



**praenatal.de**

**Praenatal-Medizin  
und Genetik**

**Information on  
prenatal diagnostics**



**Praenatal-Medizin  
und Genetik**

Medical Partnership  
PD Dr. med. P. Kozłowski und Partner  
Essen District Court PR 1853

**praenatal.de**

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### **Child's profile**

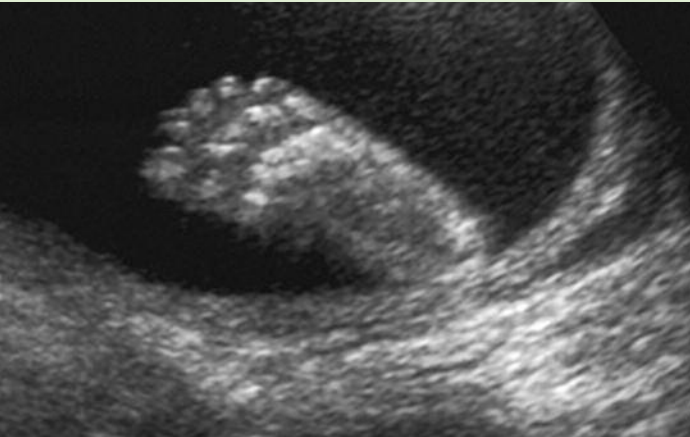
*in the 22nd WG (week of gestation)*

## **Dear patient,**

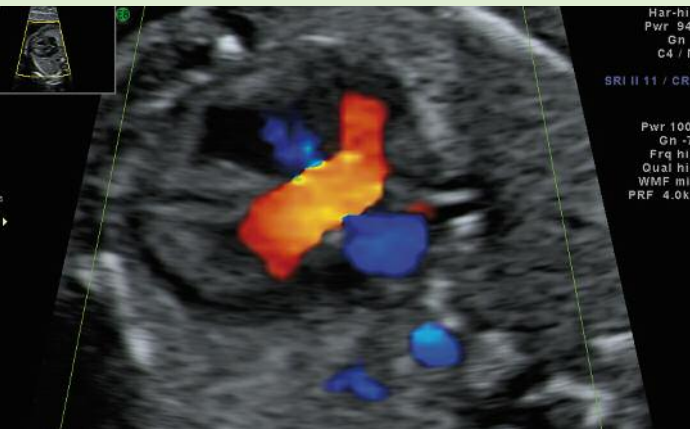
Your gynaecologist has explained to you the various prenatal screening methods. It could be that you have not yet decided whether and which of these screenings are worth considering. We, the cooperation partner of your gynaecologist and doctor are providing a summary in this brochure of what prenatal diagnostics offer and the legal regulations applicable to them from 2010 onwards. In addition, detailed information can be found on our website:

[www.praenatal.de](http://www.praenatal.de)

**Child's foot**  
*in the 22nd WG*



**Intersection of the large cardiac vessels**  
*in the 22nd WG*



## Prenatal diagnostics

Prenatal diagnostics encompasses medical screenings that are used to check the health of the unborn baby in the womb. The particular method employed depends on

- your week of pregnancy
- specific issues, e.g. your age, family history, your own previous conditions, adverse ultrasound findings

None of these screenings, however, can guarantee a completely healthy child.

### When is genetic counseling necessary?

Your gynaecologist will know whether there are any circumstances in either your own or your family's history to indicate a potential risk for your unborn child. In this case, you will be referred for a genetic counselling session where we will discuss whether, given your history and your family tree, it makes sense to perform specific screenings.

Your doctor will also tell you about the genetic counselling before a scheduled screening between your 11th and 14th week of pregnancy. If there are no special circumstances surrounding either you or your family, the genetic counselling session will deliver additional findings only in rare cases.

On the other hand, before a scheduled amniocentesis or placenta screening, a genetic counselling session is helpful to jointly decide the scope of the screening.

# Causes of diseases in the unborn child

## Chromosomal abnormalities

Every cell of a healthy person's body has 46 chromosomes, 23 of which are inherited from the mother and 23 from the father. All chromosomes therefore occur in pairs. The number or the structure of the chromosomes, however, can be altered. Replication errors can cause chromosomes to occur in threes instead of twos. This is called trisomy. The most common is the trisomy of chromosome 21, which is known as Down's syndrome.

