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The History of Genetics Research- from Mendel's Discoveries to CRISPR on the example of prenatal tests

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Abstract: *The aim of the article is to show the beginnings and development of research on genetics at the turn of the last two centuries. The article shows, that it could take up to 100 years from the invention of an innovative method to its implementation and practical use for humanity. An example of this is the proof of inheritance traits by G. Mendel in 1865, the discovery of which was popularized in 1956. A modern method in CRISPR genetics, which may be officially approved in genetic medicine in 10 years, was discovered in 1987 by a Japanese scientist Yoshizumi Ishino.*

Working on innovations in medicine requires patience and many years of laboratory experience and medical experiments. These are years of work of many generations of doctors, biologists and geneticists. Genetics opens up new possibilities for humanity to prevent the incidence of many diseases that can be eliminated by modifying human genes. Through the development of prenatal genetics, it will be possible to eliminate many diseases of the child while still in the womb.

The article shows the role and importance of the precursors of genetics and its evolution.

A review of the literature on the subject made realize how many people from the world of medicine and biology are working on innovative methods in genetics. This field is mainly dealt with by centers in Europe, the USA, China and Japan. The article is of significant historical significance, which initiated the development of genetic research.

The article consists of 5 parts. Introduction to the subject of genetics and its development from the nineteenth century to the twenty-first century, the second part is devoted to Genetic diagnostics – what it is, what are its types and importance. In the third part I made an introduction to the subject of prenatal testing (development, types, types, characteristics), and then I analyzed the role and importance of prenatal tests and the role of genetic counselling on the example of the in-depth analysis of the literature. In the end, I gave research conclusions and recommendations.

Keywords: *genetics, prenatal tests, genetics research, DNA, medicine.*

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I. INTRODUCTION

The subject of my article is devoted to genetics. The genetics can first of all detect hereditary diseases early, and secondly, knowing one's own genetic predispositions allows for early detection of diseases such as cancer, type 1 diabetes, heart disease or neurological diseases. Genetic testing can reveal the carrier of mutations that can affect the health of future children – e.g. cystic fibrosis, spinal muscular atrophy (SMA) or fragile X syndrome.

Early diagnosis of genetic and developmental defects in the foetus allows to detect trisomies (e.g. Down's, Edwards', Patau's syndromes) as early as the 10th week of pregnancy, without any risk to the baby. This means increased pregnancy safety, as early knowledge of potential problems makes it possible to prepare an appropriate medical and delivery plan, which can improve the prognosis. Tests can confirm that the pregnancy is going well, which gives mental peace and a sense of control, and if serious defects are detected, parents have more time for medical consultations, decisions and the organization of specialized care (Ashley, 2021).

For humanity, the description of the human genome has become a milestone primarily in medicine – in the prevention, diagnosis and treatment of many diseases.

Genetics can be divided into different types, depending on what is the object of its study (https://www.britannica.com/science/genetics):

1. **classical genetics, population genetics** – these branches of genetics deal with genetic diversity and genetic variability within a population;
2. **molecular genetics** - this branch of genetics deals with the study of the structure and function of genes at the cellular level;

3. **Cancer genetics** – this branch of genetics, on the other hand, studies cell cycle disorders that lead to cancerous transformation.

The subject of my article is the first type of genetics, because I will conduct an analysis of prenatal diagnostics in the light of the literature on the subject. My research goal is to prove the thesis that genetic diagnostics has an impact and importance on human health already in the prenatal phase.

This thesis will be proven on the example of the analysis of literature (scientific articles and books) in the field of prenatal testing.

The main research problem is the development of prenatal tests and their importance for future parents and their offspring.

The research question of my article is: What are prenatal tests and why are they important?

This is looking at those 3 statements:

1: Genetic testing provides valuable information about patients' health and genetic predisposition.

2. Prenatal tests assess the genotype of a child before it is born.

3. Prenatal testing is crucial in diagnosing fetal health and detecting genetic defects, as it helps detect congenital genetic defects.

