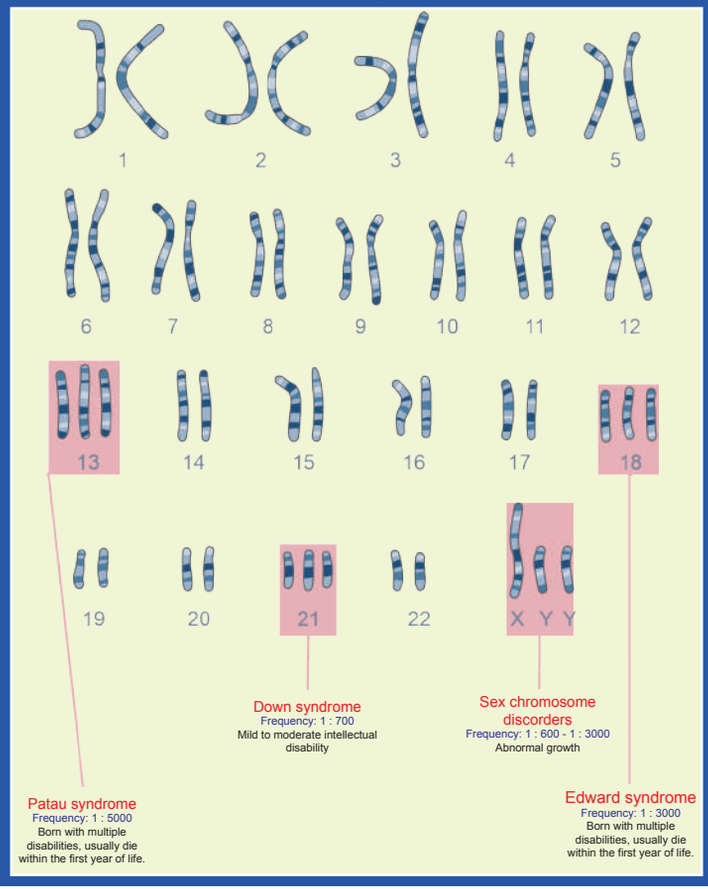
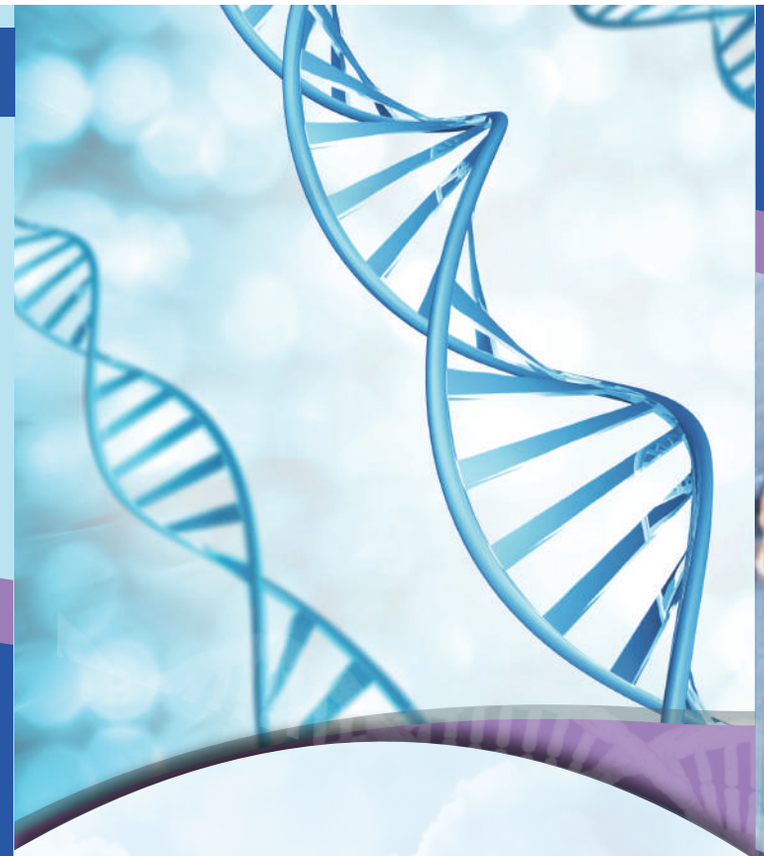
Chromosomal disorders

What are chromosomes and chromosomal discorders?

