Prenatal Testing Information

Basic Biology

Our bodies are made up of trillions of cells.

Within each cell is our genetic material, or DNA.

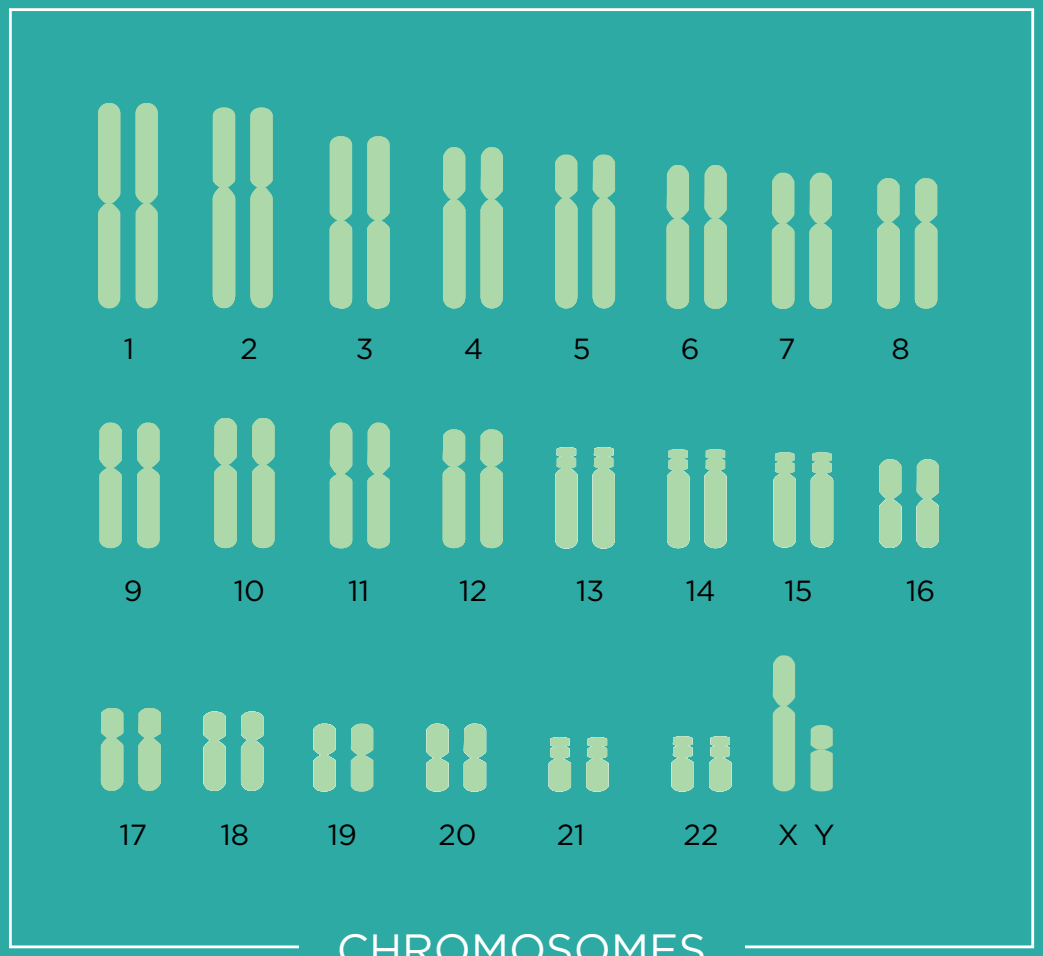
DNA is the instruction manual that tells our bodies how to grow and develop.

Basic Biology

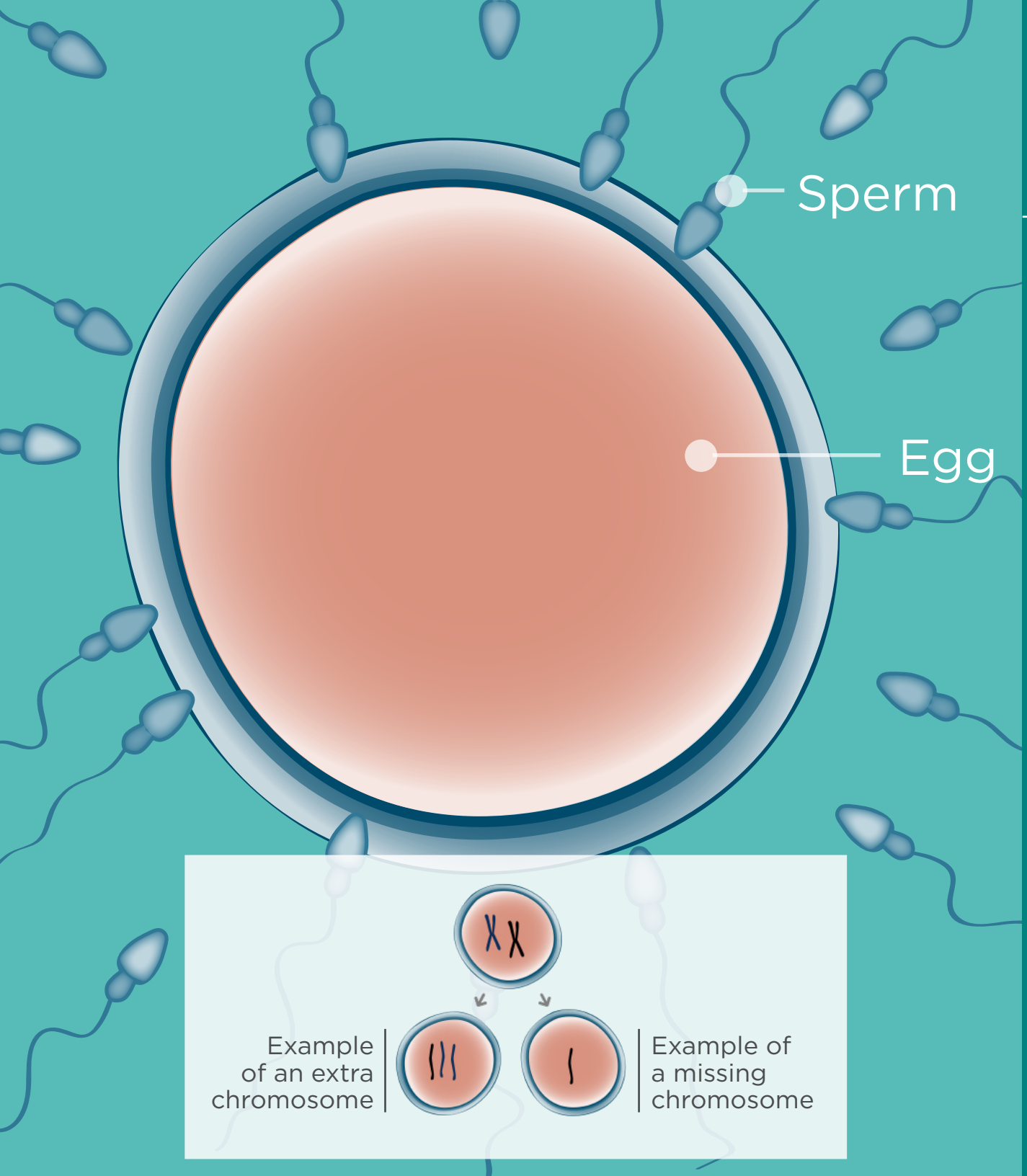
DNA is packaged into structures called chromosomes.

