

# Prenatal testing



You decide!

Questions about non-invasive screening tests to discuss with your doctor:

- What is this test for?
- How will it be carried out?
- Are there any risks associated with the test? Which ones?
- How accurate is the test?
- Is there a chance of unexpected, worrisome test results? What could they be?
- What are the follow-up examinations and procedures I may have to face?
- Until when will I have to decide in favour of or against the test?
- When can I expect the results?
- How much will the examination and potential follow-up treatment cost?
- Where can I find additional information and advice?



Questions for the discussion of positive test results with your doctor after invasive testing:

- How accurate is the result?
- What does the detected disorder mean?
- Are there any prenatal measures available to address it?
- What are my options in the current situation?
- Can you provide me with information about relevant parents' associations so I can talk to other parents concerned by life with a child with an impairment?
- How would a potential pregnancy termination be carried out?
- How long do I have to decide on how to proceed?
- Where can I find additional information and advice?

Questions for the discussion of positive test results with your doctor after non-invasive testing:

- How accurate is the result?
- What does the detected anomaly mean?
- What are my options?
- Until when will I have to decide in favour of or against further testing?
- Where can I find additional information and advice?

Questions about invasive tests to discuss with your doctor:

- What is this test for?
- How will it be carried out?
- What does the anomaly my child is being tested for mean?
- Are there any risks associated with the test? Which ones?
- How accurate is the test?
- Are there any prenatal measures available in case of a positive test result?
- What are the follow-up examinations and procedures I may have to face?
- How much will the examination and potential follow-up treatment cost?
- Until when will I have to decide in favour of or against the test?
- When can I expect the results?
- Where can I find additional information and advice?



97% of children are born without illness or disability. However, prenatal screening tests such as ultrasound scans (e.g. nuchal translucency) and blood tests for certain illnesses and disabilities of the unborn child are often carried out as a matter of course. Such tests can lead to difficult decisions for you as parents-to-be. This is why we recommend you seek information and advice as early as possible – it will help you answer the question of whether or not to test with confidence.

The results show an increased chance of the baby having an illness or a disability

Would you like to have further, invasive tests carried out in order to obtain a reliable diagnosis? Less than 0.5% of invasive tests result in a miscarriage – if carried out by experienced practitioners

Would you like to proceed with the tests?

Yes

Preliminary, non-invasive screening tests are carried out to determine the chance of the child being born with an illness or a disability

Nothing of concern has been found

No

You accept your child as he or she is

The test resulted in a miscarriage

The diagnosis is clear: Your child will be born with an illness or a disability

The diagnosis is clear: Your child will be born without illness or a disability

Terminate the pregnancy

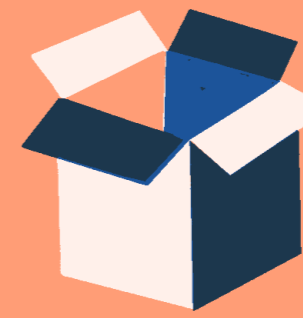
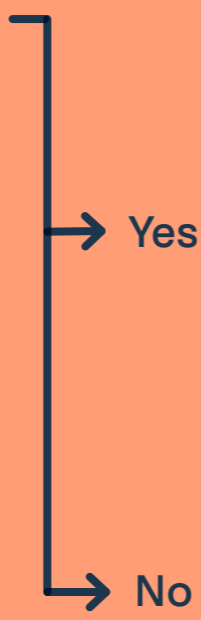
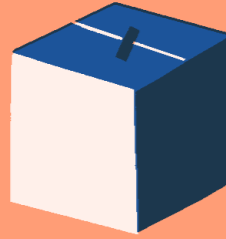
What does this diagnosis mean for you and your family? What next?

Continue the pregnancy

Miscarriage

Birth

Prenatal testing is no guarantee for a healthy child. On the one hand, prenatal tests can only detect some of the illnesses and disabilities a child might be born with and on the other, false results can occur. What is more, some illnesses and disabilities are the result of birth complications.



## Prenatal tests – you decide!

You are going to be parents. A time of heightened emotion lies ahead and, with it, many new questions and topics. A topic which does not often emerge early on without prompting is prenatal screening and testing.

They include regular monitoring of blood pressure, urine status and body weight, as well as ultrasound scans and a range of blood tests. All these examinations can help predict pregnancy complications and potential chromosomal disorders or malformations of the child. Many of these routine screenings have now become standard procedures to an extent that they are no longer even being discussed. However, expert support of parents-to-be should be based on empathy, non-directive information, advice and time – a precious commodity today.

While screening for chromosomal disorders such as Trisomy 21 has been a significant part of prenatal diagnostics for decades, the screening concept itself has evolved considerably over the past few years. Non-invasive tests in particular have become much more effective, but also more complex and more difficult to explain and, therefore, more difficult to understand.

