GOLDnatal

Highly sensitive, non-invasive prenatal test

Information brochure for patients

labor team

Dear expectant mother,

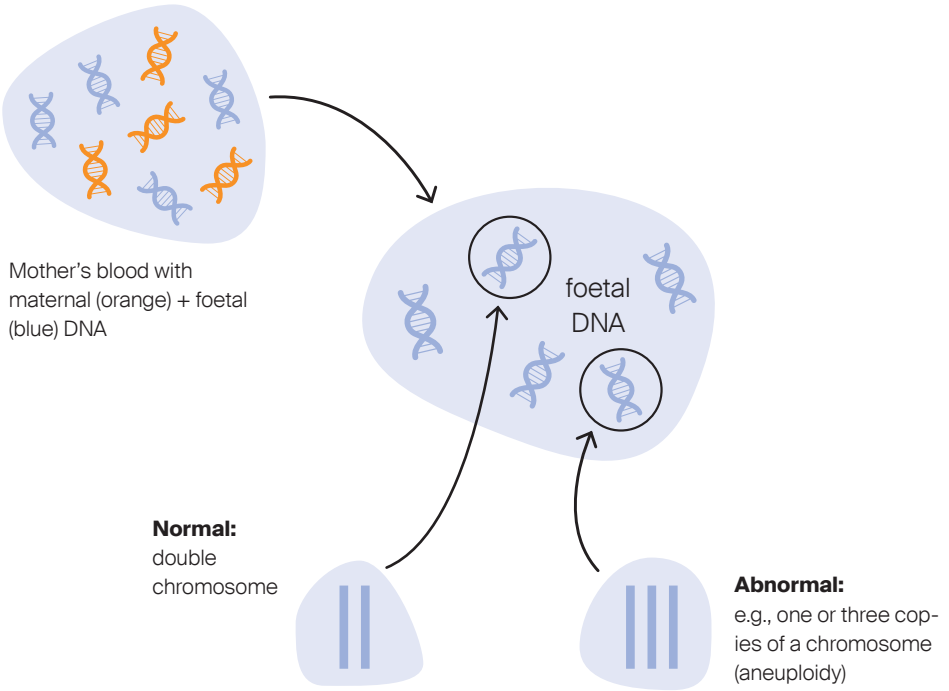
Now that you are pregnant, you may be thinking about your baby's health – be it due to your age, an abnormal first trimester test, or a child with a congenital disease in your family. GOLDnatal is a highly sensitive, non-invasive prenatal test (NIPT) for early pregnancy.

This test makes it possible to identify potential, future, or pre-existing impairments in the child. The results can be used to take action and make early preparations for postnatal treatment.

How GOLDnatal works

Each cell in the human body contains 23 pairs of chromosomes that carry genetic information (DNA). In each pair, one chromosome comes from the mother and one from the father. A pair of chromosomes determines gender: girls have two X chromosomes, boys have one X and one Y chromosome. During pregnancy, small portions of foetal and placental DNA enter the mother's bloodstream, which can be isolated from the mother's venous blood and broken down using the latest technologies. Thus, the DNA can be assigned to the different chromosomes and quantified in order to determine a possible chromosomal abnormality.

Our certified technology is based on the Illumina process and is performed in our Human Genetics Laboratory in Goldach, St. Gallen.



Chromosomal abnormalities tested for by the GOLDnatal scan

Irregularities occur when there is an additional or missing chromosome on a certain chromosome pair. In trisomy, one chromosome is present threefold instead of twofold in the body's cells. The probability of such a genetic defect increases with the age of the mother. GOLDnatal makes it possible to examine the most common trisomies.

Trisomy 21 (Down syndrome)

Trisomy 21 is the best-known and also the most common chromosomal disorder. Its prevalence in newborns is approx. 0.2%. Typical symptoms include delayed mental and physical development, and potentially the abnormal development of certain organs (e.g., hearing and vision problems, heart defects).

Trisomy 18 (Edwards syndrome)

Trisomy 18 occurs slightly less frequently in newborns with a prevalence of approx. 0.04%. This genetic defect causes severe developmental disorders. The majority of foetuses do not survive to birth. The symptoms and their severity are highly variable, with over 100 different malformations associated with Edwards syndrome.

Trisomy 13 (Patau syndrome)

Patau syndrome causes severe malformations of various organs. Most foetuses die before birth and only a few babies survive the first year of life. The prevalence of this genetic defect in newborns ranges from 0.01% to 0.02%.