We inherit our chromosomes from our parents.

Humans typically have 46 chromosomes, occurring in 23 pairs. Females typically have two X chromosomes and males have one X and one Y chromosome.



CHROMOSOMES



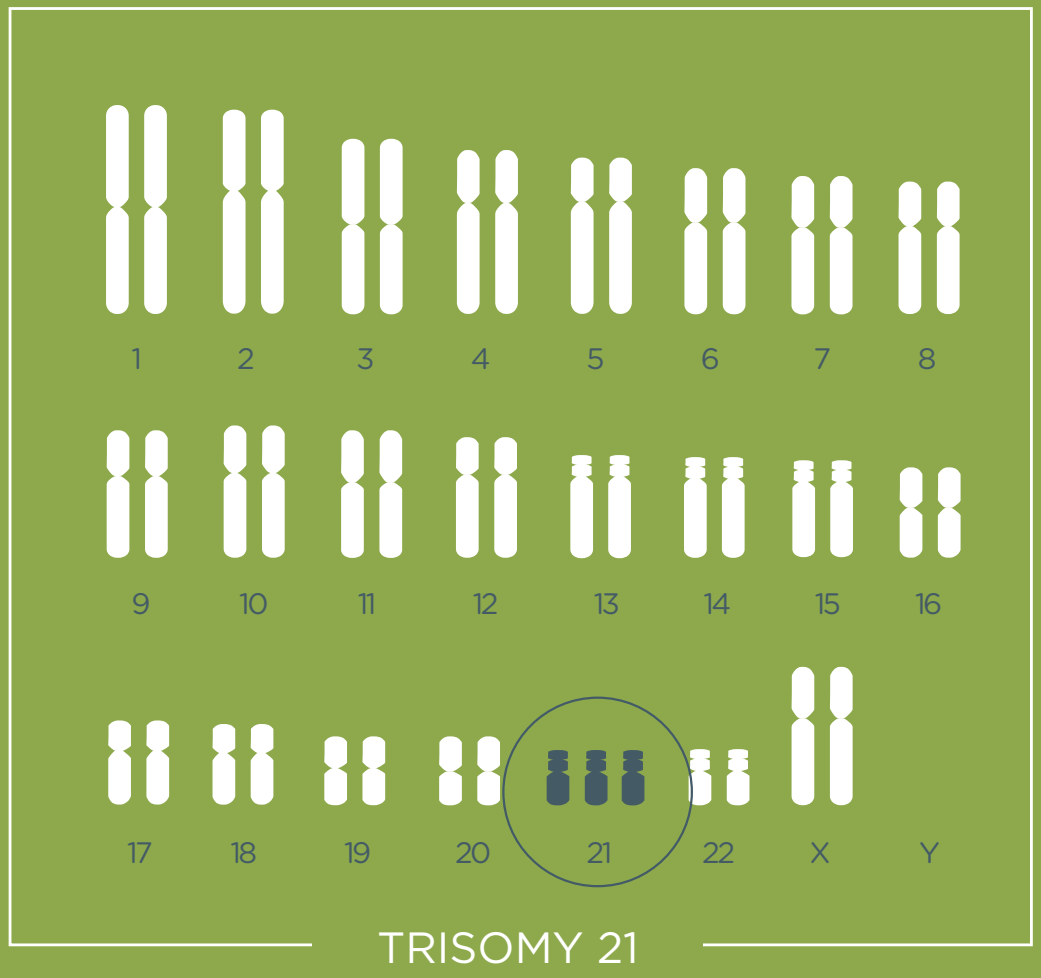
Basic Biology

Sometimes, an error can occur that leaves a sperm or egg cell with a missing or extra chromosome.

If that happens, the resulting pregnancy may also have a missing or extra chromosome.

Generally chromosome conditions are not inherited.