1. INTRODUCTION TO THE ISSUES OF GENETICS IN THE LIGHT OF SCIENTIFIC LITERATURE

Genetics in medicine deals with the study of the influence of genes on human health. Research on genetics is primarily used to determine the etiology of diseases and determine which diseases are encoded in genes. Thanks to research on genetics in medicine, scientists are able to determine the mechanisms of inheritance of genetic diseases and determine appropriate methods of their treatment. The achievements of genetics make it possible to diagnose the likelihood of genetic diseases already in the prenatal phase (Yang, 2019, p.388).

1.1.Development of genetics

The beginning of genetics was given by Gregor Mendel, an Austrian monk and scientist, who in the years 1856–1863 conducted experiments on peas (Pisum sativum) in the monastery garden in Brno. He crossed different varieties of peas according to colors, seed size or flowers. He noticed certain mathematical regularities and described three laws of heredity, which today we call Mendel's laws: 1. the law of purity of gametes, 2. the law of segregation (separation of alleles), 3. the law of independent segregation of traits (Mendel, 1865).

Table 1: Mendel's three laws

NR	Name of the law	The main idea	Example
1.	The law of gamete purity	F1 offspring are uniform (one trait dominates)	Parent 1: Red flower (genotype **AA)** Parent 2: white flower (genotype **aa**) Offspring (F1): all have the Aa genotype – red flowers, because red is dominant
2.	The law of allele segregation	In F2, the recessive trait returns in the 3:1 ratio	continuation of the F1 generation (i.e. Aa x Aa): Possible genotypes in F2: **AA, Aa, Aa, aa** Phenotypically: 75% red flowers, 25% white flowers
3.	The law of independent segregation of characteristics	Traits are inherited independently (as long as they are not coupled)	If we study the color of flowers (red/white) and the shape of seeds (smooth/wrinkled), then their combinations in the offspring can occur in any configuration (e.g. red + smooth, white + smooth, etc

Source: Own study based on Mendel, 1865.

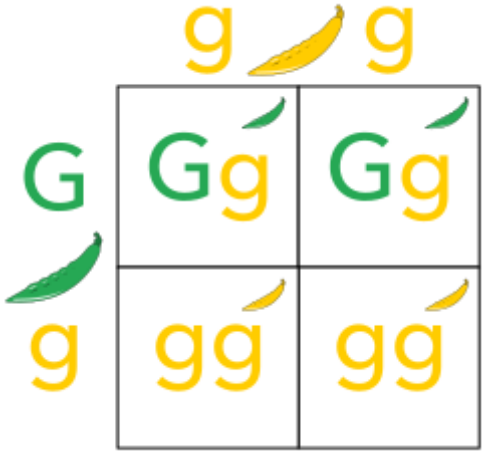
Although Mendel published his results as early as 1865, no one paid attention to them for many years. It was not until the beginning of the 20th century that his work was "rediscovered" and appreciated – then modern genetics began to develop.

On the basis of Mendel's model, the so-called sex-linked traits, including sex-linked diseases, can be explained. Sex-linked diseases are determined by the presence of alleles on the X chromosome. In the case of recessively inherited diseases, in order for the disease to manifest itself in a woman, both X chromosomes must have a recessive allele (the woman must be homozygous, a heterozygous female is a carrier), while the disease will manifest itself in males because they have only one X chromosome (the male is a hemizygous).

In 1905, the English biologist William Bateson (1861-1926) introduced the term genetics as a new field of science dealing with the inheritance of biological traits – based on Mendel's "rediscovered" principles (Crew,

1967, p.13). Together with the English geneticist Reginald Crundall Punnett (1875-1967), he discovered genetic coupling (trait coupling) and contributed to the popularization of genetics and its commercial use (Ibid).
A Punnett square showing a typical test cross. (Green pod color is dominant over yellow for pea pods in contrast to pea seeds, where yellow cotyledon color is dominant over green (Mendel, 1865).

