Prenatal Diagnosis and Pre-Implantation Genetic Diagnosis

1. Introduction

Modern medicine offers various prenatal tests which aim to determine whether particular disorders or abnormalities are evident in the child or not. These examinations are described as “Prenatal Diagnosis“ (PND).

“Pre-Implantation Genetic diagnosis“ (PGD) refers to clinical diagnostics which are performed in the course of artificial insemination prior to the implantation of the embryo into the uterus.

In countries with high standards of medical care some of these examinations are part of routine antenatal care. Other tests are offered or recommended in case of particular risks or specific questions.

PGD is banned in some countries and is legally regulated in most countries.

Parents who are expecting a baby are – like all of us – more or less influenced by their social environment and might see it as the natural thing to do or may even feel obligated to make use of the possibilities offered by PND or PGD, because they are concerned about their child’s health. They are often not aware that this may bring them into conflict and that they could be faced with uncomfortable decisions, as there could potentially be no treatment available and the only remaining option might be an abortion.

In the following sections we will discuss ethical concerns, indicate the limits of prenatal diagnosis, summarise the position of the New Apostolic Church and illustrate methods of prenatal diagnosis in the appendix.

2. Prenatal Diagnosis (PND)

Over the last decades prenatal diagnosis has contributed to lower maternal and infant mortality and undeniably plays an important part:

- It can provide reassurance for parents and deepen their relationship with the unborn child.
- Gene mutations or abnormalities that would lead to life-threatening conditions after birth (for example congenital heart disease) can be identified. This makes it possible to plan the birth in such a way that the child receives optimal care directly after birth. In some cases treatment can already commence before birth.
- If non-life-threatening abnormalities are detected in the unborn child (for instance cleft lip and palate), parents have the opportunity to prepare themselves and find out about support and treatment options.
- Recognising disabilities or untreatable conditions in the unborn child enables parents to confront the disability/disease early on and fosters an attitude of acceptance.
- If possible birth complications are identified (for instance a low-lying placenta or placenta praevia) appropriate birth planning (caesarean section) can prevent life-threatening emergencies.
- If developmental disorders are diagnosed, more closely monitored antenatal care can minimise the risk of impairment.

Nonetheless prenatal diagnosis has to be viewed critically as well and examined carefully:

- Many parents feel socially responsible and sometimes even under pressure during pregnancy to undergo tests that are offered or recommended.
- The fear of a diagnosis of abnormalities can mean that parents will perhaps only accept their new situation in life after 20 weeks of pregnancy and only then build a positive relationship with the unborn child.
- There are indications that parents adopting a wait-and-see attitude can have life-long consequences for parent-child attachment and the development of the child.
- Considering the many disorders and disabilities that could be diagnosed, treatment is - even today – only possible in some cases. If abnormalities or genetic defects are detected, the medical profession often recommends an abortion.
- Prenatal diagnosis cannot guarantee a healthy child, even if the test results don't show any pathological abnormalities.
- Some tests merely indicate a higher risk for certain disorders which might, however, potentially not be confirmed by further examinations.
- Some PND methods pose an additional risk of miscarriage.

In most countries (in Germany this is enshrined in law) people have a right to ignorance and available PND tests can be refused.

The fact that potential disorders and disabilities can be detected by PND should not make us lose sight of the fact that 95 percent of infants are born healthy; and, statistically, out of a hundred women, who are 40 years old when their child is born, only one is expecting a child with Down's syndrome – in younger women the statistical probability is even lower.

Before deciding whether to make use of prenatal diagnostic testing the following questions will be helpful:

- What is our perception of "disability" and "disease", "health" and "normality"?
- What would it mean for us to have a child who is sick or disabled from birth? Who could support us?
- Which procedure-related risks are we prepared to accept in order to obtain information about potential disorders or disabilities affecting our child (e.g. amniocentesis carries a 0.3-1 percent risk of miscarriage)?
- Could we imagine not undergoing any prenatal testing and thereby running the risk of having a sick or disabled child without knowing this before the birth?
- Are we even prepared to consider a possible termination of pregnancy at a later stage (when the child's movements can possibly already be felt and a close relationship with the child has already been established)? This would normally be an induced birth which can lead to enormous physical and psychological strain.

3. Pre-Implantation Genetic Diagnosis (PGD)

Pre-implantation genetic diagnosis is the DNA analysis of embryonic cells created by artificial insemination, prior to implantation into the uterus. Embryos with chromosome abnormalities and genetic disorders are hereby identified and disposed of, so that preferably only healthy embryos are used for the planned pregnancy. This method has been available for over 20 years and has probably been used for conceiving more than 10,000 children worldwide (as of 2012).