Chromosome Conditions



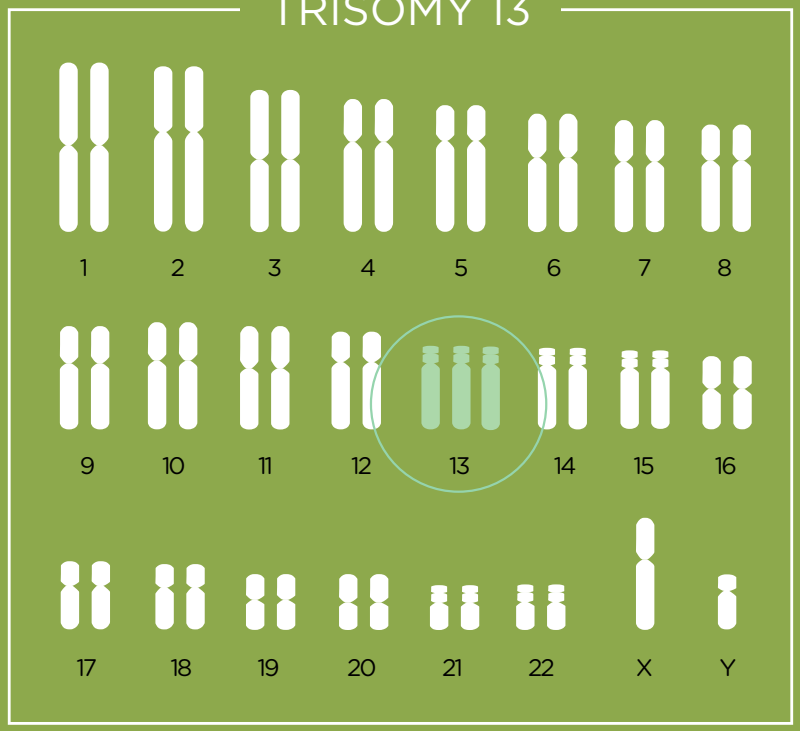
TRISOMY 21

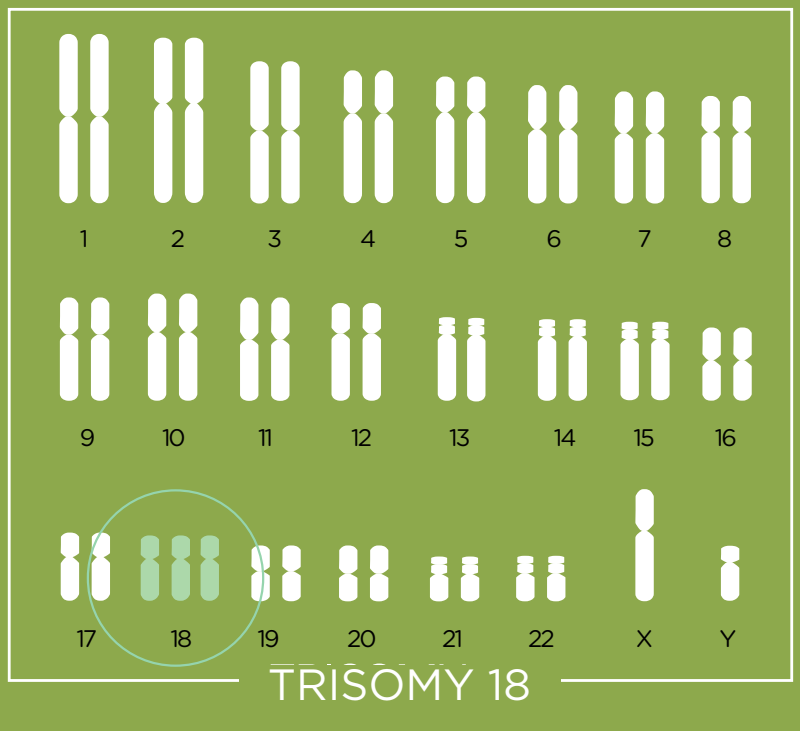
Trisomy 21:

Trisomy 21 refers to an extra copy of chromosome 21. Trisomy 21 causes Down syndrome.

Down syndrome is a condition that affects physical and mental development.[1]

