Prenatal diagnosis

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1. Preface

Currently the ratio of genders of new-borns in Albania is 100 girls to 116 boys. Normal would be 100 to 105. These shocking numbers can easily be related to our topic, prenatal diagnosis. In Albania, where girls have a much smaller chance to get a job and earn money later in life, the wish of a young couple to get a boy instead of a girl is huge. Therefore when the doctor tells the young pregnant woman after the ultrasound that it is a girl, the chance of her deciding to get an abortion is tremendous. Sadly in countries like China and India this is also the case. The worst about these not wanted girls is, that the couple will even go as far as getting an illegal abortion. This means the abortion takes place, when the girl is already developed so far, that an abortion is not even allowed anymore. We chose this topic, because firstly we were shocked by such stories and secondly we were interested how far the prenatal diagnosis goes. How many disabilities and diseases can be predicted? And how? In addition we were interested in different methods and if there are any advantages or disadvantages. And last but not least we were fascinated by the thought, that one-day we may be living in a world consisting of “perfect humans”. However, we guess that science is still very far away from this imagined world of perfection, but the question stays: Is such a goal even possible to achieve?

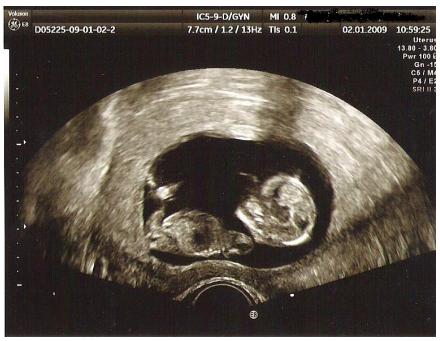
2. Introduction

As mentioned in the preface, the actuality of prenatal diagnosis in countries like Albania is huge. However prenatal diagnosis is an important topic all around the world. The goal of prenatal diagnosis is to determine the health and condition of an unborn fetus.
It includes a variety of different techniques, which will be examined later in this paper. The most common reason for prenatal diagnosis is to check for chromosomal abnormalities (of which trisomy 21 is the most common), genetic diseases and other conditions. The first ultrasonography was performed by Ian Donald in 1958. Since then a variety of different techniques have been developed.

3. Methods

3.1 Ultrasonography

The beyond dispute most used prenatal diagnosis is ultrasonography. It is used since 1958 and has no harmful effect on the unborn or on the mother. The technique by itself works as follows: Sound waves are used to produce an image of the fetus. This picture can then be used to measure the fetus. For example a crease in the neck of the fetus is measured and a probability if the child will have trisomy 21 can be calculated. Of course the gender can also be told. This method gives you a basic idea of the health of the fetus, but is never as precise as amniocentesis, because the genes of the fetus can not be found out with ultrasonography.
Ultrasonography was used in 1958 for the first time to create a picture of an unborn. Of course in 1958 ultrasonography was far away from the easy and common way it is used today. In Switzerland nearly every pregnant woman gets three ultrasounds examinations, in the 10^{th,} 20th and 30th pregnancy week.

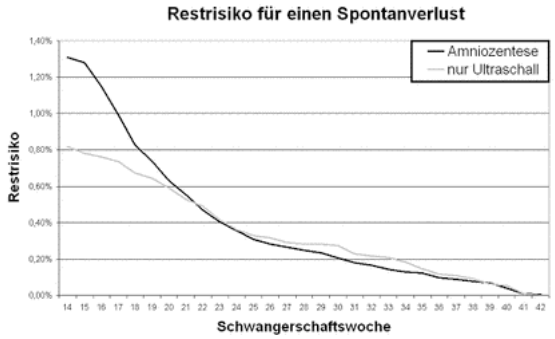


Picture 1: Result from Sonography

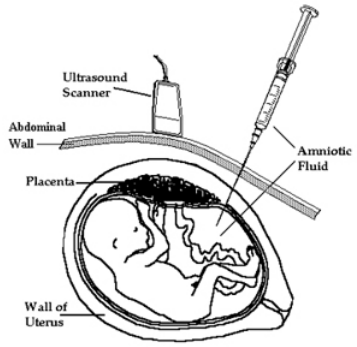

Picture 2: Family having Sonography

3.2 Amniocentesis

Amniocentesis is a prenatal test in which amniotic fluid is removed from the uterus with a needle, which allows gathering information about the health of the fetus. The amniotic fluid is the fluid that surrounds and protects a baby during the pregnancy. This fluid is containing fetal cells, and the fetal DNA is then examined for genetic abnormalities. It can detect nearly all known chromosomal disorders (e.g. Down syndrome) several hundred genetic disorders (such as cystic fibrosis) and neural tube defects. Amniocentesis is usually performed between the 14 and 20 week of the pregnancy. However the risk of having a miscarriage is very low. Nevertheless when such a test is performed too early the risk of harming the fetus is high. The American College of Obstetritians and Gynecologists estimate the loss rate due to miscarriage from the procedure as low as 1 in 300 births.



