Client Handout:
Genetic Screening/Testing

Pineapple Babies Birth Services

Genetic screening/testing can be a hard topic to discuss. First, I want to explain the difference between screening and testing. Screening – tells you that you have the markers that indicate there could be a genetic difference with your baby. Testing – can confirm with confidence that your baby has a genetic condition. Often times, you will need several tests (after an abnormal screening) to get a positive confirmation of a genetic anomaly. It is important that you have an idea of what you will do with the information you receive should you choose to have screenings or tests done. It is also important to understand that one screening or a combination of a screening and a test alone may not be enough to diagnose an anomaly. You should be prepared to commit to all screenings, tests, and ultrasound measurements to have a meaningful risk assessment.

This handout will contain several examples of screenings and tests that may be offered or you may wish to request. All screenings are not right for everyone. Making a decision about whether to go through genetic testing is intensely private and often emotional. Your midwife can assist you in determining how you wish to proceed.

Reasons you may consider Genetic testing…

- If you have a family history of a specific condition or you have delivered a baby with a genetic condition
- If you would consider termination for certain genetic conditions
- If you just want to know
- If you have a disease or condition, knowing may help you (and your provider) determine what medication and dosage is right for you.
- If you have some of the risk factors for a specific condition

Here are some Risk Factors…

- General:
  - Family history of birth defects or congenital anomaly
  - Alcohol consumption during pregnancy
  - Smoking during pregnancy
  - Drug use
  - Maternal age of 35 years or older
- Taking certain medications
- Having uncontrolled diabetes before or during pregnancy, obesity

Having one or more of these risk factors does not mean you will have a baby affected by a birth defect, it just means that you have a risk factor. Knowing you have a risk factor or more might mean that you will want to consider genetic testing.

What now?

Now you can make the decision as to whether genetic screening/testing may be right for you and your family. If you desire genetic testing be sure to let your midwife know. If you should choose not to undergo any genetic screenings or tests, or if you choose to just do specific ones your midwife will have a document for you to sign stating that you do not desire genetic screening/testing at this time. It will also mention that you understand that some tests must be done at certain times in your pregnancy for accuracy. Should you reconsider genetic testing after signing this form you will need to speak with your midwife as soon as possible to access the screening/testing you desire or alternatives if available.

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Screening/Test	What Can Be Detected	Accuracy	Risks
CVS (Test) When: 10-12 weeks (16 weeks for RhD-) *Ultrasound w/placental tissue sample	Chromosomal abnormalities, various genetic conditions. Does NOT test for Neural tube defects.	96% accurate	Damage to embryo, miscarriage, cervical lacerations, hemorrhage, and infection.
Nuchal Translucency (Screening) When: 11-14 weeks *Ultrasound	Chromosomal abnormalities, cardiac conditions.	70-80% alone, 80-90% accurate when combined with first trimester screening.	Ultrasound is generally accepted as low risk.
MSAFP (Screening) Maternal Serum Alpha-fetoprotein When: 16-18 weeks *Maternal blood draw	Open neural tube defects, fatal Trisomies 21 and 18, and Smith-Limil-Opitz syndrome.	10% screen abnormal. Of those 2-3:100 babies born with a birth defect.	Risk from site of blood draw.
Anatomy Ultrasound (Screening) When: 18-22 weeks *Ultrasound	Physical abnormalities suggestive of possible conditions.	Very accurate, dependent upon skilled technician and radiologist.	Ultrasound is generally accepted as low risk.
Amniocentesis (Test) When: 12-18 weeks *Ultrasound w/amniotic fluid aspiration	Chromosomal abnormalities, neural tube defects, and genetic disorders	98-99% accurate	Damage to fetus, miscarriage, cervical lacerations, hemorrhage, and infection.

Other tests may be available upon request.

As you can see, there are many options for prenatal screening and testing. These are just a few of the most popular tools used in this area. If there are specific things you are interested in screening/testing for please contact your midwife. Many genetic screenings/tests are costly and often insurances will not cover those costs. Please be aware of what your insurance will and will not cover when considering genetic tests. Also, if you have a history of or have delivered a baby with a genetic condition in the past, you may be able to request special consideration from your insurance provider.

References

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