1. http://www.nature.com/scitable/topicpage/trisomy-21-causes-down-syndrome-318



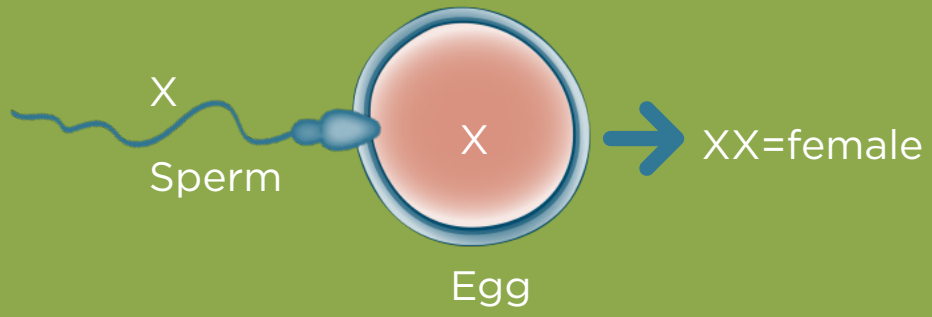


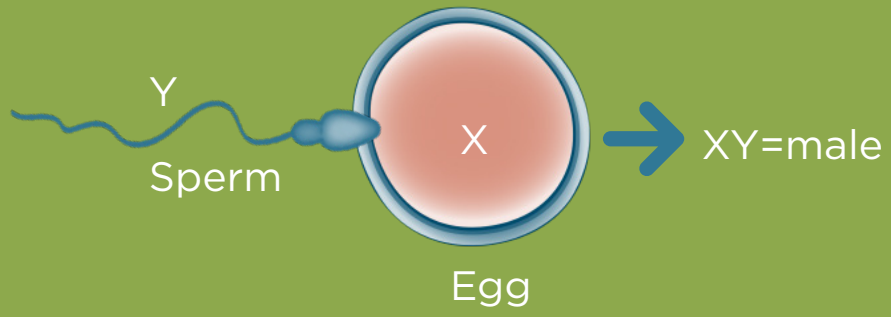
Chromosome Conditions

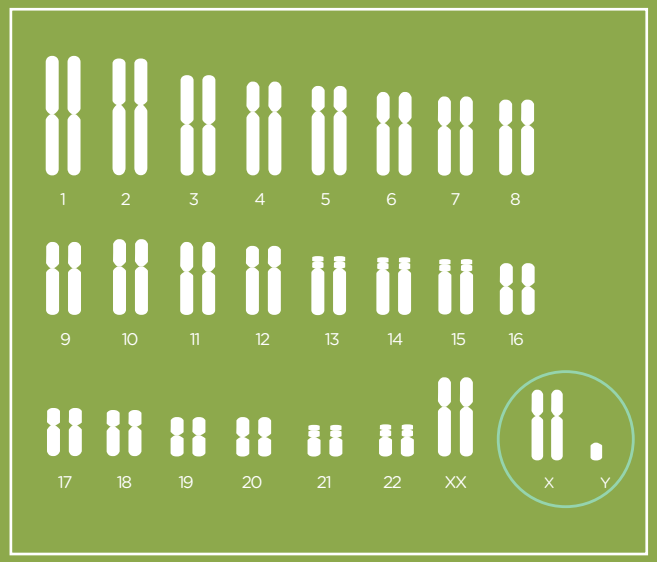
Trisomies 13 and 18:

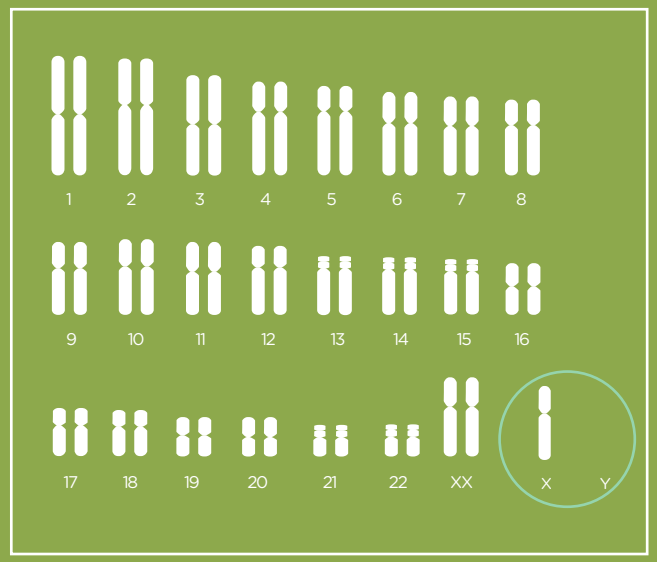
Trisomy 13 and trisomy 18 are less common than Down syndrome, but also involve an extra chromosome in the cells. Affects on development are so serious that many fetuses with these conditions will not survive.[1]

1. U.S. National Library of Medicine. Genetics Home Reference. Trisomy 13. https://ghr.nlm.nih.gov/condition/trisomy-13. Accessed April 25, 2018.









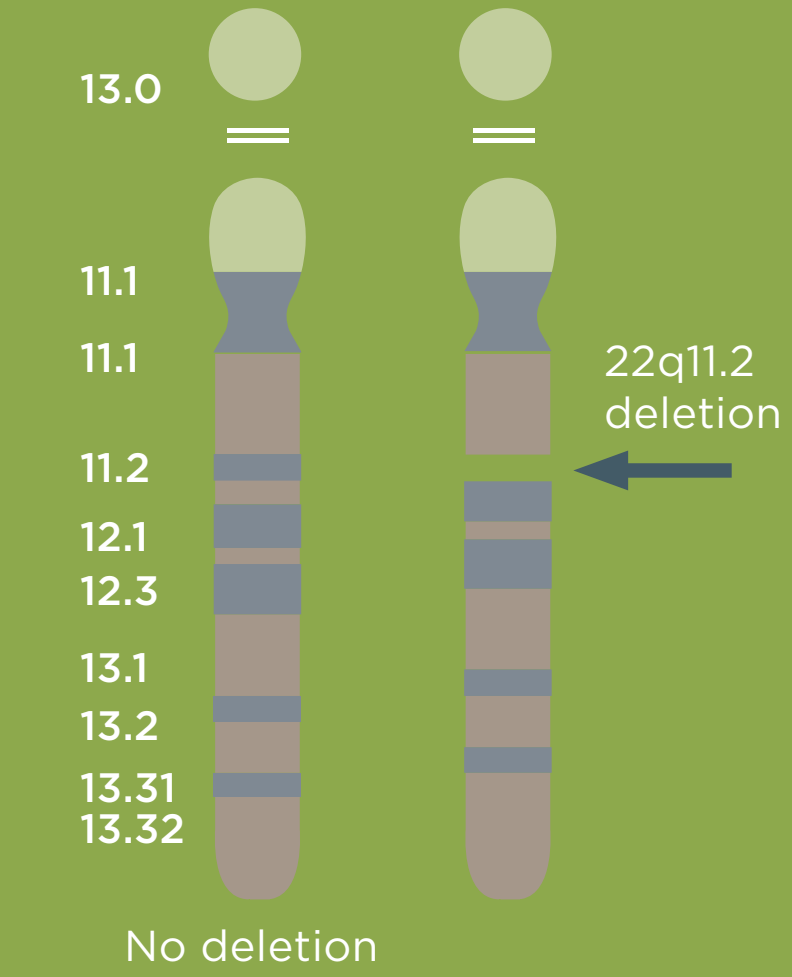
Chromosome Conditions

Chromosomes that determine whether we are male or female are labeled “X” and “Y” and are also called ‘sex chromosomes.’