Diagram 1. Punnetts's square



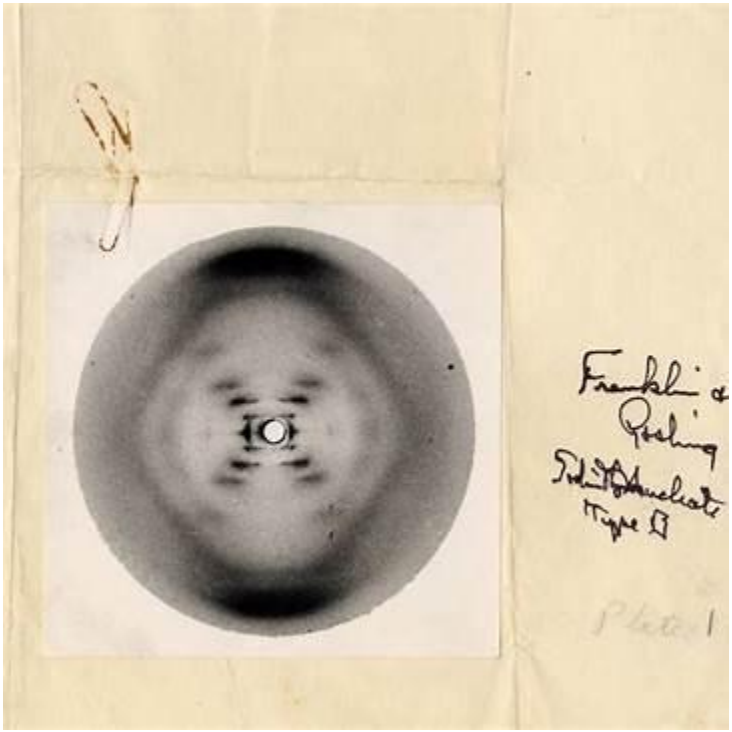
Source: Fairbank, 2012, p.222.
Punnett squares for each combination of parents' colour vision status giving probabilities of their offsprings' status, each cell having 25% probability in theory.

Table 2. Punnett crossword puzzle

A | a --- A | AA | Aa --- a | Aa | aa	**Genotype** **Phenotype (appearance)** **Proportion** **AA** red flower 1/4 **Aa** red flower 2/4 **aa** White Flower 1/4 **End Result (phenotypically):** 1. **75%** (3/4) of the plant will have **red flowers** 1. **25%** (1/4) the plant will have **a white flower**

Source: F.A.E. Crew, *Reginald Crundall Punnett. 1875–1967. „Biographical Memoirs of Fellows of the Royal Society", 13, 1967.*
Then, in 1910, Thomas Hunt Morgan (1866-1945) demonstrated that genes are on chromosomes and explained the genetic basis of sex determination *(A Dictionary of Scientists*). He did this research on a fruit fly. His discovery is a link between a specific gene and the X chromosome – he studied the inheritance of eye colour in flies.
The beginning of the development of genetics was the demonstration by Thomas Hunt Morgan (1866-1945) that genes are on chromosomes and the explanation of the genetic basis of sex determination (Ibid.).
The name gene itself (i.e. from the Greek genos – beginning) was introduced in 1909 by the Danish scientist Wilhelm Johannsen (1857-1927), who used this term to describe certain elements found in the nucleus of a cell of a higher organism, conditioning the occurrence of a specific morphological feature (Johannsen, 1909, p.123).
At the same time, Johannsen also proposed the term "allele", applying it to different variants of the gene. The concepts of gene and allele appeared in 1927, during a lecture at the International Congress of Genetics in Berlin (Nils, 1983, p. 482).
In 1952, Raymond Gosling, a graduate student at Rosalind Franklin, took a crucial X-ray called "Photo 51".

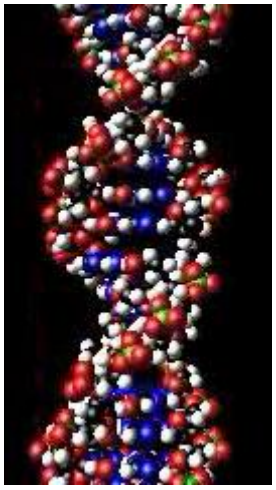
Photo 1 : Photo 51



Source: National Institute of Health

This X-ray image showed the diffraction pattern of X-light on DNA fibers, revealing the characteristic shape of the helix. This image was decisive for understanding the structure of DNA (Franklin, Gosling, 1953, p.740).

