Genetic Testing Options

Parents: (This can be done prior to or during the early part of pregnancy)

- **No testing** (other than the standard Cystic Fibrosis or Ashkenazi Jewish Panel, if clinically appropriate)

- **Carrier screening**---PAMF offers the Counsyl Foresight test which screens for carrier status of over 175 conditions. Please go to the following website address to obtain additional information: http://www.counsyl.com/foresight.

Baby:

1. **No testing** (If the testing will not change your management of the pregnancy---i.e. you would not terminate or do further testing to see if there is a problem, you would not want to know for preparation purposes, etc…) You would still be getting a 20 week anatomy ultrasound.

2. **Screening options** (non-invasive and with no risk of miscarriage)

   Standard screening options (Generally covered by insurance regardless of your age):

   - **Full serum integrated screen**---(First trimester blood test, transabdominal nuchal translucency U/S, and 2nd trimester blood test); Detects 90% of Down's syndrome; 81% Trisomy 18, 80 to 90% of neural tube/abdominal wall defects, 60% of Smith Lemli Opitz Syndrome.

   - **Serum integrated screen**---(1st and 2nd trimester blood tests); Detects 85% of Down's syndrome and the same percentages for the above listed conditions

   - **Quad marker screening**--- (2nd trimester blood test); Detects 80% of Down's syndrome and the same percentages for the above listed conditions.

   - **NOTE**: 1st trimester blood test is at 10 to 13 6/7 wks, Nuchal translucency U/S on baby at 11 2/7 to 14 2/7 wks, and 2nd trimester blood test is at 15 to 20 wks.

New screening option (generally covered only by select insurance plans, if initial genetic screening is abnormal or if patient is 35 or older at time of delivery. There is a discounted self pay rate---please refer to the web link.):

- **NIPS** (non-invasive prenatal screening; previously known as NIPT or cell free DNA testing)---one blood test anytime after 10 wks=>Detection rate of 99% for Down's Syndrome, 99% for Trisomy 18, 99% for Trisomy 13, sex chromosome abnormalities and microdeletions. Please go the following website link to get more information: http://www.counsyl.com/prelude.

3. **Diagnostic options** (Invasive, carries small risk of miscarriage, better detection rate, and generally covered by insurance if patient is 35 or older at time of delivery or if initial screening test is abnormal)

- **CVS—Chorionic Villus Sampling**---done between 10 to 13+ weeks---a high risk OB MD inserts a sterile catheter through the abdomen or the vagina under ultrasound guidance to obtain a sample of placental cells; The genetic material in the cells is grown and all 46 chromosomes of the fetus are examined.

- **Amniocentesis**---Done after 15 to 16 weeks---a high risk OB MD inserts a sterile needle through the abdomen under ultrasound guidance to obtain a sample of cells. The genetic material in the cells is grown and all 46 chromosomes of the fetus are examined.