Some people have a missing or an extra sex chromosome. These conditions are called “Sex chromosome aneuploidies, or SCAs”.

People with SCAs can have birth defects, infertility and learning differences. Some people with an SCA have such subtle features that the condition isn’t identified until after childhood.[1]

1. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1634840/



22q11.2 deletion syndrome

22q11.2 deletion syndrome is caused by a missing piece of chromosome 22. It occurs in close to 1 in 1,000 pregnancies.[1]

Children with 22q11.2 deletion may have heart defects, immune system problems, learning challenges, and specific facial features.[2,3]

Any pregnancy has a chance to have 22q11.2 deletion. Fetuses born with 22q11.2 deletion usually do not have a family history of the condition.[4]

Women of all ages have an equal chance to have a fetus with 22q11.2 deletion.[4]

1. Grati et al. Prenat Diagn. 2015 Aug;35(8):801-9.
2. McDonald-McGinn et al. Genet Couns. 1999;10(1):11-24.
3. Bassett et al. J Pediatr. 2011 Aug;159(2):332-9.
4. McDonald-McGinn et al. Genet Med. 2001 Jan-Feb;3(1):23-9.

Who is at risk?

Women of any age can have a fetus with a chromosome condition, but we know that the chance increases as women get older. Other factors that can increase the chance for a chromosome condition in a pregnancy are family history and things identified by prenatal ultrasound, such as a heart defect.

The American College of Obstetricians and Gynecologists (ACOG) and the Society for Maternal-Fetal Medicine (SMFM) recommend offering aneuploidy screening, or diagnostic testing early in pregnancy to all women regardless of maternal age.[1]

1. ACOG Practice Bulletin 163. Obstetrics & Gynecology May 2016 Vol 127 Issue 5 (e123–e137).

Traditional Screening

Traditional screening tests measure protein and hormone levels in a blood sample from the pregnant woman and estimate the chance that the developing fetus has a chromosome condition.

Because it is a blood test, there is no risk to the pregnancy.

The blood test is often combined with an ultrasound measurement, called the nuchal translucency (NT), to improve result accuracy.

This type of testing is generally restricted to certain periods of time during the pregnancy.[1]

1. ACOG Practice Bulletin 163. Obstetrics & Gynecology May 2016 Vol 127 Issue 5 (e123–e137).

TRADITIONAL SCREENING

Traditional Screening

The detection rate is the chance that the test will correctly identify a pregnancy that has one of these conditions.

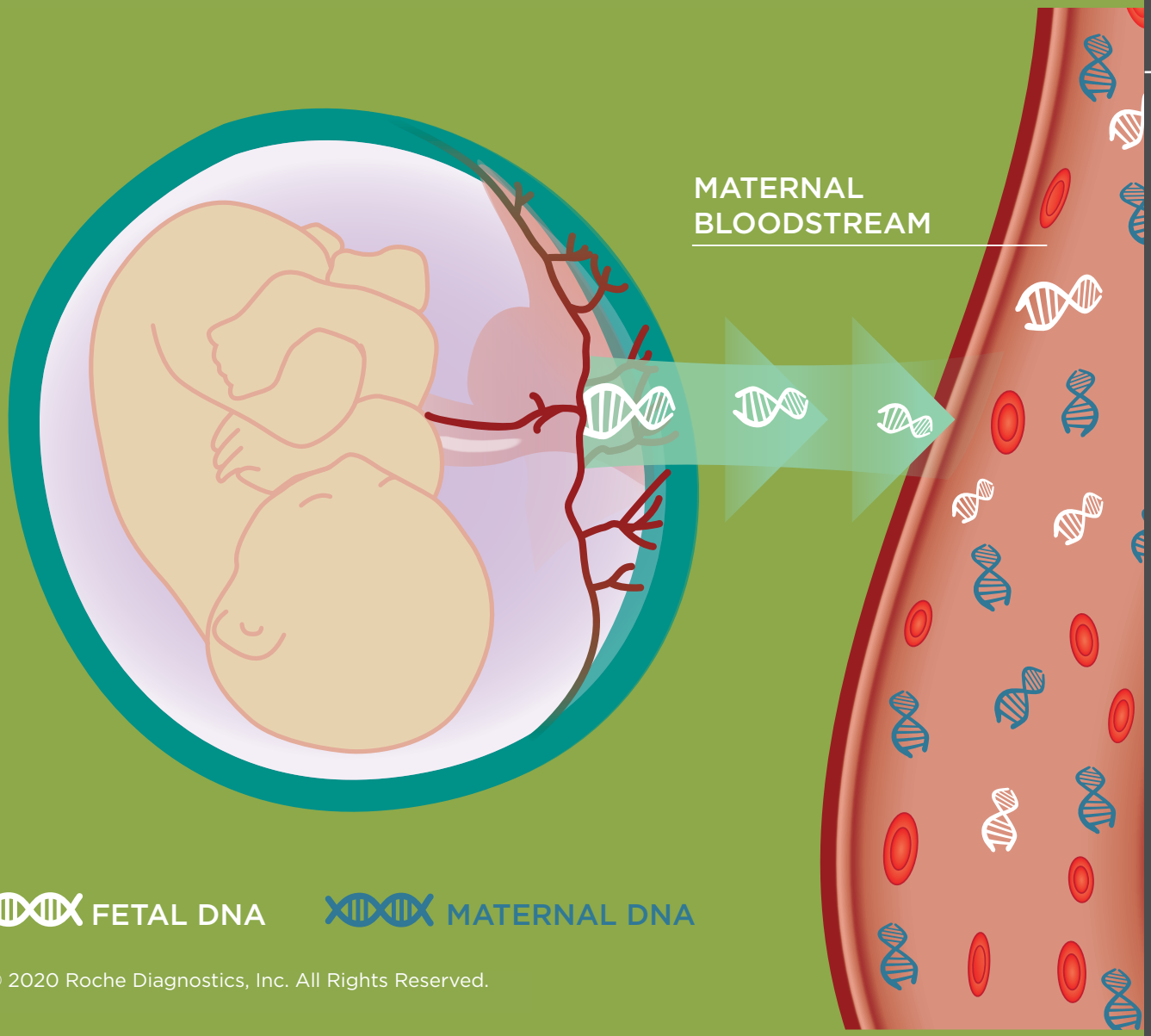
Detection rates with traditional screening may range from 69-96% for trisomy 21, depending on the method used.