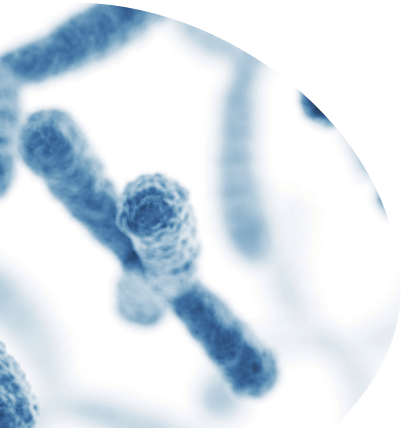
Gonosomal aneuploidies
Gonosomal aneuploidies are abnormalities of the sex chromosomes. GOLDnatal tests for the following anomalies:

Turner syndrome (monosomy X)
In monosomy X, an X chromosome is missing. There is no mental disability and life expectancy is average. Hormone treatment can support a largely normal development.

Triple X syndrome (XXX)
In trisomy X, a woman has an additional X chromosome. Women who have trisomy X are often above-average in height. Most women are only slightly affected or have no symptoms.

Klinefelter syndrome (XXY)
Healthy men have an X and a Y chromosome, while in Klinefelter syndrome there is an additional X chromosome. Height is above average and it often causes testosterone deficiency.

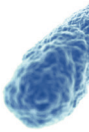
Jacobs syndrome (XYY)
Jacobs syndrome has an additional Y chromosome in men. Men who are affected are often above-average in height. Their intelligence, life expectancy, and fertility are largely unimpaired.



Microdeletion syndrome

Microdeletions represent a form of genomic modification in which small pieces of chromosome were lost. The GOLDnatal assay is used to detect the most common microdeletions, which are significant in the following anomalies:

- DiGeorge syndrome (22q11.2)
- 1p36 deletion syndrome
- Angelman syndrome (15q11.2)
- Cri-du-chat syndrome (5p)
- Prader-Willi syndrome (15q11.2)
- Wolf-Hirschhorn syndrome (4p)

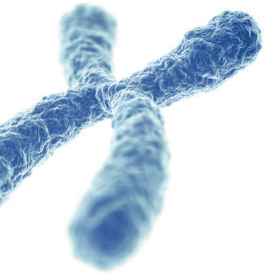


Reliability of GOLDnatal

GOLDnatal is a prenatal test in which a venous blood sample is sufficient for the entire analysis. There is no risk to you or your child. Compared to other non-invasive tests (e.g., first-trimester test), GOLDnatal is much more reliable.

Benefits of GOLDnatal

- Reliable: More than 99% of trisomies 21, 18, and 13 are detected
- Fast: Results of trisomies and aneuploidies within 4 working days, results of microdeletion syndromes within 6–9 working days
- Simple: non-invasive blood test



Limitations of GOLDnatal

GOLDnatal provides a risk assessment (high/low risk) for each syndrome. If there is a high risk of a certain syndrome, further testing using amniotic fluid/choriovillus sampling is strongly recommended. A low risk means that a child is highly unlikely to carry the chromosomal abnormalities examined. As with all non-invasive prenatal tests, 100% safety cannot be guaranteed.

In rare cases, an insufficient amount of foetal DNA in the maternal blood may lead to a test result with limited validity. A vanishing twin or a different chromosomal configuration of the placental cells (placental mosaic) may also lead to results with limited informative value. The test is also suitable for the detection of trisomies 21, 18, and 13 in monozygotic and dizygotic twin pregnancies as well as in ART pregnancies (IVF, ICSI, egg donation). The test is not suitable for the detection of gonosomal aneuploidies and microdeletions in the case of dizygotic twin pregnancies. GOLDnatal can be used starting from the
10th week of pregnancy (9+0 weeks of pregnancy).

Duration and costs of GOLDnatal

GOLDnatal costs CHF 480.60. If the first trimester test is abnormal, GOLDnatal (without gonosomal aneuploidies and microdeletions) is covered by health insurance. The test for gonosomal aneuploidies costs an additional CHF 100.00, the test for microdeletions costs
CHF 230.00 – neither is reimbursed by health insurers. There are no costs for tests without conclusive results. The test results of the trisomy assessment and the gonosomal aneuploidies are available within four working days. Assessment of microdeletion syndromes take 6–9 working days.

labor team w ag
Blumeneggstrasse 55
CH-9403 Goldach
+41 71 844 45 45
info@team-w.ch
www.laborteam.ch

M14783/0225