PGD is mainly used for identifying hereditary diseases and chromosomal anomalies in couples with a high risk of genetic disorders. Alternatively it can be used to conceive a baby who is a suitable stem cell donor for a sick sibling (“saviour sibling”). PGD is socially controversial; legislation varies between countries and extends from a complete ban to liberal practices without restrictions.

Supporters of PGD perceive it as an advantage that – in the case of artificial insemination - a pregnancy is not started on a ‘trial and error‘ basis, to be either terminated or continued depending on the PND result.

4. The NAC's Position on Prenatal Diagnosis and Pre-Implantation Genetic Diagnosis

The New Apostolic Church is an advocate for life.

Human life begins with the union of the ovum and sperm cell. The fertilised egg already constitutes life – an individual human being with the right to protection.

We also believe that a child's soul is impressionable during the prenatal stage. Just as God protects us and holds us in His hand according to His grace, likewise does the unborn child develop under His protection and according to His grace. As an expression of this realisation the New Apostolic Church offers to dispense a prenatal blessing at the request of the parents. This blessing especially applies to the spiritual development of the new life, but also covers the child's entire prenatal development.

Position on PND

The New Apostolic Church approves of making use of prenatal diagnostics as long as it is beneficial for:

- supporting the healthy development of the unborn child
- preventing damage by identifying disorders early
- planning the right method of childbirth based on the anomalies identified
- improving parent-child attachment
- providing certainty and reassurance for parents
- helping parents who are expecting a sick child to prepare for this situation

From the perspective of the Church those types of prenatal screening tests that carry an increased risk of miscarriage should be avoided. Such types of PND should only be employed in isolated cases, if the life of the unborn child was in serious danger, for example.

Seen from a psychological and a faith perspective prenatal diagnosis can be helpful but can equally be a potential source of stress for parents. The decision for or against prenatal screening is the responsibility of the parents. Accepting the new life, even if the screening tests predict or even prove a child's future disability is crucial from the perspective of Christian faith.

Position on PGD

Pre-implantation genetic diagnosis which is designed to deselect embryos with undesirable physical characteristics and to kill them, is not compatible with Christian ethics.
The Church will always accept the personal decision made by the parents and will offer soul care and support (further information on the topic can be found in the document 'Termination of pregnancy').

5. Short Statement

The New Apostolic Church is an advocate for life. Human life begins with the union of the ovum and sperm cell. The fertilised egg already constitutes life – an individual human being with the right to protection.

Prenatal diagnosis which safeguards and supports the development of the new life is a valuable medical achievement. Methods that compromise or even prevent the uninterrupted development of the child should – from the perspective of the Church – be avoided.

The decision for or against prenatal screening is the responsibility of the parents. Accepting the new life, even if the screening tests predict or even prove a child's future disability is crucial from the perspective of Christian faith.

To deselect life with undesirable physical characteristics and to kill it, is not compatible with Christian ethics.

Appendix

A. Methods of Prenatal Diagnosis

There is a general distinction made between invasive and non-invasive methods.

Non-invasive methods are ultrasound scans and blood tests of the mother which allow - individually or in combination – a personal risk assessment for specific genetic disorders to be made.

Although invasive methods usually facilitate the concrete diagnosis of a genetic disease, chromosomal defect or metabolic disorder, they do, however, necessitate an intervention with the risk of miscarriage.

A.1. Non-Invasive Methods

A.1.1. Ultrasound (Sonography)

An ultrasound scan (sonography) is a harmless procedure, according to current medical knowledge, which generates an image of the unborn child. The timely development of the unborn child can be monitored and multiple pregnancies can be identified.

It is possible to…
- check the placenta and amniotic fluid
- see an image of the child’s body and organs
- determine the gender at a later stage of pregnancy

Several conditions and developmental disorders can also be identified by means of an ultrasound scan. At the same time indications for certain diseases can emerge which have to be investigated further using other methods (for example in the case of genetic defects or viral infections).

A.1.2. 3D Ultrasound

Three-dimensional ultrasound is a computer-assisted conversion from a two-dimensional to a three-dimensional image. This allows a spatial view of the surface of the body and other body structures.

The medical indication for a three-dimensional ultrasound can only be determined by a specialist in the case of specific medical questions (e.g. spina bifida, cleft lip and palate or congenital heart disease).

A.1.3. Doppler Sonography

Doppler Sonography generates an image and produces a measurement of blood flow through the umbilical cord and important blood vessels of mother and child. If there are concerns about the normal development of the unborn child (in the case of high blood pressure in the mother for example), more accurate information relating to the wellbeing of the child can be collected by performing this scan than by the mere measurement of the baby's growth.

