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PRENATAL DIAGNOSTICS AND TREATMENT

START TO LIFE

**Prof. Mirosław Wielgoś, MD**

is a gynecologist. He is currently Rector of the Medical University of Warsaw (2016–2020 term). Prior to this role he was the head of the First Department of Obstetrics and Gynecology, and Dean of the First Faculty of Medicine at the University. His professional interests revolve around issues related to obstetrics, pregnancy pathology, prenatal diagnostics and intrauterine treatment.

miroslaw.wielgos@wum.edu.pl

Prof. Mirosław Wielgoś, MD, Rector at the Medical University of Warsaw, talks about why women of all ages should undergo prenatal testing and where they can turn for help.

ACADEMIA: What is the difference between prenatal screening and prenatal treatment?

MIROSLAW WIELGOŚ: Prenatal screening encompasses all the tests performed during pregnancy, which are aimed at detecting various types of abnormalities in the fetus. Among the different techniques used for this, ultrasound is the most common. This is the basic method for visualization of the fetus, allowing for accurate assessment of fetal anatomy and detection of malformations. There are also ge-

proper pregnancy management, planning the birth at the right time and place. Some defects can be treated after birth, but then the birth must take place in a properly equipped medical facility, because sometimes the operation must be carried out immediately and there is no time to transport the newborn to the appropriate facility. Also, diagnosing even the most serious of defects is not tantamount to terminating the pregnancy, as ultimately such a decision is made by the mother, not the doctor.

So, the woman may choose to continue the pregnancy even if serious defects were detected in the fetus?

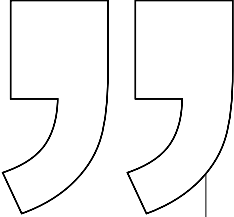
She has every right to do so. It is her choice, but it should be a conscious one. If a woman wants to give birth and raise a sick child, prenatal diagnostics can help her prepare for it, mentally, logistically, and financially. This way she does not suddenly learn of her child's disability at childbirth.

What percentage of pregnancies require intervention at the fetal stage?

In the general population, malformations occur in about 3% of cases. However, statistics include all abnormalities, from cosmetic ones, such as a cleft lip, to serious defects, which represent a small portion of these 3%.

These days an increasing number of older women are getting pregnant and giving birth. Have you observed a related increase in the incidence of fetal defects?

The risk of genetic defects, which medicine is still unable to fix, increases with the mother's age. But these types of defects have various degrees of gravity. They may relate to different chromosomes, but even within the same abnormality, such as the trisomy of chromosome 21, or Down syndrome, the degree of disability can vary greatly. This illness may be accompanied by a whole range of structural defects and delayed intellectual development. However, we are not able to predict the degree of this mental development. Some children do relatively well, others are severely impaired. At the time of diagnosis we cannot tell the mother to what degree her child will be disabled, as we simply don't know yet.



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netic and biochemical methods which can assess the risk of defects and confirm, or exclude, their presence at the chromosome level.

When a defect is detected, in many cases we can begin prenatal treatment. Sometimes it's an actual prenatal correction of defects, and sometimes the treatment includes managing the pregnancy so that the child survives the prenatal period, and can undergo an operation soon after birth.

You mentioned that treatment may be undertaken in specific cases. How many fetuses diagnosed with defects can be helped?

It's difficult to determine the percentage. Everything depends on what type anomaly we are dealing with. I should stress that the notion that detection of fetal defects essentially amounts to abortion is a big misunderstanding. Even if a defect cannot be corrected prenatally, the knowledge of its presence can aid in

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What conditions are usually treated at the prenatal stage?

A prime example of a disorder which qualifies for intrauterine treatment is fetal hemolytic disease, caused by a serological conflict [which Dr. Ewa Brojer from the Institute of Hematology and Blood Transfusion in Warsaw wrote about in *Academia* magazine 2/2014 – ed.]. Thanks to effective prevention measures this conflict occurs less frequently, but we cannot eliminate it completely. With prenatal screening, a test to detect certain antibodies in the blood of the mother, we can detect the condition with non-invasive methods and without jeopardizing the fetus, and then, using the Doppler test, determine the degree of its severity. In the case

the world are currently using the fetoscope method. We are making efforts to introduce this method at our clinic as well.

What determines whether the child is operated on in the womb or after birth?