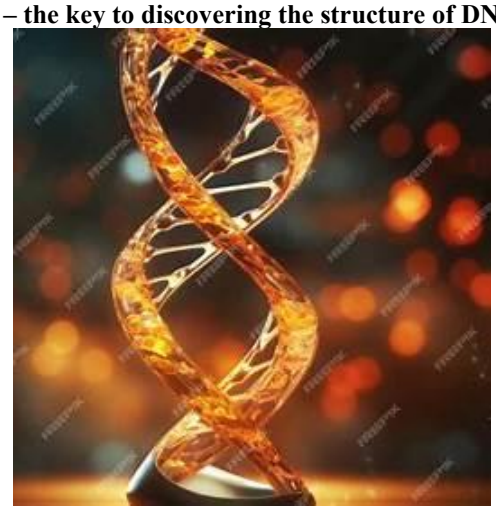
Photo 2: Model of DNA



Source: National Institute of Health

In 1953, James Watson (American) and Francis Crick (British) described the structure of the DNA molecule – deoxyribonucleic acid, which stores the genetic information of all living organisms.

Photo 3. Photo 51" – the key to discovering the structure of DNA – the DNA model



Source: National Institute of Health

Their discovery showed how DNA acts as a carrier of hereditary information and how it can copy itself and pass it on from generation to generation. (Watson, Crick, 1953, p. 737).
The structure of DNA was described as **a double helix** – two twisted strands that form a stable, spiral form. Each strand is made up of repeating units called **nucleotides**, each of which contains:

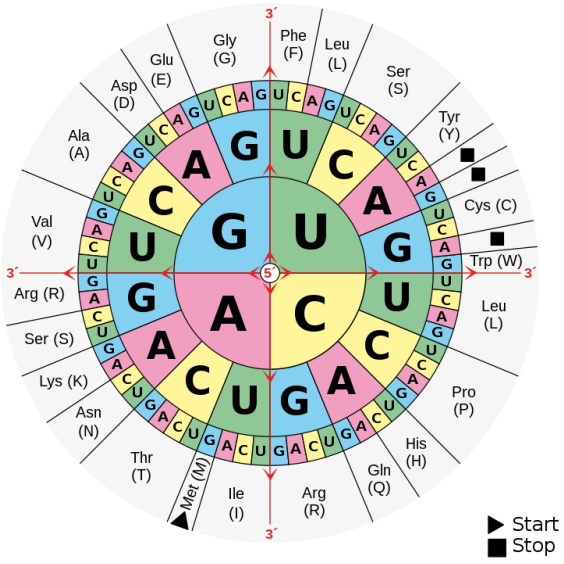
1. deoxyribose sugar,
2. phosphoric acid residue,
3. one of the four nitrogenous bases: adenine (A), thymine (T), cytosine (C) or guanine (G).

Nitrogenous bases combine in pairs: A with T and C with G, forming the so-called hydrogen bonds. These base pairs form the "rungs" in a ladder-like structure, and the skeleton is made of sugars and phosphates, forming the "frames" of this ladder.

- A - T
- T - A
- G - C
- C - G

The sequence of these letters (A, T, G, C) is like a "code" that tells the cell how to make proteins and thus how the body works.
The determination of the physical structure of DNA by James Watson and Francis Crick laid the foundations for the development of molecular genetics (Ibid.).

Diagram 2. Genetic code diagram



Source: National Institute of Health

After Watson and Crick discovered the structure of DNA, scientists began to understand how genes affect health and disease. That was the moment when genetics ceased to be just a theory and began to have real medical applications.