A.1.4. Foetal Echocardiography

Foetal echocardiography generates an image of the child's heart and major blood vessels surrounding the heart. It makes the identification of about 85 percent of congenital heart disease possible, even before the baby is born. This opens up the possibility of facilitating the birth under optimal conditions (confinement in a perinatal centre with specialist paediatricians for the treatment of congenital heart; neonatal surgery if necessary).

A.1.5. First Trimester Test (First Trimester = first 3 months of pregnancy)

Nuchal translucency (a collection of fluid under the skin at the back of the baby's neck) can be measured by means of a special ultrasound scan at a specific stage in the embryo's development. An increased amount of fluid can indicate the presence of disorders (e.g. congenital heart disease) or chromosomal defects.

In addition a blood sample is taken from the expectant mother and screened for the pregnancy hormones chorionic gonadotropin (hCG) and estriol as well as the proteins PAPP-A and AFP. The test results, the time of pregnancy and the age of the pregnant woman are taken together and used to calculate the overall risk.

A.1.6. Triple Screen Test/Quadruple Screen Test

The triple screen test screens for levels of the pregnancy hormones hCG and estriol in the blood of the expectant mother. At the same time AFP, a protein originating from the child and present in the mother's blood, is also screened for.

The quadruple screen test is an extension of the triple screen test, which also screens for the hormone inhibin A in the blood of the mother. This parameter aims to reduce the rate of false-positive and false-negative results of the triple screen test. Nonetheless the advantages and disadvantages/risks are the same as those of the triple screen test. Blood tests are carried out between the 15th and 18th weeks of pregnancy. The test results are looked at in the context of the expectant mother's age and the duration of the pregnancy. Hereby the individual statistical risk for Down's syndrome or neural tube defect (spina bifida) can be calculated. In case of an abnormal result the pregnant woman often undergoes further invasive tests (see amniocentesis). The results take longer than the nuchal translucency measurements.

A.1.7. Evidence of Foetal Cells in Maternal Blood

Because foetal blood cells also circulate in the mother's blood it is possible to identify some chromosomal abnormalities, like for example trisomy 21 (also trisomy 13 and 18), or the gender of the child early on in pregnancy by means of a simple blood test.

A.2. Invasive Methods

A.2.1. Chorionic Villus Sampling (CVS) (chorion = outer membrane)

The ultrasound-guided sampling of placental tissue with a thin hollow needle is normally done through the abdomen. Chorionic villus sampling is usually performed between the 11th and 13th week of pregnancy. The risk of miscarriage is approximately 0.5-1 percent. The risk of an ambiguous result which necessitates a follow-up examination is ca. 2 percent, the risk of an incorrect diagnosis is under 0.2 percent.

Chorionic villus sampling is often carried out when a result needs to be available as early as possible or after an abnormal result of a first trimester test. The presence of particular diseases, like metabolic disorders or muscular diseases (muscular dystrophy, cystic fibrosis et al.), which run in families, can also be actively explored by the possibility of examining chromosomes directly.

This method cannot give any indication on the presence of neural tube defect (e.g. spina bifida).

A.2.2. Amniocentesis or Amniotic Sac Puncture

A few millilitres of amniotic fluid are extracted from the amniotic sac with a hollow needle through the abdominal wall of the pregnant woman under ultrasound guidance. The extracted amniotic fluid contains – amongst other things - cells shed by the unborn baby, which are then grown in a cell culture. This procedure is normally carried out in the 16th week of pregnancy. The results are usually available after two-four weeks.

The results of amniocentesis are very precise and incorrect diagnoses are rare. Chromosomal anomalies, like for example trisomies, can be detected – of which Down's syndrome is the most common. Neural tube defects (spina bifida) as well as genetic diseases, e.g. severe muscle and metabolic diseases can also be identified. The risk of miscarriage is between 0.3 and 1 percent.

A.2.3. Umbilical Cord Puncture (Cordocentesis)

Cordocentesis involves a blood sample being taken from the umbilical cord under ultrasound guidance and examined for – amongst other things - specific infections and blood group incompatibility between mother and child. The child can also directly be given medication in this way or a blood transfusion can be carried out in case of anaemia, for instance. The test carries a risk of miscarriage of about 1 percent.

A.2.4. Foetoscopy

Foetoscopy allows the child to be observed through a thin pipe, which is inserted through the abdomen into the amniotic cavity under local anaesthetic. Tissue samples can be taken from the skin or liver in order to exclude or detect very rare skin or metabolic diseases. The examination carries a risk of miscarriage of about 5 percent.