We, the *insieme* parents' association, care deeply about informing parents-to-be without pressuring them into deciding in favour of or against prenatal testing, or in favour of or against having a child with an impairment. This brochure is intended to highlight that you have the right to make an informed decision yourselves on how much you would like to know about your child at the prenatal stage. We would like to provide contacts and links where you can obtain further information and advice, and give you the opportunity to consider the implications of prenatal testing early on in the process of becoming a parent.

To launch the discussion, we invite you to retrace the decision-making path on the front page.

## Prenatal tests for the unborn child

Prenatal tests – also known as prenatal diagnostics – which are carried out to examine the unborn child, are medical tests to examine the unborn child's chromosomes and to find evidence of possible anomalies. The tests generally fall into one of two categories: invasive and non-invasive

## Non-invasive tests

Non-invasive tests include ultrasound scans and various blood tests, as well as the first trimester test (ETT) or foetal DNA tests (NIPT).

The ultrasound scan is a tool to determine the number of unborn children, to detect anomalies and to monitor amniotic fluid levels as well as the child's growth.

The ETT provides data for a Trisomy 21 risk assessment, but it cannot predict if the unborn child is, in fact, affected. This can only be determined with an invasive test.

In the NIPT test, foetal DNA in the mother's blood is analysed. None of these tests can provide a 100 % certainty as to the chromosomal health of the child.

There are various NIPT tests available today – screenings for a range of syndromes and other chromosomal abnormalities. A positive NIPT result is not a certain indicator that the child is affected – an invasive follow-up test is required for confirmation.

## Invasive tests

Invasive testing is the only way to conclusively examine the unborn child's DNA.

Invasive tests involve the ultrasound-assisted sampling of tissue through a needle from the placenta (chorionic villus sampling), from the amniotic fluid (amniocentesis), or, in rare cases, directly from the umbilical chord (cordocentesis). Unlike the non-invasive tests, these examinations can result in a miscarriage. Nowadays, the associated risk is less than 0.5 % if the procedure is carried out by a seasoned practitioner.

## Prenatal tests are voluntary

Prenatal tests are voluntary. You, as a mother and father-to-be, have the right to decide whether you want to take advantage of all available test methods or just some of them, or if you would rather not have any tests at all.

You are entitled to change your mind at any point of the pregnancy and you have the right to ask to not be notified of some or all of the test results if you so wish.

## Make sure you are given all the information and advice you need

Before any prenatal tests are carried out, your doctor will inform you about their advantages and disadvantages and about their cost and possible consequences. If necessary, ask for more information and use our lists of questions to help you discuss the matter with your doctor. Complement the lists with your own questions if you so wish.

In addition, you can always consult any advice centre or parents' association in the field.

## Take your time

Make sure to carefully consider prenatal testing before you agree to have it. After consulting your doctor, you can always ask for a second opinion from an advice centre or parents' association, talk to your friends and family.

Information and advice centres for prenatal testing:

- Sexuelle Gesundheit Schweiz (Sexual Health Switzerland, register of regional information and advice centres)  
[www.sante-sexuelle.ch](http://www.sante-sexuelle.ch)
- Verein Ganzheitliche Beratung und kritische Information zu pränataler Diagnostik (Association for holistic advice and critical information about prenatal diagnostics)  
[www.praenatal-diagnostik.ch](http://www.praenatal-diagnostik.ch)
- [www.appella.ch](http://www.appella.ch)
- Kindsverlust.ch (Information, counselling and support in the event of the death of a child during pregnancy, at birth and in a child's early life)  
[www.kindsverlust.ch](http://www.kindsverlust.ch)

Parents' organisations:

- insieme (parents' associations for people with a learning disability and their families)  
[www.insieme.ch](http://www.insieme.ch)
- insieme21 (parents' association for people with Trisomy 21 and their families)  
[www.insieme21.ch](http://www.insieme21.ch)
- Konferenz der Vereinigungen von Eltern behinderter und langzeitkranker Kinder – KVEB (Conference of parents' associations of children with chronic illnesses and disabilities, contacts to various associations)  
[www.behindertekinder.ch](http://www.behindertekinder.ch)
- Elternsofa (a platform to connect with other parents (-to-be) of a child with a learning disability)  
[www.elternsofa.ch](http://www.elternsofa.ch)

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insieme – together with and for  
people with a learning disability

The insieme parents' association campaigns for a society open to all human beings. insieme provides a framework for the inclusion of people with a learning disability in our society so they can live, work and thrive in our midst.

[www.insieme.ch](http://www.insieme.ch)  
[www.prenatal-testing.ch](http://www.prenatal-testing.ch)