The number of the sex chromosomes (X/Y chromosomes) can also be altered. Anomalies in the structure of chromosomes can also affect the development of the unborn child. Every pregnancy carries a slight risk of such random chromosomal abnormalities. This is not influenced by either your family's history or your own lifestyle.

However, the risk of some chromosomal abnormalities increases with age (see Table).

### Age risk for Down's syndrome (for a pregnancy in the 12th week)

20 years	1 : 1068	36 years	1 : 196
25 years	1 : 946	38 years	1 : 117
30 years	1 : 626	40 years	1 : 68
32 years	1 : 461	42 years	1 : 38
34 years	1 : 312	44 years	1 : 21

## Hereditary diseases

Hereditary diseases are caused by genetic modifications. They occur for the first time or already exist undetected in one, more often in both parents. Both metabolic functions and physical build can be affected. The potentials of detecting a hereditary disease prenatally and consequences must be clarified beforehand in a human genetic

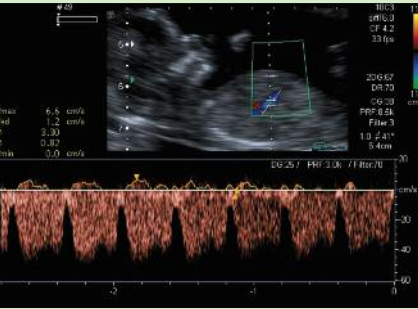
counselling session. Many diseases can be diagnosed before birth using molecular genetics.

## Organ malformations

The majority of these malformations occur during the second and third month of pregnancy. Kidneys, urinary tract, heart and brain are the most commonly affected. Often, no cause can be found. In rare cases, external factors such as medication, exposure to radiation or maternal infections can play a role. It is highly likely that an ultrasound scan will reveal organ malformations. However, a clear screening does not always rule out the existence of these malformations.

Normal set of human chromosomes





**Blood flow to the heart**  
*(Ductus venosus)*  
*in the 13th WG*



**Child's face**  
*in the 30th WG,*  
*3D imaging*



**Aortic arch**  
*in the 22nd WG*

## Screening methods

Test	Optimum time
Early organ screening (with risk calculation for trisomy where applicable)	12 + 0 - 13 + 2 WG
Chorionic villus sampling	from WG 11 + 0
Amniocentesis	from WG 14 + 0
Alpha-Feto-Protein (AFP)	from WG 15 + 0
Cordocentesis	from WG 18 + 0
Ultrasonic differential screening / echocardiography	19 + 0 - 20 + 6 WG (or earlier if recommended by your doctor)
Doppler ultrasound	from WG 26 + 0

**Explanation:** *WG 13+2 means 13 complete weeks plus 2 days, i.e. the same as the 2nd day of the 14th week*

## Early organ screening

(11 - 14 WG)

This screening has gained increasing importance in recent years. Your doctor will discuss with you whether he / she will perform this screening him- / herself or refer you to us.

The focal point of early diagnostics is a detailed ultrasound screening. Good screening conditions provide us with a host of information about your child's development and organs. **We recommend that this screening is performed between 12+0 and 13+2 week of pregnancy.**

# Risk calculation for trisomy

(first trimester screening)

Trisomy 21 (Down's syndrome) in the unborn child can occur whatever the age of the mother, although the probability increases with the mother's age.

If you want to calculate your own personal risk for a trisomy 21, we will record additional markers (nasal bone, tricuspid valve, Ductus venosus, facial angle) during the screening and carry out a probability calculation according to the method of the Fetal Medicine Foundation (FMF) London ([www.fetalmedicine.com](http://www.fetalmedicine.com))

## However, you should be aware that

- *A clear ultrasound scan cannot guarantee a healthy child. The result of the risk analysis is not a clear indication of whether your child has a chromosomal abnormality or not.*
- *The probability calculation records only the risk for a trisomy of chromosomes 21, 13 and 18.*

*The table shows the risk of abnormalities in other chromosomes in a pregnant woman, whose first semester screening for trisomy 21, 18 and 13 has revealed a risk of less than 1 to 1000:*

Age	Residual risk
35 years	1 : 550
37 years	1 : 420
40 years	1 : 210
41 years	1 : 170
42 years	1 : 140

It is very important that you understand beforehand how helpful a pure probability analysis will be for you and what conclusions you would draw from a high or low risk value. If you want to safely rule out the most frequent chromosome abnormalities, we recommend a chromosome analysis on your child's cells. This requires either a chorionic villus sampling (from WG 11 + 0) or an amniocentesis

(from WG 14 + 0). We will be happy to explain the potentials and risks of these screenings to you in a genetic counselling session.