If the diagnosed defect does not pose a threat to the life of the fetus, then it is easier to wait and treat the child after birth. Usually this type of correction does not constitute a problem for pediatric surgeons. It's a different story if the abnormality can cause irreversible damage, as in the case of an obstructive defect of the urinary tract. It may be caused by a trivial anomaly, such as blocking of the urethra by the valve. This stops the urine from flowing and increas-



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of severe anemia, intrauterine treatment is started, consisting of a blood transfusion directly into the circulatory system of the fetus. Even in a severe form, the hemolytic disease is reversible, provided it is detected early and appropriate treatment is undertaken.

The current spectrum of intrauterine treatment is still wide open. There are increasingly diverse treatments being performed throughout the world today and we would like to make them available in Poland as well. A good example is the myelomeningocele defect, a form of spina bifida. In Poland, this defect is treated only in Bytom, but there the operation is performed through an open uterus. This procedure is very invasive. Several centers around

es the size of the bladder and ureters, which ultimately results in hydronephrosis and kidney damage. This can be prevented by a procedure in which catheters are placed inside the bladder to bypass the valve. This way the kidneys can develop properly, and after birth the child can function normally. There is also another method, which is still not performed in Poland: a fetoscope is inserted into the fetal bladder, which helps pierce the obstructed urethral valve.

A fetoscope is an endoscope is used for fetal intrauterine procedures. It is a tube with a diameter of slightly more than 3 mm, which has three channels: optical, through which we can see the operated area, a working channel, and a channel for fluid delivery.

This is hard to imagine. After all, the fetus is tiny and is connected to the mother's body. It's like operating on two patients at the same time.

This procedure resembles other laparoscopic procedures. Because they require high precision, it is necessary to eliminate fetal movements by providing adequate resources. Of course the fetus also receives painkillers and analgesic agents. The mother is given local anesthesia so she can be conscious and help us, for example, by changing her body position or controlling her breathing.

Intrauterine procedures are performed with an ultrasound, which helps us see what is happening inside the womb. We also use ultrasound with fetoscopic procedures, allowing us to locate the right

untreated, TTTS leads to the death of one or both twins. The most effective method of treatment is a fetoscopic procedure, using a laser to sever these connections and separate the blood flow.

Is your center the only facility performing these procedures in Poland?

We are the only ones who perform the procedure to treat diaphragmatic hernia. Such facilities are also scarce throughout Europe. Diaphragmatic hernia is a relatively rare malformation, but extremely serious, associated with a high mortality rate. It occurs when an opening forms in the diaphragm separating the abdominal cavity from the chest. Because there is a difference in pressure between the abdominal cavity and the chest, the abdominal organs, including the intestines, stomach, and sometimes liver, are pushed through this opening to the chest. There they press on the lungs, which are unable to develop, leaving the child unable to breathe on its own after birth. Even artificial ventilation will not help here because the lungs are rudimentary, and the child dies shortly after birth.

Surgical repair of diaphragmatic hernia is relatively simple, and consists of returning the displaced organs back to the abdominal cavity and mending the hole in the diaphragm. But something must be done to help the lungs develop before birth. Lungs are not needed in utero, but they must have a chance to develop. The following solution was found: a balloon is inserted into the trachea that clogs the airways and prevents the fluid produced in the alveoli and the bronchial tree from escaping, keeping it in the lungs. As a result there is a change in pressure and it's the lungs that begin to press on the intestine, not vice versa. Even if the organs affected by the hernia remain in the chest, they can be operated on soon after birth, because at that point the child actually has lungs, and therefore stands a chance of surviving.

If such a defect is diagnosed in some rural area, how can the patient receive treatment at your center?

Physicians performing diagnostic tests should know that such patients should be referred to us. In Poland there are several similar centers and they specialize in specific treatments. We treat diaphragmatic hernia, at the Bielany Hospital in Warsaw they treat heart defects, in Bytom they specialize in myelomeningocele, or repairing spina bifida, whereas the Twin-to-Twin Transfusion System is performed in Łódź, Gdańsk, and at our center.

These defects don't occur frequently, so there is no point for each center to offer the full set of treatments. Moreover, due to the centers concentrating on different areas, each center takes on a number of

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places, so that after entering the uterus the fetoscope is automatically in the immediate vicinity of the operated area.

Is there a minimum age at which these procedures can be performed on a fetus?

