrier Testing

(4) What kind of carrier testing would you offer the patient? How would you present these options?

(5) The patient and her family elect to pursue the following carrier tests. The results, completed after 5 weeks, are as follows:

Mr. Risk:

Hexosaminidase A = 62% (WNL)

DNA Analysis for Tay-Sach's disease = No mutations detected

DNA Analysis for Canavan's disease = No mutations detected

CF DNA analysis = No mutations detected

Ms. Genetics:

Chromosome analysis = 46,XX

DNA analysis for Fragile X = Normal

CBC = WNL

Hem. Electrophoresis: Hb A = 98%

Hb A₂ = 2% (WNL)

CF DNA analysis = No mutations detected

Ms. Genetics' Sister:

Chromosome analysis = 46,XX

DNA analysis for Fragile X = 300 CGG repeats ("full mutation")

Mr. Risk's Father:

CF DNA analysis – deltaF508 positive

Mr. Risk's Mother:

CF DNA analysis - deltaF508 positive

(6) Were any of these carrier tests unnecessary? What are some other issues that might make these tests impractical?

(7) How do these carrier test results modify the percentages calculated in question (2)B)?

(8) What risks remain and what tests would you offer the patient?

Prenatal Diagnosis

(9) At this point, Ms. Genetics elects to have an amniocentesis. After a 10 day waiting period the results are:

Karyotype = 46,XX

Amniotic Fluid AFP = 0.6 M.O.M.

Targeted ultrasound = No detectable abnormalities

(10) What's wrong with starting a genetics consultation at 15 weeks gestation?

(11) What referral sources are available for such a patient?