## How is the risk for trisomy calculated?

The first trimester screening can be performed only between WG 11+1 and 13+6. Ultrasound is used to measure what we refer to as the nuchal translucency (also called nuchal fold) of the unborn child. If the nuchal translucency is enlarged, the probability of your child having a disease increases.

In parallel, two values (the concentration of the protein **PAPP-A** and of the hormone **free  $\beta$ -HCG**) can be determined in the mother's blood.

The determined risk is given as a ratio: A 1 in 500 risk for Down's syndrome means that out of every 500 pregnant women, one will have a child with Down's syndrome.

If we include the markers of nasal bone, tricuspid valve, Ductus venosus and facial angle, the accuracy of the first trimester screening is around 90%.

According to the German Genetic Diagnosis Act (GenDG) in force since February 2010, an ultrasound scan followed by a risk calculation is considered a genetic screening. Just like before an amniotic fluid puncture, your doctor is under an obligation to explain the screening to you, offer you genetic counselling and provide you with written information. You must declare your consent or refusal in writing. At every genetic screening, you can revoke your consent to the test or to the results being stored at any time.



### Other causes of enlarged nuchal transparency

Other reasons for an enlarged nuchal transparency include heart defects, diaphragm/umbilical hernia, skeletal or metabolic defects. In some cases, there is no particular reason and the pregnancy continues without complications.

Besides the first trimester screening, other additional tests on the mother's blood are offered although these are less relevant.

Low values of the protein PAPP-A can also indicate an increased risk of problems with the placenta function and the mother's blood pressure as the pregnancy progresses.

#### **Please note:**

*In normal pregnancies, the first trimester screening is not part of the regular check-ups. The costs for consultation, ultrasound scans and lab screenings are not paid by public health insurance.*

# Ultrasonic differential screening

During the **18th and 21st weeks of pregnancy**, we can conduct a detailed **organ scan**. This is much more extensive than the ultrasound scan set out in the German Maternity Guidelines during this period. Besides special equipment, it also requires the physician to have a great deal of experience.

Your doctor will decide whether there are reasons to refer you to ultrasonic differential screening. If this is not the case, you can of course have this screening as a self-payer service.

We look at all reproducible organs and features of the unborn child:

- the child's growth for its age
- the amount of amniotic fluid
- the appearance and function of all visible organs
- the position and appearance of the placenta

In many cases, the detailed ultrasound scan can reveal features that increase the statistical risk of Down's syndrome. These irregularities are not malformations and they do not impair the organs concerned. If, during the screening, we discover one or several of such "markers", we can carry out a risk assessment for Down's syndrome (trisomy 21) based on your age.

This screening also involves assessing your baby's heart and the large blood vessels by means of an **echocardiography**. We monitor:

- The heart's position, size and symmetry
- Anatomy of the heart's structures
- Function of the heart valves and chambers
- Heart rate
- Position of the large arterial and venous vessels

The blood flow in the umbilical cord, blood vessels of the child's circulation and in the blood vessels in the womb are also shown in colour and acoustically using the Doppler ultrasound scan. The **Doppler ultrasound scan of the child's and mother's blood vessels** is used mainly during late pregnancy (**WG 26 – 38**) if an acute or chronic deficiency in the supply to the unborn child is suspected.

More and more pregnant women are interested in the potentials of the **3D/4D ultrasound scan**. The main aspect is the fascinating image. We consider this modern procedure a complementary method when special issues arise. For that reason, we use 3D/4D imaging only if we can expect it to deliver additional diagnostic findings. We do not perform 3D/4D imaging without an organ scan at the same time.

## **Please note:**

*Even with good equipment quality and care, it is not possible to completely rule out malformations and diseases. Ultrasound technology reaches its limits under poor screening conditions or in case of very minor defects such as a small defect in the ventricular septum or minor defects in the area around the spinal column.*

*An ultrasound scan can never rule out the possibility of chromosomal abnormalities as such diseases do not necessarily involve organ defects.*

## Amniocentesis

(from WG 14 + 0)

The baby's cells can be taken from the amniotic fluid and the chromosomes analysed. In this way, numerous chromosome abnormalities can be ruled out with a very high degree of certainty. We perform an amniocentesis sometime after the 14th week of pregnancy.