Sometimes a test can indicate a high chance for a chromosome condition when the developing fetus does not have it. This is called a false positive result.

With traditional screening, about 1 in 20 women will receive a false positive result.[1]

1. ACOG Practice Bulletin 163. Obstetrics & Gynecology May 2016 Vol 127 Issue 5 (e123–e137).

harmony®



Harmony Prenatal Test

The Harmony blood screening test is for trisomy 21 (Down syndrome), trisomy 18, and trisomy 13. It can be performed as early as 10 weeks of pregnancy.

During pregnancy, the pregnant woman's blood contains fragments of the developing fetus's DNA. The Harmony blood screening test is a test that analyzes this DNA in a blood sample to predict the probability of trisomy 21 (Down syndrome), as well as trisomies 18 and 13.[1,2] These type of tests are also called cell-free DNA tests and or Non-invasive prenatal test (NIPT).

NIPT based on cell-free DNA analysis is a screening test.

The Harmony test can also screen for conditions caused by having an extra or missing copy of the X or Y chromosome, and 22q11.2 microdeletion.

Talk to your healthcare provider to understand which options are right for you.

HARMONY PRENATAL TEST

The Harmony non-invasive prenatal test is based on cell-free DNA analysis and is considered a prenatal screening test, not a diagnostic test. Harmony does not screen for potential chromosomal or genetic conditions other than those expressly identified in this document. All women should discuss their results with their healthcare provider who can recommend confirmatory, diagnostic testing where appropriate. The Harmony prenatal test was developed and its performance characteristics determined by Ariosa Diagnostics, Inc. a CLIA-certified and CAP-accredited clinical laboratory in San Jose, CA USA. This testing service has not been cleared or approved by the US Food and Drug Administration (FDA).

1. Stokowski et al. Prenat Diagn. 2015 Dec;35(12):1243-6.
2. Norton ME et al. N Engl J Med. 2015;372(17):1589-1597.

TRADITIONAL SCREENING:

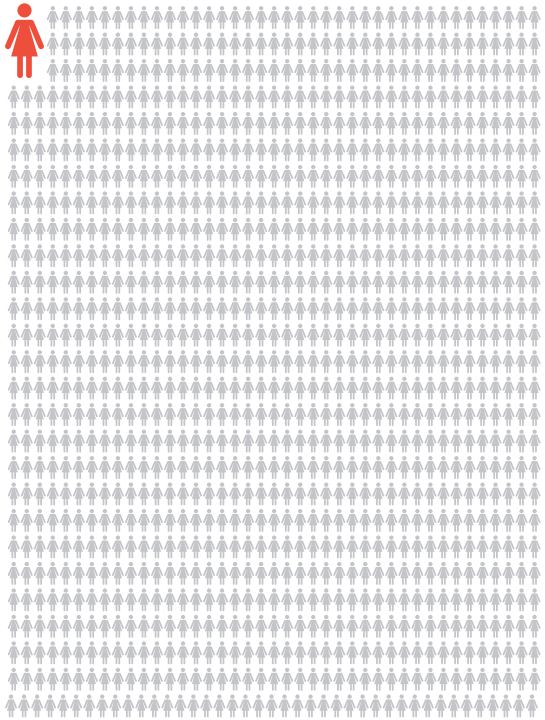
1 in 20

will have a false positive result

HARMONY PRENATAL TEST:

Less than 1 in 1600

will have a false positive result



The Harmony prenatal test is a screening test that is more accurate than traditional screening tests for trisomy 21 (Down syndrome).[2]

With NIPT, there is a lower chance that follow-up testing, such as amniocentesis, will need to be considered.[4]

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Harmony Prenatal Test

Clinical studies have demonstrated the Harmony test to have high detection rates, including >99% detection rate for Down syndrome.[1]

Less than 1 in 1,600 women will have a false positive test result with Harmony. This is at least 50 times lower than traditional screening tests.[2]

The Harmony test is more accurate than traditional screening tests.[2]

In a small percentage of women, a result from a non-invasive prenatal test may not be obtained. This is most often due to not enough fetal DNA within the maternal blood stream but can also be related to the general quality of information in the sample.[2]

Ensuring enough fetal DNA is an important quality metric so that your result is not based on maternal DNA alone.[3] Consult with your healthcare provider regarding all results.