In the 60s of the twentieth century research on hereditary diseases began, e.g. the first newborn screening tests were created (Guthrie, 1961, p. 864).
In 1973, Herbert Boyer, an American biotechnologist, together with Stanley Cohen, introduced the human gene into bacterial cells and created the first recombinant DNA molecule (recombinant DNA) (Cohen et al., 1973, p.3240). So, from the 70s of the twentieth century, the development of genetic engineering, gene cloning, DNA testing, in vitro began.
In 1983, Kary Mullis (American biochemist, Nobel Prize laureate) invented the PCR method that allows copying DNA fragments (polymerase chain reaction (PCR). It is a technique that allows for quick and repeated copying of a selected fragment of DNA in laboratory conditions. Thanks to PCR, millions of copies of DNA can be obtained from a very small sample – e.g. from a drop of blood, saliva, hair (Mullis, Faloon, 1987, p.335).
During this period, research on hereditary diseases such as phenylketonuria (PKU), cystic fibrosis, sickle cell anemia, Down syndrome (trisomy 21), cytogenetic techniques, chromosome studies, were developed to detect chromosome number disorders (https://www.genesis.pl/).

2. GENETIC DIAGNOSTICS

In the 21st century, genetic diagnostics began to develop. It is a branch of medicine that involves the analysis of genetic material (DNA) to detect mutations or abnormalities in genes, chromosomes, or the entire genome. This makes it possible to assess the risk of developing genetic diseases, diagnose genetic diseases, and adjust treatment to the patient's genetic profile (Marques, 2010,p.238).
Genetic diagnosis can concern single genes, entire chromosomes or entire genomes.
In the area of my project, prenatal diagnostics can be distinguished, which examines the genes of the fetus during pregnancy. It can be invasive or non-invasive (Ibid.).
With genetic diagnostics, diseases can be detected earlier (Roberts & Middleton, 2018).
Prenatal tests are tests performed during pregnancy to assess the health and development of the fetus, detect genetic diseases or congenital malformations, assess the risk of trisomy (e.g. Down syndrome), metabolic diseases, heart defects, etc. This is the result of the work of many doctors and scientists developing the technique over decades.

2.1. Invasive prenatal tests

The development of prenatal tests took place as early as the 1950s, when the first attempts were made to collect amniotic fluid for diagnostic purposes. In 1956 – German gynaecologist Fritz Fuchs and his colleague Pol Tietze are considered to be among the pioneers who were the first to describe the safe collection of amniotic fluid for the diagnosis of fetal sex and genetic diseases. They were pioneers in the field of prenatal diagnosis, especially in the field of fetal sex determination and prenatal blood type determination (Fuchs & Riis, 1956, p.330).

In the 1960s, amniocentesis began to be used more widely as a diagnostic tool, m.in. to assess fetal lung maturity and identify certain genetic defects, such as Down syndrome (Haddow, 1992).
Invasive tests – they are more precise, but they are associated with some risk for the mother and fetus. They are performed when the results of non-invasive tests indicate an increased risk of genetic defects.

Table 3. Type of medical examination -invasive prenatal procedure

Type of medical examination	description
Amniocentesis	It involves taking a sample of amniotic fluid containing fetal cells. This allows for a thorough analysis of the genetic material
Chorionic villus sampling	involves taking a sample of chorionic villus tissue, which has the same genetic material as the fetus.
Cordocentesis	It involves taking a sample of umbilical cord blood, which allows for the analysis of the genetic material of the fetus.

Source: Own study based on Roberts, Middleton, 2018.

2.2. Non-invasive prenatal tests

In the 80s of the twentieth century, the development of ultrasound examinations and biochemical markers in the mother's blood took place two huge milestones in prenatal diagnosis.
Ian Donald, a Scottish gynecologist, he is considered the "father of ultrasound in obstetrics". Together with Tom Brown, he developed the first ultrasound devices. They used an industrial apparatus (previously used to detect engine defects) for medical examinations. In 1958, the first scientific paper on the use of ultrasound in pregnancy diagnostics was published (Donald, MacVicar, Brown, 1958, p. 1188).

Professor Stuart Campbell, who collaborated with I. Donald, developed 2D ultrasonography, and later also 3D and 4D ultrasonography, and contributed a lot to the imaging of fetal structures (Campbell, 2005, p.12). His work had a huge impact on prenatal diagnosis and the development of fetal medicine.

Dennis Lo, a physiologist from Hong Kong, discovered the presence of free fetal DNA (cffDNA) in the mother's blood in 1997. This discovery sparked the development of non-invasive prenatal testing (NIPT) (Lo, Y. M. D. et al., 1997).