Picture 3: Risk of miscarriage



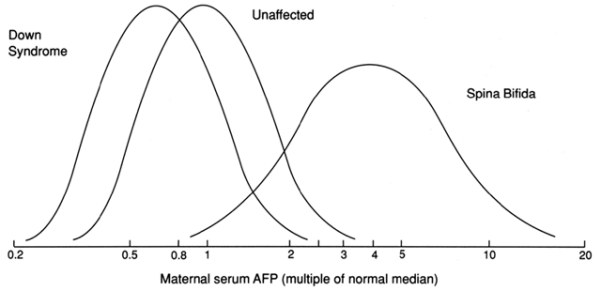
Picture 4: Amniocentesis

3.3 Chorionic Villus Sampling (CVS)

Chorionic villus sampling is a prenatal diagnosis test for identifying chromosome abnormalities and other inherited genetic disorders in the fetus with high levels of accuracy. The testing involves sampling of the chorionic villus (placental tissue) and testing it for chromosomal abnormalities. The test does not identify neural tube defects. There are two ways that samples are collected. Transcervical: A catheter is inserted through the cervix into the placenta to obtain the tissue sample. Transabdominal: A needle is inserted through the abdomen and uterus into the placenta to obtain the tissue sample.
CVS is usually performed between the 10th and 12th weeks of pregnancy. CVS may be chosen over amniocentesis because the procedure collects larger samples and it can be performed earlier in the pregnancy. It also provides faster results than amniocentesis. Although CVS is considered to be a safe procedure complications may occur, as with any invasive procedure. Miscarriage is the primary risk related to CVS occurring 1 out of every 100 procedures.

3.4 Maternal serum alpha-fetoprotein measurement (MSAFP)

Another prenatal test given to most women measures the maternal serum alpha-fetoprotein levels. The test measures the mother's blood for the amount of AFP. AFP is a protein produced by the fetal liver. In 1984 it was reported that low maternal serum alpha-protein levels were associated with Down's syndrome. With this knowledge, the need for more detailed testing can be identified earlier during the pregnancy.



Picture 5: MSAFP

3.5 The newest method: blood test

The newest method has big chances to make amniocentesis completely needless, because it has no effects at all on the fetus. It is now possible to find fetal cells in the blood of the mother that the fetus gave off to the blood of the mother. By finding these rare cells the genes of the fetus can also be evaluated, just as in the amniocentesis, but with no probable damage of the fetus. This blood test is getting very common, because now that it is so easy, every woman wants to test the genes of her child. Opponents of this test say that if it gets so normal to test the genes of a child, then much more abortions will take place.

4. Interview with an expert

As our topic is often discussed not only on a scientific base, but also on an ethical and moral base, we decided to chose an expert who would also discuss prenatal diagnosis not only on the scientific base.
Dr. iur Matthias Bürgin who also studied biology was the perfect expert for our topic. He works at the “Bundesamt für Gesundheit” and his job is as he said himself: “Where law, medical science and ethic meet.”
For the first 30 minutes we talked about what he is recently working on. Even so it doesn’t directly have to do with our topic, we thought it would be enlightening if we would shortly summarize what he is working on, as it is very interesting.
Matthias Bürgin is working on the law of in vitro fertilization of Switzerland. In many countries in vitro fertilization is allowed, but not in Switzerland. Basically in very rare cases, where there is for example a 25% chance of a child to have Cystic Fibrosis (because of the restrictive genes of the parents), the egg cell of the mother is fertilized in a lab with the sperm of the father. That means the whole process of fertilization takes places outside of the body. Then when the Embryo is around three days old one to two cells are taken away from it and genetically tested, if it is affected or not. For this testing there are only 24 hours left, because if it is not affected you want to “put” the embryo in the mother within the same cycle. If 24 hours pass, you will have to freeze the embryo to wait for the next cycle of the mother and the defrosting often goes wrong. Matthias Bürgin is now working on this law, which should allow an in vitro in fertilization in very rare cases. This is of course also in an ethical way very delicate, because people get scared that parents would start to test the colours of the eyes of their future child or similar characteristic traits. Mathis Bürgin explained to us, that this anxiety is legitimate, but in Switzerland this will not be possible, because it will only be allowed to test harmful genetic diseases. And then we started to talk about our topic.

Hello Matthias Bürgin. Thank you for your time. We would like to talk about prenatal diagnosis. First of all we would like to discuss what you think the future of prenatal diagnosis will be. Do you think that in 20 years there will be a big difference from today?

Of course! Today we are able to predict around 300 genetic diseases. The problem is, that today, we are “only” able to predict diseases which have to do with one gene. But we are not able to predict diseases, which are a combination of different genes. I think that prenatal diagnosis is at the beginning of its development. We have to find out more about the diseases, which are produced by a combination of genes. Of course, this is an endless work and we will never be able to predict everything, but in 20 years from now, we will know a lot more. And maybe also new methods will be developed. Such as recently the blood test method, which is very easy.

