Prenatal Screening, Testing and Diagnosis, and Perinatal Palliative Care

In the first and/or the second trimester of pregnancy a combination of maternal blood or serum tests and fetal ultrasound may detect the possibility of a fetus having trisomy 21 or trisomy 18. When a possibility of trisomy 13 is suspected, it is usually through ultrasound findings and sometimes might be detected through a first trimester blood test called pregnancy associated plasma protein (PAPP). (Best, 2006)

A **negative result** of screening **does not exclude the possibility of a trisomy condition** but it does mean there **is no increased risk** detected for a possible trisomy condition from that particular test. Unless there are other risk factors such as maternal age or prior pregnancy with a genetic disorder, further testing is usually not recommended.

A **positive result from the screening does not mean that a fetus has a trisomy condition.** It does mean there is an **increased risk detected for a possible** trisomy condition and further diagnostic testing is warranted, but follow-up tests are invasive. Diagnostic tests (*cytogenetic testing*) study the genetic make-up of cells in amniotic fluid obtained through amniocentesis, or tissue from chorionic villus sampling (CVS), or fetal blood. Some parents choose not to do follow-up invasive testing due to a possible 1% or less risk to the fetus and/or personal beliefs.

New, reliable **non-invasive** prenatal testing **(NIPT)** was marketed by four companies in 2012**. NIPT** can detect a trisomy condition with a single blood draw from the expectant mother as early as the tenth week of pregnancy. It is very appealing as optional follow-up invasive testing (e.g. amniocentesis) is not necessarily needed, and results are obtained earlier than with other tests. NIPT looks at cell-free DNA fragments (cfDNA) from the fetus, circulating in the mother's blood and can detect trisomy 21, 18 or 13 with higher accuracy than the traditional maternal serum screening. Studies find NIPT is effective with a false positive rate (calling the result a trisomy 18 or 13 when it was not) of less than 1%. (Carey, JC 2013, the SOFT Times newsletter, http://trisomy.org/?page_id=7344/)

There is a **lack of agreement** within medicine as to whether NIPT is a **screening or diagnostic test** and to whom it should be offered; all pregnant women or only to women at high risk of fetal aneuploidy (abnormal number of chromosomes). A pretest explanation of NIPT benefits and limitations and the fact that **testing for chromosomal abnormality is optional** should be provided to the expectant woman**.** If a physician views this test as only a screening and not a diagnosis, invasive testing will likely be suggested for confirmation of a positive result or a parent may request further testing.

Genetic amniocentesis and chorionic villus sampling (CVS) are the only tests that can confirm a trisomy or other chromosomal problems. (*Preparing for the Arrival of your Baby,* Berg et al., 2015; free E-book at http://www.internationaltrisomyalliance.com/)

Another recently approved genetic test that can detect trisomy conditions and more, **Chromosomal Microarray (CMA),** is done using samples obtained during amniocentesis or chorionic villus sampling. CMA looks at submicroscopic changes within a chromosome, finding information not routinely identifiable by karyotype and gives more information than NIPT, including findings of unknown significance that can create anxiety in a parent. (Stokowski and Klugman, 2013) CMA might help with the diagnosis of some rare genetic conditions.

For those not diagnosed prenatally:
Karyotype has been in use since the sixties and remains useful today for trisomy 18 and 13. A chromosome analysis is performed with chromosomes of a single cell arranged in a specific order and photographed. Many SOFT families have a photo of their child's karyotype. (http://www.geneticseducation.nhs.uk/laboratory-process-and-testing-techniques/karyotyping). When more rapid results are needed, a test called fluorescence in situ

hybridization (**FISH**) is available. (http://www.rarechromo.org/information/Other/FISH%20FTNW.pdf)

Genetic counseling is recommended for all women with "at risk pregnancies." If a diagnosis of trisomy 18 or 13 is identified before 24 weeks of pregnancy, the provider will discuss the option of termination. Approximately 90% of trisomy 18 or 13 pregnancies are terminated in Europe and 75% in the USA. For those who choose to continue their pregnancy, referral to Perinatal Palliative Care (PPC) is occurring more often. Participation in these programs is optional. PPC offers guidance for creating a birth plan, end of life wishes and care options/decisions if the infant is born alive. All PPC programs encourage comfort care but interventions for new born infants with a life-limiting diagnosis vary by region and team. If it is important, a parent needs to ask about interventions and surgeries allowed, particularly if considering a possible cardiac repair. PPC services are consultative and can be done by phone.

www.prenatalpartnersforlife.org **was founded by Mary Kellett, mother of Peter who had trisomy 18. This organization provides support information and encouragement for carrying to term with an adverse prenatal diagnosis and support for raising your child with special needs after birth.**

http://perinatalhospice.org/Perinatal_hospices.html **Provides a list of Perinatal hospice/palliative care programs and support in the USA and Internationally. These programs are a relatively new concept of care for parents who choose to continue a pregnancy after learning their expected baby has a life-limiting diagnosis.**

Wiipedia link for cell free DNA screening: http://en.wikipedia.org/wiki/Cell-free_fetal_DNA
Wikipedia link for traditional prenatal screening: http://en.wikipedia.org/wiki/Prenatal_diagnosis

Excerpts from ***Care of the Infant and Child with Trisomy 18 and Trisomy 13*** [Barnes, Carey 2014] free E-book at www.trisomy.org were provided by co-author, Ann Barnes.