1. Stokowski et al. Prenat Diagn. 2015 Dec;35(12):1243-6.
2. Norton ME et al. N Engl J Med. 2015;372(17):1589-1597.
3. Takoudes T, et al. Performance of non-invasive prenatal testing when fetal cell-free DNA is absent. Ultrasound Obstet Gynecol. 2015;45(1):112.
4. Wax et al. J Clin Ultrasound 2015 Jan; 43(1):1-6.

Screening Reminders

No screening test is designed to detect all possible conditions. Screening tests help identify pregnancies with an increased chance of certain chromosome conditions. With screening tests, false positive and false negative results can occur.

Women who have an increased chance on their screening test should consult a medical professional who can discuss follow-up confirmatory options.

Invasive Confirmatory Testing

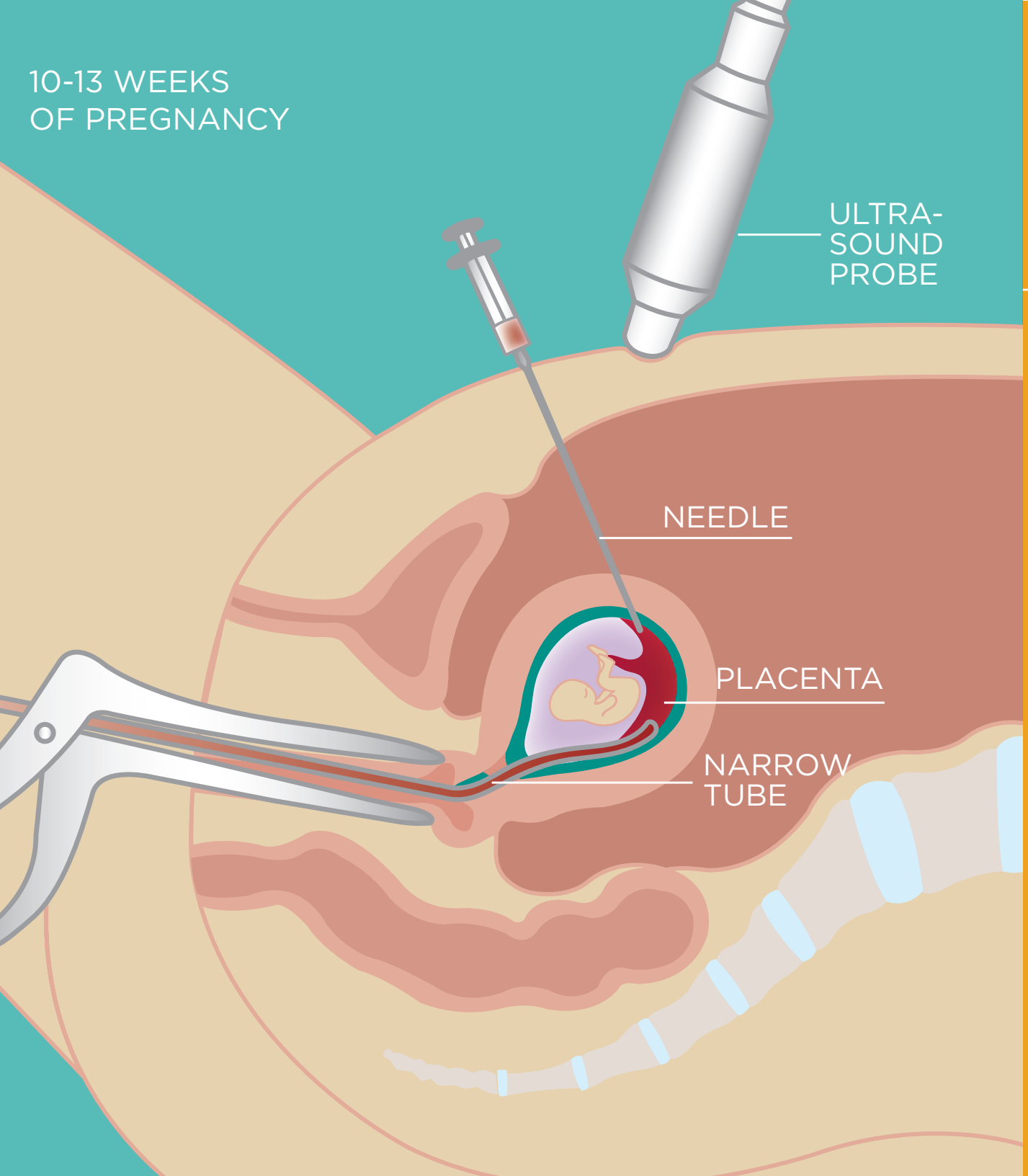
Invasive confirmatory testing involves a procedure to obtain cells from the pregnancy for testing.[1]

Invasive confirmatory testing also known as ‘diagnostic testing’ can give a definitive answer about most chromosome conditions in a pregnancy.

It is most often performed for pregnancies at increased chance for a chromosome condition.

There are two types of invasive procedures, chorionic villus sampling and amniocentesis.

1. ACOG website: http://www.acog.org/Patients/FAQs/Diagnostic-Tests-for-Birth-Defects (082516)



Chorionic Villus Sampling (CVS)[1]

CVS can be done between 10-13 weeks of pregnancy.

