the reassurance of

knowing

Shedding added, valuable light on the chromosomal and subchromosomal health of a pregnancy.

verifi®
prenatal test

your questions–answered

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TEST PRENATAL NO INVASIVO | ADN FETAL LIBRE EN SANGRE MATERNA

veragen

Responsibly expanding the most accurate, reliable, and relevant Non-invasive Prenatal Test (NIPT) available.

An elective panel of clinically significant trisomies and microdeletions.

Built on the trusted foundation of the verifi prenatal test

- The same accurate answers you've come to rely on from the verifi test—without the risks of invasive procedures
- Thorough and responsible test expansion—recommended in conjunction with clinical context such as abnormal ultrasound and family history
- Proven technology for confident results—takes advantage of the most widely used Illumina next-generation sequencing technology
  - Available as *elective* choices—not as mandatory components of the verifi test

The option to detect two key trisomies associated with an increased risk of miscarriage

- **Trisomy 9**—A rare chromosomal condition with the vast majority of instances resulting in miscarriage in the 1st trimester. While the majority of live births will not survive during early postnatal period, those that do will have serious health concerns, including intellectual disability and cardiac defects. It can also occur in mosaic form[1–4]
- **Trisomy 16**—The most commonly occurring autosomal trisomy seen in first trimester miscarriages. Rare survivors with mosaic trisomy 16 are at increased risk for health concerns including intra-uterine growth restriction, intellectual disability, and cardiac defects[2, 4–6]

There is a small increased risk for a woman to have a pregnancy with a viable trisomy following a miscarriage with trisomies 9 or 16.[1–6] The ability to identify these important chromosomal causes of miscarriage can help with risk assessment as well as monitoring and management of subsequent pregnancies.

The verifi test option for trisomies 9 and 16 can be ordered separately from the microdeletion panel.

What are microdeletions?

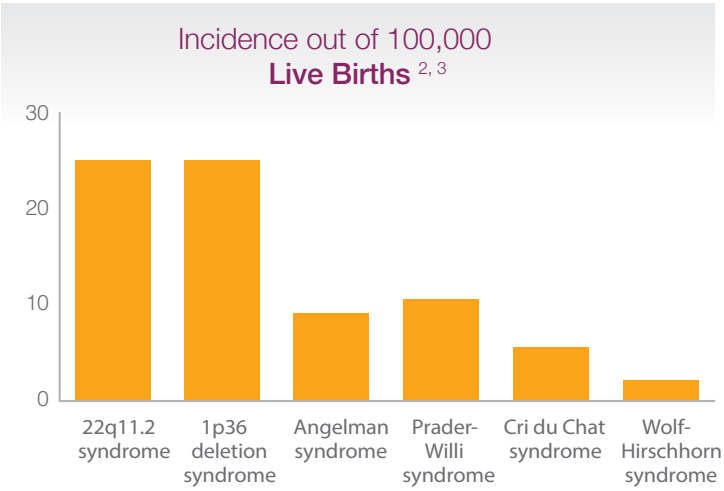
Microdeletions are chromosomal disorders caused by small missing pieces of chromosome material. They are usually not visible by standard methods of chromosome analysis. Microdeletions can occur on any of the 23 pairs of chromosomes. Some occur more commonly in a specific area of a particular chromosome and have been linked to known genetic syndromes. Most occur by chance, rather than being inherited from a parent, and can occur with no prior family history and without other risk factors, such as advanced parental age. Results from routine pregnancy screening are usually normal.

Why is it valuable to test for microdeletions?

Many microdeletion syndromes can cause serious health issues including both physical and intellectual impairment—the severity of which can vary from individual to individual. These conditions usually cannot be detected by traditional serum screening and may or may not be associated with ultrasound abnormalities. Until now, an invasive procedure, such as chorionic villus sampling (CVS) or amniocentesis, was the primary way to detect such conditions prenatally.

Why choose the verifi microdeletion panel?

The capabilities of the verifi test have been expanded to help detect the five microdeletion regions. By doing so, the verifi microdeletion panel can provide valuable information to aid in pregnancy management and preparing for newborn care. This additional panel provides physicians with further non-invasive testing options in certain clinical situations.



Going to greater lengths for the answers that matter most.

Taking a deeper approach to the science of genetic sequencing, the verifi test provides a clearer picture of chromosomal health—detecting even small abnormalities with a high degree of accuracy and reproducibility.[20] The verifi microdeletion panel has been validated on actual clinical and analytical samples. Its optimized algorithm addresses the complexities of specific chromosomal regions to provide accurate answers about the loss of genetic material. The result is better overall performance, including a low false positive rate compared to other NIPTs, and the lowest test failure rate in the industry.[21] Responsibly investing the time and technology to do things right, and leading the field in clinical publications, the verifi microdeletion test means greater reassurance and peace of mind.

