Medical Genetics and Dysmorphology

1. A three-year-old girl presents for evaluation of recurrent pneumonia (five times in two years) and chronic diarrhea. She is at the fifth percentile for height and weight. What are your differential diagnoses? Construct a family pedigree as if her aunt (mother's sister) and cousin (mother's brother's child) have the same condition.

2. A newborn infant has prominent epicanthal folds, small ears, hypotonia, short, broad hands and feet, brachycephaly, and a heart murmur. The mother notes that the baby "looks different" than her two previous children. How would you evaluate this infant and counsel the mother?

3. The parents of a two-year-old boy with developmental delay report a history of mental retardation in several male members of their family. What are your differential diagnoses and what diagnostic screening would you recommend?

4. A 16 year-old girl presents with primary amenorrhea. She has been doing well at school but is not athletic and her height is less than the 5% for age. On physical examination, she has redundant neck skin and broadly spaced nipples. How would you evaluate and counsel this her?

5. The mother of a newborn tells you she has taken phenytoin throughout her pregnancy and wants to know what effect this may have on her baby. How would you counsel the mother?

6. You are asked to evaluate a baby in the nursery who is small for gestational age and microcephalic. How would you evaluate the infant? What questions would be important to ask the mother?

7. A mother of a two-year-old child with sickle cell disease is pregnant and wants to know the likelihood that this child will have sickle cell disease. How would you counsel her?

8. The pregnant mother of one of your patients calls to say that a prenatal ultrasound revealed that her fetus might have spina bifida. She wants to know the implications of the disease. How would you counsel her? Should any special precautions be taken at the time of delivery and in neonatal period?