Chromosomes contain the genetic information that tells our cells how to grow and function. Normally, babies get one set of 23 chromosomes from each parent, for a total of 46, but in some situations, a developing baby may have extra or missing chromosomes, or pieces of chromosomes. These chromosome abnormalities usually occur sporadically, however can impact the baby's health.

Trisomy disoreders include:





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Non-invasive Prenatal Screening (NIPS)

American O&G College Recommended[1,2]

"Offer NIPS to all pregnant women"

International Accreditations :

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Clinical Laboratory Improvements Amendments

What is Non-invasive prental screening?

Non-invasive prenatal screening (NIPS) is a genetic test that you can take as early as 10 weeks into your pregnancy to screen for specific chromosomal abnormalities that can impact the health of your baby. NIPS can also determine your baby's sex earlier than ultrasound.

What chromosomal changes does Groken NIPS look for?

- **Trisomy screening** checks if there are any extra copies of specific chromsomes. The most common example of trisomy is Down Syndrome, Also known as Trisomy 21.
- **Microdeletion analysis** checks if there are any missing sections, or deletions, of specific chromosomes. Microdeletions, like DiGeorge syndrome, are relatively rare.
- **Sex chromosome analysis** checks for extra or missing X or Y chromosomes, which are the chromosomes that determine your baby's sex. Sex chromosome analysis can also help determine your baby's sex.

Trisomies screened	Microdeletions screened*	Sex chromosome disorders screened**
Down syndrome Trisomy 21	1p36 deletion syndrome	Turner syndrome Monosomy X
Edwards syndrome Trisomy 18	DiGeorge syndrome 22q11.2 deletion syndrome	Triple X syndrome 47,XXX
Patau syndrome Trisomy 13	Angelman syndrome/ Prader-Willi syndrome 15q11.2 deletion syndrome	Klinefelter syndrome 47,XXY
	Cri du Chat syndrome 5p15.2 deletion syndrome	Jacob's syndrome 47,XYY
	Wolf-Hirschhorn syndrome 4p16.3 deletion syndrome	

*Microdeletion analysis is not avaiable for twin pregnancies.
**Sex chromosome analysis for twins can tell you if you are carrying at least one male baby. However, it is unable to determine if there is more than one male or identify which twin is male.

Is NIPS right for me?

NIPS is the earliest screening test for chromosomal disorders and an early opportunity to understand potential risks to your baby. The American College of Obstetricians and Gynecologists recommends all women should be offered the option of aneuploidy screening for fetal disorders regardless of maternal age.

What do I need to know?

NIPS is:

- safe, with **no increase risk of miscarriage**
- non-invasive: tests are performed using a small sample of your blood
- a screening test, not a diagnostic test: that means it can only provide an estimate of risk, not a definitive answer
- not able to test for all possible chromosomal abnormalities: NIPS looks for the most common, medically impactful conditions
- fast: after the lab receives your sample, results will be ready in **5 to 7 working days** on average

What will my results tell me?

Most women discover that their pregnancy is at low risk for a chromosomal condition. If your screening test contains a positive result, your doctor will discuss what your results mean and will provide you with options for what to do next.

Negative
A negative result indicates that your pregnancy is not at an increased risk for the disorders screened. Continue to work with your doctor, who may recommend other types of testing throughout pregnancy.

Positive
A positive result indicates that your pregnancy may be at an invreased risk for a specific chromosome abnormality. Diagnostic testing, via chorionic villus sampling (CVS) or amniocentesis, is recommended for confirmation of NIPS results.

Groken NIPS + Carrier Screening

- Carrier screening is another type of genetic testing that helps identify if you and your partner are carriers of a genetic disorder that you can pass on to your child, even if you do not have the disorder yourself.
- While NIPS looks at your baby's chromosomes, carrier screening looks for smaller types of genetic changes in you and your partner.
- The American College of Obstetrics and Gynecologists **recommends carrier screening for all pregnant women**, regardless of age or risk.[2]
- Your doctor may recommend carrier screening along with NIPS for a more comprehensive look at your baby's genetic health in early pregnancy.

1. Practice Bulletin No. 163: Screening for fetal aneuploidy. Obstet Gynecol. 2016;127(5):979-81.
2. Gregg AR, Skotko BG, Benkendorf JL et al. Noninvasive prenatal screening for fetal aneuploidy, 2016 update: a position statement of the American College of Medical Genetics and Genomics. Genet Med. 2016;18(10):1056-65