Do you think that this blood test method will replace amniocentesis?

Yes I think so. I think it already has. It is of course better, because there is no chance of affecting the fetus in a negative way. As you can make this test earlier in the pregnancy, it is therefore easier to get an abortion. Nevertheless, I think we have to think about the costs. I mean the Swiss politicians have to discuss who will be paying these tests, which are now so popular.

Now we would like to talk about what happens if for example trisomy 21 is diagnosed. I mean now that the parents know about it, they have to decide if they want have an abortion or not. Do they really decide freely? Furthermore, we talked about Albania before. Are the parents really taking an autonomous decision?

I don't think so. Of course it should be the case that the parents decide freely, but that nearly never happens. All around the world the pressure of the society is very high. Every couple wants to get a healthy child who will be able to earn money.

Do you think now that parents are able to know if their child has a genetic disease and they don't get an abortion, they will get discriminated by the society?
I don't think so. Especially trisomy 21 is a good example. I would say that 90% of the children born with this disease were cases in which the parents didn't know about it. I also have to say that this is another topic that we have to work on. We are able to predict if a child has trisomy 21, but not the severity of it. That means that many abortions take place, even though the born child would “not be affected too badly” by it. Likewise, this is also a huge leak with other diseases, where we can't actually tell how strongly the born child will be affected. We can only diagnose if it will be affected or not.

Now lastly we would like to talk about a big fear of the population. We think that many people are scared of the idea of a “perfect” and uniform human, that there will be a day, where everything is predictable and only healthy children are actually given born. What is your opinion on this topic?
This is a complete misunderstanding! I can understand that fear, but this will never be possible. As I see it, many people don't know that only the slightest part of diseases and handicaps have to do with genes. Many of the handicaps and diseases come with the ageing of the child and are not predictable. For instance when something goes wrong during the birth of a child, disabilities develop as a consequence. For example if the baby doesn't have enough oxygen and this is obviously not predictable at all and will never be.

Thank you for this great interview and for your time.
You are welcome!

5. Discussion

Prenatal diagnosis gives the mother assurance about the health of her child, but it is also a reason for many abortions. In some cases a prenatal test may raise more concerns about the pregnancy than it can answer. This discussion will deal with the ethnical and moral aspects, as well as the advantages and disadvantages of prenatal diagnosis.

Prenatal diagnosis is considered as a prevention method. Normally, prevention of a disease means that, after prevention happened, there is at least someone who is prevented from getting a disease. However, in the preventive act of prenatal diagnosis the actions are different. The prevention rather includes a person who would have been sick if he would have been born is prevented from ever existing. Whether being spared a sick life, with pain and suffering is a benefit to the child is a difficult ethical question.
As a consequence, the prevention by selective abortion does not result in anyone being helped. Opponents of prenatal diagnosis have the opinion that the question to what extent it is desirable to prevent a person with certain unfavourable characteristics from ever existing should be considered.

Another controversial aspect is how prenatal diagnosis affects family planning. The decisions the woman has to make after a prenatal test can be very stressful. The choices she makes can be life changing. Nevertheless, it can be a huge advantage to prepare for a handicapped child, if the choice to keep the child is made. Moreover it can be a huge benefit to the child, if the diagnosed condition can be treatable before birth.
However, the results can be misused for immoral reasons. As stated in the preface, there are many women who decide to abort their child, simply because it is a girl. Thus, the results have an impact on the attitude to abortion.

What's more, prenatal testing is intended to benefit society. Viewed in terms of medical economics, the burden of genetic diseases on society is essential. "Genetic disorders account for about 20 percent of pediatric hospital admissions and for an even higher percentage of long term admissions" (Simpsons, 1986). However, the problem is that most diagnosable conditions are not able to be treated prenatally. As a result abortion is the only prevention from the burdens of genetic diseases.

A very important aspect is how the doctors influence the decision of the mother or parents. Opponents of prenatal diagnosis state that nobody should be pressured into

having a prenatal test. The choice should not be influenced by the doctor. There is always the possibility of a miscarriage after such an interference and as a result a healthy child could die.

A further controversial aspect is the question if prenatal tests should be available to the whole society, or if it should only be accessible to patients with known genetic or chromosomal diseases in their family history. If it becomes available for the public, it could generate discrimination of certain people and groups with specific characteristics and lead to an approach to the perfect and uniform human.

Concluding this topic is very up-to-date and will become more important in the future, as the techniques are continually improving. The opinions will stay split and how prenatal diagnosis will develop is strongly associated to how the society will evolve and its moral views.

6. Summary

Prenatal diagnosis may improve the quality of life for women, both during and after pregnancy. On the other hand it also has its downsides. The method gets misused, such as in countries like Albania. Still it can be very helpful for making a decision for abortion. In any case prenatal diagnosis has an impact on women in a number of ways, which are not always positive.
The ethical aspects to this topic are very controversial and the opinions will stay split. Furthermore, prenatal diagnosis still has a big potential to improve in the future.

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