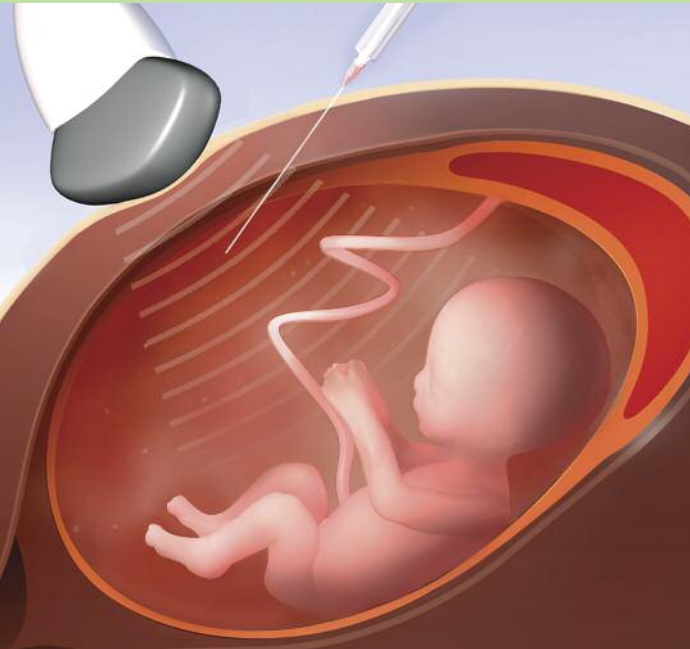
### What does amniocentesis entail?

Under continuous ultrasound monitoring, a very fine needle (0.7 mm diameter) is inserted through the mother's abdominal wall and into the amnion in such a way that no injury to the unborn baby can occur. Around 10 ml of amniotic fluid are removed. That's less than a tenth of the entire amount. The puncture itself lasts one to two minutes and only causes a slight pull in the abdomen. Immediately after the puncture, the minute injection canal closes again as the tissue is highly elastic. The baby's cells from the amniotic fluid are used to create cultures that require some time for cell growth. The result of the amniocentesis is available around 10 to 14 days after the puncture.

### Which diseases can be detected from the amniotic cells?

An analysis of the amniotic fluid cells reveals microscopically visible chromosomal abnormalities. If special circumstances exist, molecular-genetic, invisible chromosomal changes and a number of hereditary diseases can be diagnosed.

To rule out spina bifida with a high degree of certainty, a protein (Alpha-Feto-Protein, AFP) and an enzyme (Acetylcholinesterase, AChE) are investigated in the amniotic fluid.





**Amniotic fluid preparation in the lab**

### **What risks does amniocentesis carry?**

The risks are often overestimated. Every pregnancy – even without intervention – carries a natural risk of loss. In very rare instances, amniotic fluid is lost through the vagina after a puncture. In most cases, however, this defect in the amniotic sac closes again and the pregnancy continues normally. During the last 18 years, we have performed more than 60,000 amniocenteses. A puncture from the 15th week increases the risk of a natural miscarriage by an average of 0.3%. In uncomplicated pregnancies without heavy previous bleeding, the individual risk is much lower. We discuss with you your personal risk and the ways you can reduce it, by taking an antibiotic for example, during the consultation.

### **Quick test after amniocentesis**

To ease the waiting time until the final result is available, we offer you a quick test (PCR). The result, usually available within half a working day of the puncture, safely rules out the most common chromosomal abnormalities (trisomy 13, 18, 21) and can determine you your baby's sex. The final chromosomal findings are available 10-14 days following the puncture. To conduct the PCR quick test, we require only minute quantities of genetic material (DNA), which means that no additional amniotic fluid needs to be removed. The costs for the quick screening are paid by public health insurance only when there are serious irregularities and they are not always paid by private insurers.