Unmatched flexibility

The verifi microdeletion panel can be added up to 60 days after basic test results have been issued—no additional blood draw needed! Our lab will save a patient's blood sample for 60 days. **This incomparable choice provides the unique ability to look more deeply should an abnormal ultrasound occur after the initial NIPT.**

Test performance and accurate answers you can rely on

Overall Sensitivity of 91.6% and Specificity of 99.84%

Syndromes	22q11.2 deletion syndrome	1p36 deletion syndrome	Angelman / Prader Willi syndrome	Cri du Chat syndrome	Wolf-Hirschhorn syndrome
Min. Syndrome Region Size	2.7 Mb	5 Mb	5.8 Mb	9.8 Mb	3.6 Mb
Sensitivity					
No. Affected Samples Tested	8	0	0	2	2
No. Samples Detected	7	0*	0*	2	2
% Sensitivity [95% CI]	87.5% [47–99]	†	†	100% [15–100]	100% [15–100]
Specificity					
No. Putative Unaffected Samples Tested	1797	1797	1797	1797	1797
No. Samples Detected	0	0	1	0	2
% Positive Call Rate	0% [0–0.2%]	0% [0–0.1%]	0.05% [0.01–0.31%]	0% [0–0.2%]	0.11% [0.01–0.4%]
% Specificity [95% CI]	> 99.8%	> 99.9%	> 99.7%	> 99.8%	> 99.6%

* Titration of fragmented genomic DNA derived from cell lines containing either a 1p36 or 15q11.2 deletion demonstrated a linear dose response and confirmed the assay's ability to measure copy number change at these loci.

† No estimates of confidence intervals or sensitivity were performed for sample sizes < 2.

The verifi microdeletion panel identifies 6 important genetic syndromes.

	Incidence	Clinical Features (may include but not limited to)	Life Expectancy
22q11.2 syndrome (DiGeorge syndrome, Velocardialfacial syndrome)[2, 5–7]	1 in 4,000	Learning problems, congenital heart defects, palatal abnormalities	Usually normal, can be reduced for DiGeorge syndrome
1p36 deletion syndrome[2, 8–11]	1 in 4,000 to 1 in 10,000	Characteristic craniofacial features, intellectual disability, seizures, brain and heart defects	Depends on the severity of features, but can be normal
Angelman syndrome (15q11.2 deletion syndrome)*[2–3, 5, 12]	1 in 12,000	Intellectual disability, speech impairment, seizures	Normal
Prader-Willi syndrome (15q11.2 deletion syndrome)*[2–3, 5, 13]	1 in 10,000 to 1 in 25,000	Hypotonia, morbid obesity, delayed motor and language skills, intellectual disability, hypogonadism	Normal, may be reduced depending on the severity of symptoms
Cri du Chat syndrome (5p-syndrome)[2–3, 5, 14–16]	1 in 20,000 to 1 in 50,000	Intellectual disability, speech delay, cat-like cry	10% mortality in the first year; otherwise usually normal, but will depend on the severity of features
Wolf-Hirschhorn syndrome (4p-syndrome) [2, 3, 17–19]	1 in 50,000	Growth deficiency, hypotonia, craniofacial features, intellectual disability, heart and brain abnormalities	Depends on severity of features

*The microdeletion region is the same region for Angelman and Prader-Willi syndromes (15q11.2). NIPT will not distinguish between these two syndromes. Further testing is necessary.

Simple, safe, non-invasive blood test— just one tube of maternal blood needed.

- Performed as early as 10 weeks gestational age
- A deeper approach to the science of sequencing for the most accurate answers in NIPT
- Fast turn-around time; results usually available within 3–6 business days
- The most clinically relevant elective panel of trisomies and microdeletions

verifi® prenatal test

your questions–answered

Disclaimer
The manner in which this information is used to guide patient care is the responsibility of the healthcare provider, including advising for the need for genetic counseling or additional diagnostic testing. Any diagnostic testing should be interpreted in the context of all available clinical findings.

This test was developed by, and its performance characteristics were determined by, Verinata Health, Inc. It has not been cleared or approved by the U. S. Food and Drug Administration. Although laboratory-developed tests to date have not been subject to U.S. FDA regulation, certification of the laboratory is required under the Clinical Laboratory Improvement Amendments (CLIA) to ensure the quality and validity of the tests. Our laboratory is CAP-accredited and certified under CLIA as qualified to perform high-complexity clinical laboratory testing.

Limitations of test
This test is designed to detect subchromosomal deletions and is validated for common deletions in chromosomal regions 15q11.2, 5p15.2, 22q11.2, 1p36, and 4p16.3. The test is validated for singleton pregnancies with gestational age of at least 10 weeks as estimated by last menstrual period. These results do not eliminate the possibility that this pregnancy may be associated with other chromosomal or subchromosomal abnormalities, birth defects, and other conditions. This test is not intended to identify pregnancies at risk for open neural tube defects. A negative test result does not eliminate the possibility of Angelman syndrome, Prader-Willi syndrome, 5p-/Cri du Chat syndrome, 22q11.2 deletion syndrome, Williams syndrome, 1p36 deletion syndrome, or 4p-/Wolf-Hirschhorn syndrome. In addition, conditions caused by other molecular mechanisms cannot be detected with this assay. There is a small possibility that the test results might not reflect the chromosome status of the fetus, but may reflect subchromosomal changes of the placenta (confined placental mosaicism), or of the mother.

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