Most procedures are performed in the second half of pregnancy, that is after 20 weeks. This is because these operable defects can only be diagnosed during a routine examination that every pregnant woman must undergo between 18 and 22 weeks of gestation. It is very rare for the hemolytic disease to appear in a severe form before the twentieth week. It may occasionally happen, however, that a serious state occurs earlier, such as in cases of severe defects of the urinary tract. They require immediate intervention to unblock the bladder.

Intrauterine procedures may also be performed earlier in case of TTTS, Twin-to-Twin Transfusion System in a twin pregnancy. It is a common complication of a monochorionic twin pregnancy, where both fetuses share a single placenta, connecting the blood circulation of the twins. One fetus becomes a donor, the second a recipient – one doesn't receive enough blood, while the other gets too much. If left

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cases of one type, which allows for specialization and better proficiency. Two years ago, when we first performed surgery on the diaphragm it took us two hours, and now it takes a few minutes.

Where did you get the idea to perform this procedure at the clinic?

Previously, these types of procedures were not being performed in Poland so patients had to seek help abroad. Even if you managed to get reimbursed by the National Health Fund (NFZ), it still meant incurring additional costs of travel and accommodation. So I thought that, since we have a lot of experience in prenatal treatment, and I believe we perform the most intrauterine transfusions in Poland, as well as treat TTTS, then why not treat diaphragmatic hernia?

I realized that it's difficult, but if it wasn't, anyone could do it. The first step was to bring in special equipment, which proved not to be too difficult. However, it's having the right people that is most crucial. Surgery has to be learned, you can't just train by doing "dry runs" We had to find the right person and it turned out to be Dr. Przemysław Kosiński. He went to London, where he spent two years as part of the team under Prof. Kypros H. Nicolaides. For the first year he never even came close to the operated patients, but luckily the professor eventually recognized his potential and allowed him to assist during procedures. Dr. Kosiński was able to learn from a true master, and my only worry was whether he would return to us after these two years, because he had many attractive offers.

How did the first operation go?

It was very difficult because in London Dr. Kosiński had not performed these procedures himself. He had participated in them, but it's one thing to observe, or even assist, and another to manage it alone. The first few operations were therefore complicated, but we have gained experience and now, given favorable conditions, i.e. if the fetus is in a good position, etc., the procedure lasts a few minutes. The first patient who survived is now two years old and is thriving. We are keeping in touch with his parents.

How do you work with the parents in these situations?

Parents are first told that something may be wrong with their child during the screening tests. At that point, the faster they end up in a specialized center, the better. There, the presence of the defect is verified. In the case of diaphragmatic hernia we can determine the severity of the defect, a hole in the diaphragm may be small, but it may also cover almost the entire diaphragm. We treat only the most severe cases, where the risk of mortality is high. The sur-

vival rate of children with severe diaphragmatic hernias is less than 20% if the defect is not treated, which is a very small percentage. Moreover, the treatment doesn't guarantee that the baby will survive. Although it doubles the chance for survival, there is still about 60% risk of death. I have not yet come across a parent who decides against the surgery.

Parents are willing to do anything to save their child, but we must make them aware that we are not able to guarantee complete success. Even if we insert a balloon into the trachea, the lungs develop, the child is not born prematurely, and will be immediately operated on, its chance for survival is still 40%. We communicate this very clearly, but there are still cases of parents accusing us of negligence.

In the general population, malformations occur in about 3% of pregnancies. However, these statistics include both minor and severe cases.

Sometimes, however, the defect is so severe that the child has no chance to survive, and the pregnancy endangers the mother's life. This is an indication to terminate the pregnancy.

No one should be denied the opportunity to continue a pregnancy with a defective fetus, or to care for a heavily handicapped child, but at the same time, no one should be forced to give birth to such a child. This is consistent with the applicable law, which in some cases permits abortion. Besides, it is never a decision for the doctor, but for the parents to make. If a patient really wants to have an abortion, she will have one, but we should make sure that it takes place in civilized conditions, ensuring safety and proper care. In every case that can be helped, when there is a chance to save the baby, we inform the patient. We offer help and strive to ensure that every patient can take advantage of it. Therefore I stress the importance of prenatal testing, which women of all ages should undergo. Screening is very important. Without it, some children would die in the womb or be born seriously ill.

INTERVIEW BY AGNIESZKA KLOCH