## Chorionic villus sampling

(from WG 11 + 0)

As the placenta (called chorion during the early weeks of gestation) comes from the fertilised ovum, cells taken from it can be used to analyse your baby's chromosomes. A chorionic villus sampling makes sense if a chromosomal analysis is required in the very early stages of pregnancy, e.g.:

- if the ultrasonic image shows irregularities
- if the first trimester screening reveals a high risk
- in the case of hereditary disease or metabolic disorders
- if you request one after an early screening.

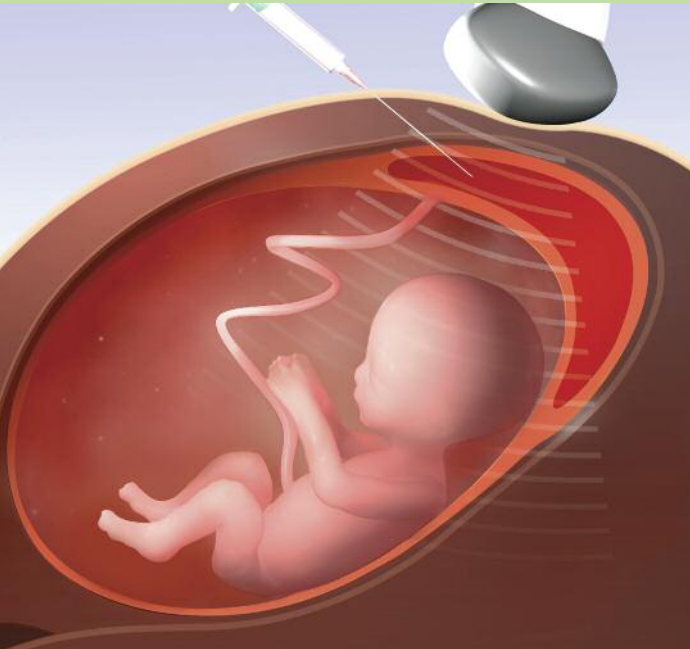
We perform a chorionic villus sampling any time from the end of the 11th WG. This screening does not provide any information on spina bifida.

### What does a chorionic villus sampling entail?

After a careful ultrasound scan, a fine needle is inserted under constant visual monitoring through the mother's abdominal wall into the placenta and minute pieces of tissue are removed. The puncture usually lasts only one to two minutes. During the operation, most women feel a sharp pull in the abdomen. The cells are used to create cultures that require some time for cell growth and replication. The result is available after about ten days. If the amount of tissue is adequate, a short-time test is performed, which is able to rule out the most common chromosomal abnormalities just two working days following the puncture.

### What risks does a chorionic villus sampling carry?

The risks of a chorionic villus sampling are very similar to those of amniocentesis. The risk of natural loss is increased by an average of 0.3 to 0.5%. As with amniocentesis, the individual risk is lower in uncomplicated pregnancies.

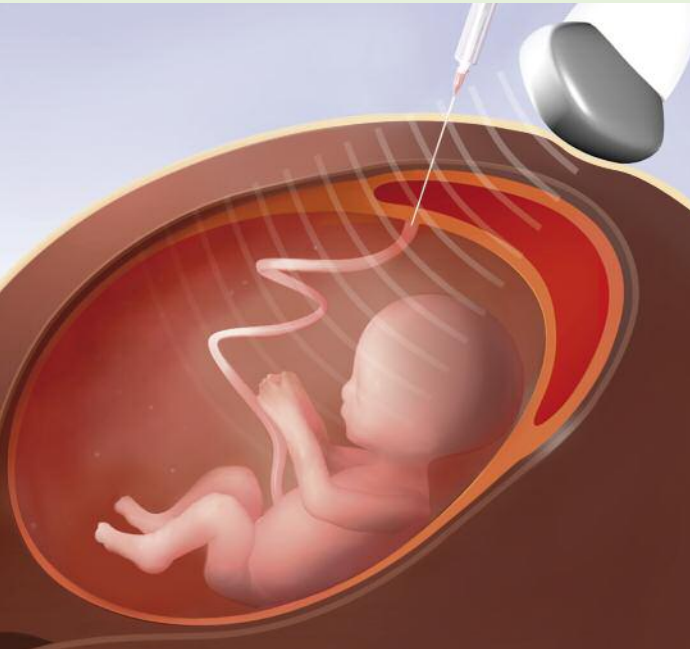


Chorionic villus sampling

## Cordocentesis

(from WG 18 + 0)

The umbilical cord puncture is used only when there are special circumstances, e.g. baby's anaemia, infection or irregularities discovered relatively late. With some diseases, blood or medicines are injected into the baby's circulation.