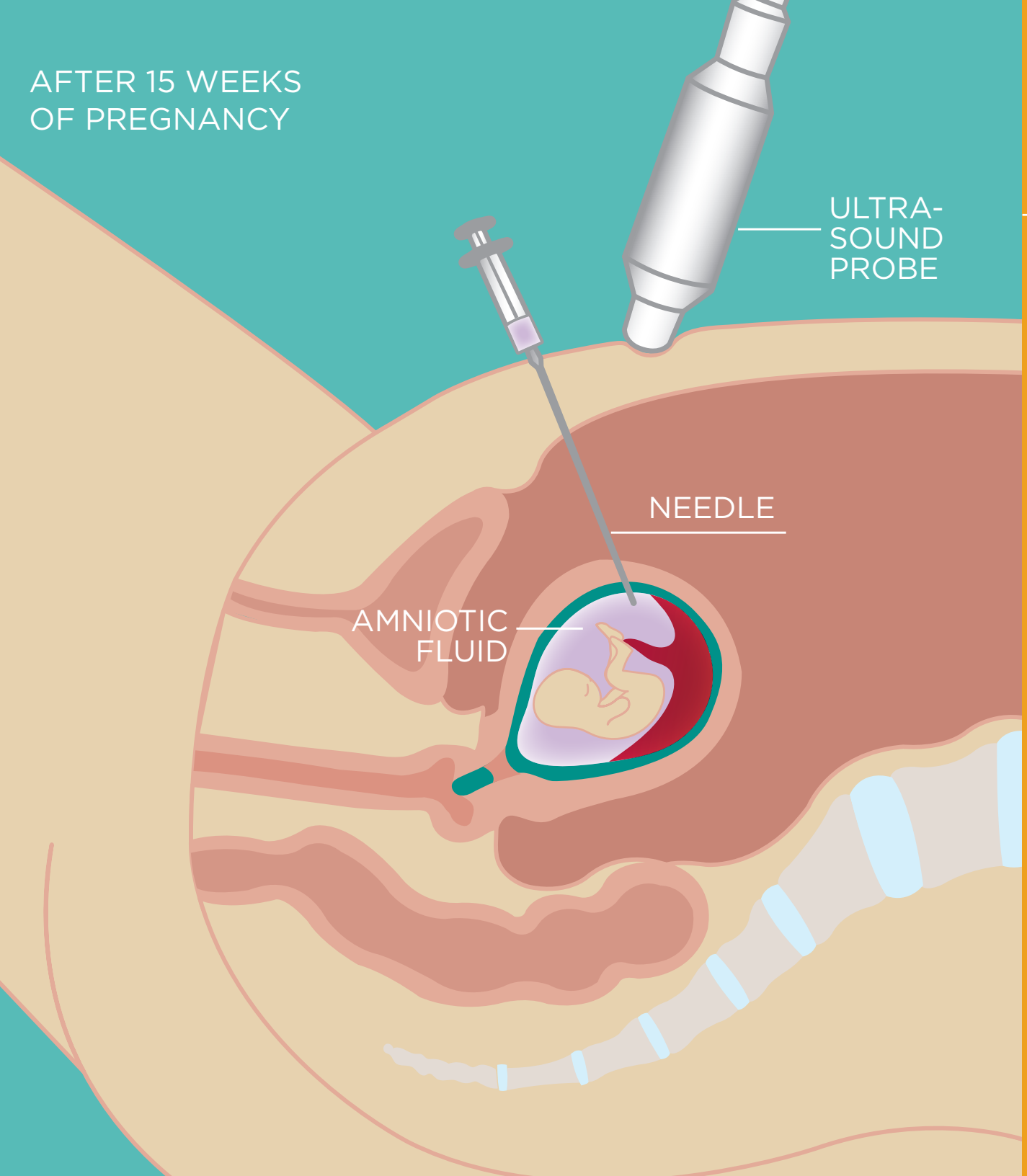
This procedure removes a small sample of cells from the placenta using a needle (through the abdomen) or narrow tube (through the vagina).

All chromosomes within these cells are studied to see if there are any extra or missing.

CVS can provide greater than 99% detection rate for all chromosome conditions.

There is a small chance for miscarriage following the procedure.

1. Chorionic villus sampling. Retrieved from http://www.mayoclinic.com/health/chorionic-villus-sampling/MY00154/DSECTION=chances



Amniocentesis[1]

An amniocentesis is performed after 15 weeksof pregnancy.

Amniotic fluid contains cells from the developing fetus, and a small sample of this fluid is removed using a needle that is inserted through the belly.

All chromosomes within these cells are studied to see if there are any extra or missing.

Amniocentesis can provide a 99.8% detection rate for chromosome conditions.

This procedure also carries a small chance for miscarriage.

1. Amniocentesis. Retrieved from http://www.mayoclinic.com/health/amniocentesis/MY00155/DSECTION=chances

Invasive Confirmatory Testing Reminders

While some women would only consider an invasive test if a screening test like NIPT or ultrasound indicated a high chance for chromosomal conditions, invasive testing is available for any pregnancy. ACOG (American College of Obstetricians and Gynecologists) recommends offering screening or invasive diagnostic test to all pregnant women, regardless of age.[1]

Some women may choose invasive confirmatory testing even if there is not a high chance of a chromosome condition in their pregnancy.

1. ACOG Practice Bulletin 163. Obstetrics & Gynecology May 2016 Vol 127 Issue 5 (e123–e137).

Choosing Your Prenatal Testing Option

Now that you have learned about your prenatal testing options, the choice of screening, invasive or no testing is yours.

Discussing your questions and needs with your healthcare provider or genetic counselor may help you decide which option is best for you.

The Choice is Yours!

During the discussion with your healthcare provider, the following screening or invasive confirmatory testing options may be offered:

- ☐ Harmony® prenatal test (cell-free DNA or non-invasive prenatal screening)
- ☐ Traditional Screening
- ☐ CVS
- ☐ Amniocentesis
- ☐ No screening/testing

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