

The California Prenatal Screening Program: Helpful terms and their definitions

Amniocentesis

Amniocentesis is a diagnostic test. An experienced doctor takes fluid from around the fetus to find out if a fetus may have a birth defect with more certainty than prenatal screening allows.

Birth defects

Birth defects are conditions in a fetus that can cause physical changes and intellectual disabilities. Birth defects most often happen by chance and usually do not run in families. Genetic conditions are a type of birth defect.

cell-free DNA (cfDNA) screening

This screening uses genetic information to find out whether the fetus has an increased chance of certain genetic conditions. A blood sample is used to look at pieces of DNA from the placenta.

Chromosomes

Chromosomes help the fetus develop and are found in every cell in the body. If there are extra or missing copies of a chromosome, the fetus can have a genetic condition.

Chorionic villus sampling (CVS)

Chorionic villus sampling is a diagnostic test. An experienced doctor takes a small number of cells from the placenta to find out if a fetus may have a genetic condition with more certainty than prenatal screening allows.

Diagnostic testing

This term refers to amniocentesis and chorionic villus sampling. Both are tests to find out if a fetus has a birth defect with some certainty. Diagnostic testing gives more definite information than prenatal screening. Prenatal screening only estimates whether there is an increased chance of birth defects.

DNA

Material in the body that carries the information necessary for the fetus to develop and for the cells in our body to work.

Down syndrome (Trisomy 21)

This genetic condition is caused by having three copies of chromosome 21 instead of two copies. It causes mild to severe intellectual disabilities, and serious health problems such as heart defects.

Fetus

A developing baby inside a pregnant individual.

Genes

Pieces of DNA that act as instruction manuals. They tell our cells how to grow and act and help the fetus develop.



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Genetic conditions

Genetic conditions that the PNS Program screens for are caused by an extra chromosome in a fetus or newborn.

Intellectual disability

A condition where a person has some limits in mental functioning and in skills such as communicating, taking care of themselves, and social skills.

Klinefelter syndrome (XXY)

This genetic condition is caused by having an extra X chromosome instead of the typical X and Y chromosomes. There may be mild developmental delays, learning disabilities, and infertility.

Maternal serum alpha-fetoprotein (MSAFP) screening

This screening looks at a protein produced in the pregnant individual's bloodstream. An increased amount of that protein may show the fetus has an increased chance of birth defects called neural tube defects.

Neural tube defects

Birth defects in the neural tube that extends from the top of the head to end of the spine. The tube forms into the fetus's brain and spinal cord. These defects include problems in the development of the brain or spine, like spina bifida (open spine).

Prenatal screening

Prenatal screening uses blood samples to check if a fetus has an increased chance of having a birth defect. Screening gives an estimate of the chances and does not tell if the fetus does have a birth defect. Chorionic villus sampling or amniocentesis are diagnostic tests that may give a clear answer on whether the fetus has the birth defect.

Placenta

The placenta is the organ inside a pregnant individual that provides nutrients and oxygen to the fetus.

Prenatal care provider

A doctor, physician assistant, nurse practitioner, or midwife who has special training in pregnancy and health among pregnant individuals.

Trisomy 21 (Down syndrome)

This genetic condition is caused by having three copies of chromosome 21 instead of two copies. It causes mild to severe intellectual disabilities, and serious health problems such as heart defects.

Trisomy 18 (Edwards syndrome)

This genetic condition is caused by having three copies of chromosome 18 instead of two copies. It causes severe intellectual disabilities and serious health problems.

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Trisomy 13 (Patau syndrome)

This genetic condition is caused by having three copies of chromosome 13 instead of two copies. It causes severe intellectual disabilities and serious health problems

Trisomy X

This genetic condition is caused by having three copies of the X chromosome instead of two copies. There may be mild developmental delays and learning disabilities.

Turner syndrome

This genetic condition is caused by only having one X chromosome instead of two copies. There may be mild developmental delays, learning disabilities, heart defects, and infertility.

Ultrasound exam

A prenatal care provider uses sound waves to get a detailed view of the fetus and the area surrounding it to check on the health of the fetus.

X and Y chromosome variations (Sex Chromosome Aneuploidies or SCAs)

A term for the conditions checked by the Prenatal Screening Program. These include Turner syndrome, Klinefelter syndrome (XXY), Trisomy X, and XYY.

XXY (Klinefelter syndrome)

This genetic condition is caused by having an extra X chromosome instead of the typical X and Y chromosomes. There may be mild developmental delays, learning disabilities, and infertility.

XYY

This genetic condition is caused by having an extra Y chromosome instead of the typical X and Y chromosomes. There may be mild developmental delays and learning disabilities with this genetic condition.