## How safe is it to screen the baby's cells?

Screening cells taken from the amniotic fluid, the placenta tissue or the unborn baby's blood are still the only possibility to rule out numerous chromosomal abnormalities and genetic defects (though not all) with a high degree of certainty. However, all parents carry a so-called basic 4-7% risk for their child to be born with more or less severe malformations or diseases. The risk of a severe malformation or disease depends on the age of the parents and is around 1%. Even if invasive screening has eliminated the possibility of certain chromosomal abnormalities, a basic risk of other diseases and malformations still exists. Physical malformations, mental handicaps or metabolic diseases usually cannot be ruled out by chromosomal analysis.

- Changes in minute parts of chromosomes or individual genes can not be detected under the microscope.
- If, in very rare cases, a smaller part of a foetus's cells carries an abnormal set of chromosomes (mosaic), this cannot always be reliably detected.
- In just a handful of cases, cell cultures grow very slowly, meaning that the results are available later than expected.
- Very rarely, no analysis is possible. In that case, we would have to discuss the consequences with you and your doctor. If a quick-test has been performed, it may be possible to dispense with a new puncture.

## What should I do after a puncture?

Immediately after any puncture (chorionic villus sampling, amniocentesis, cordocentesis), you should lie quietly in our rest room for at least half an hour. Any complications occurring after a puncture will usually appear within the first 24 hours. We therefore recommend that you stay at home and rest, lying down, on the day of the puncture and the day after. On the day of the puncture and the day after, avoid physical exertion (e.g. sport, heavy lifting, frequently climbing stairs). If you work, have your gynaecologist write you a sick note for these two days. One to two days after the puncture, you should see your gynaecologist for a check-up. If after a puncture you loose fluid or blood or suspect this, experience severe lower abdominal pain or other discomfort, you should visit your doctor or your clinic immediately. If no complications occur after the puncture, you can safely continue to play sport, fly and even sex – unless your doctor has already recommended otherwise.

## What happens if the result is abnormal?

Although the vast majority of children are born healthy, we diagnose an abnormality or disease in a small percentage of unborn children. In this case, we will not of course leave you alone with this result. In this situation, it is important that you get both medical and non-medical advice and support. We, a team of gynaecologists, genetic scientists and psychosocial counsellors, decide with you the best way forward for you and your unborn child. Counsellors from *donum vitae* are engaged in our practices in Düsseldorf and Cologne. If necessary, we consult experts from other areas, such as paediatricians, cardiologists or paediatric surgeons. Whenever you have to make decisions, we offer you all the support you need.

### Your appointment in our practice:

Before we begin with a scheduled screening, we answer all your questions in a one-to-one discussion. Therefore, we would ask you to sign the consent form for the scheduled screening after this discussion.

- We take a lot of time for every screening. In addition, any prenatal screening may require discussions of procedures that we cannot foresee at the scheduling stage. Please understand if the waiting time is occasionally longer than usual. To be on the safe side, you should allow at least two hours for your appointment.
- As well as your maternity record, please also bring along previous findings, lab findings etc.
- After your visit to our practice, you will find a green card in your maternity record. The personal data on this card are very important to the screening procedure and to the quality of our work.

The entire practice team is there to help you before, during and after your appointment.

We look forward to welcoming you  
Your prae-natal.de team



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## prae natal.de in Düsseldorf Pränatal-Medizin und Genetik

Medical Partnership  
 PD Dr. med. P. Kozłowski und Partner  
 Essen District Court PR 1853

### Public transport

Tram 701, 704, 709, 711, 715, 719 or Bus 780, 782, 785  
 Berliner Allee stop

### Parking

Car park at Kaufhof Galeria opposite the practice, open 24 hours  
 Car park at Jecht, Luisenstraße 33, open 24 hours

## prae natal.de in Cologne Pränatal-Medizin und Genetik

Medical Partnership  
 PD Dr. med. P. Kozłowski und Partner  
 Essen District Court PR 1853

### Public transport

Tram 12, 15, Christophstraße / Mediapark stop

### Parking

Underground car park at Kaiser-Wilhelm-Ring 26, open 24 hours