



# POSNA

## The Core Curriculum

### Prenatal testing

#### Objectives

1. Describe currently available methods used to obtain samples or images for prenatal testing
2. List 2 disorders of orthopaedic interest which can be detected with prenatal testing

#### Discussion points

1. Should prenatal testing be under legislative control?

#### Discussion

Since the 1960's amniotic fluid has been cytogenetically analyzed. Amniocentesis consists of aspirating amniotic fluid, which is largely fetal urine. Cells are separated by centrifugation and analyzed. A wide variety of chromosomal disorders (Down syndrome), neural tube defects (myelomeningocele), metabolic defects (homocystinuria, Lesch-Nyhan syndrome, X-linked muscular dystrophy, Charcot-Marie-Tooth) can be detected by prenatal testing. Amniocentesis is generally not performed until the 16th week of pregnancy, due to the small amount of amniotic fluid before that time. There is a small but definite risk associated with amniocentesis. Chorionic villus sampling, analyzing tissue which forms part of the placenta, can be performed at 9-12 weeks. Ultrasound and fetoscopy provide anatomic information, but fetoscopy carries a 2-5% risk of spontaneous abortion. In vitro fertilization allows testing of a single cell at the 6-10 cell stage of the preimplantation embryo. The embryo can then be implanted in vivo. This field is rapidly growing, and there is a journal specifically devoted to prenatal diagnosis. Prenatal screening is generally restricted to populations at special risk (advanced parental age, child with neural tube defect, etc.). Issues related to prenatal testing are sensitive, and have been the subject of intense and emotional debate in the United States.

#### References

1. Asch A. Prenatal diagnosis and selective abortion: a challenge to practice and policy [see comments]. *American Journal of Public Health* 1999;89(11):1649-57.
2. Brambati B. Prenatal diagnosis of genetic diseases. *European Journal of Obstetrics, Gynecology, & Reproductive Biology* 2000;90(2):165-9.
3. Drugan A, Johnson MP, Evans MI. Ultrasound screening for fetal chromosome anomalies. *American Journal of Medical Genetics* 2000;90(2):98-107.

4. Kashork CD, Chen KS, Lupski JR, Shaffer LG. Prenatal diagnosis of Charcot-Marie-Tooth disease type 1A. *Annals of the New York Academy of Sciences* 1999;883:457-9.
5. Kent-First M. The critical and expanding role of genetics in assisted reproduction. *Prenatal Diagnosis* 2000;20(7):536-51.
6. Kuppermann M, Nease RF, Learman LA, Gates E, Blumberg B, Washington AE. Procedure-related miscarriages and Down syndrome-affected births: implications for prenatal testing based on women's preferences. *Obstetrics & Gynecology* 2000;96(4):511-6.
7. Singer E, Corning AD, Antonucci T. Attitudes toward genetic testing and fetal diagnosis, 1990-1996. *Journal of Health & Social Behavior* 1999;40(4):429-45.