Nicolaides Kypros is a professor of fetal medicine. He is known for his research on nuchal translucency (NT) and the use of biochemical markers in the early detection of Down syndrome (Kypros, 1994).

Howard Cuckle, a British geneticist, is a co-author of the triple test and the composite test, which use markers such as (Cuckle, 2003, p.636):

1. PAPP-A (Pregnancy-Associated Plasma Protein A).
2. β-hCG (free chorionic gonadotropin subunit).

After 2010, non-invasive genetic testing (NIPT) emerged, which can detect genetic diseases from the mother's blood, without risk to the baby (Chitty S., Hill M., Lewis M., et al., 2015, p.113).

Table 4. Non-invasive tests

Prenatal Ultrasound (USG)	It is used to assess the development of the fetus and detect possible abnormalities in the anatomical structure.
Dual test	involves determining the level of two substances in the mother's blood: free beta-hCG and PAPP-A protein. This helps to assess the risk of Down syndrome, Edwards syndrome and Patau.
Genetic testing NIPT (Non-Invasive Prenatal Testing)	analyze free fetal DNA present in the mother's blood, allowing the detection of trisomy of chromosomes 21 (Down syndrome), 18 (Edwards syndrome) and 13 (Patau syndrome), as well as other chromosomal abnormalities. These tests are available from around 10 weeks of pregnancy and are highly sensitive and safe.

Source: Own study based on Roberts, Middleton, 2018.

2.3. Examples of diseases that can be treated prenatally

1. **Pompe disease**

It is a rare metabolic disease that leads to the accumulation of glycogen in the muscles and heart, which can lead to organ failure. As part of the experimental study, infusions of the enzyme directly into the fetus through the umbilical cord were carried out, which allowed to inhibit organ damage and improve the development of the baby after birth.

2. **X-linked hypohidrotic ectodermal dysplasia (XHED)**

It is a genetic disease that causes, m.in other things, a lack of sweat, teeth and lacrimal glands. The first successful prenatal treatment of this disease involved the introduction of protein into the amniotic fluid, which improved the development of sweat glands and teeth in newborns.

3. **Beta-talasemia**

It is a blood disease that causes severe anemia. Experimental studies have used prenatal stem cell transplantation or gene therapy to correct a genetic defect before birth.

4. **Genetic hearing loss**

Some forms of congenital hearing loss can be treated with gene therapy before birth. Research on the use of gene therapy to treat hearing loss is promising.

5. **Cystic fibrosis**

It is a genetic disease that affects the functioning of the lungs, pancreas, and other organs. Experimental gene therapies delivered prenatally have shown promising results in improving organ function in mice, offering hope for future use in humans.

3.THE ROLE AND IMPORTANCE OF PRENATAL TESTS ON THE EXAMPLE OF THE ANALYSIS OF PREVIOUS STUDIES (ANALYSIS OF THE LITERATURE)

Over the past few years, numerous studies have been published on the role of prenatal testing (NIPT) in prenatal diagnosis.

There are different types of prenatal tests. The role of prenatal tests is enormous. Thanks to them, it is possible to detect diseases such as:

1. Trisomy 21 (Down syndrome),
2. Trisomy 18 (Edwards syndrome),

1. Trisomy 13 (Patau syndrome),
2. Sex chromosome aneuploidies (e.g. Turner-Klinefelter syndrome),
3. microdeletions and other rare diseases.

Thanks to these tests, genetic and developmental defects can be detected early, as they allow to identify diseases such as Down syndrome, Edwards syndrome or neural tube defects.

In addition, they allow to assess the growth, structure of organs and the functioning of the placenta as well as the amount of amniotic fluid.

4. BENEFITS OF PRENATAL TESTING AND THE ROLE OF GENETIC COUNSELING

The prenatal testing gives future parents time to prepare themselves mentally, emotionally and medically (sometimes also for the decision to continue or end the pregnancy in the case of severe defects). In some cases, it is possible to schedule intrauterine surgery, delivery in a specialized center, or treatment of the newborn immediately after birth.

Another benefit of prenatal testing is the fact that some birth defects, such as neural tube defects, can be detected as early as the first trimester of pregnancy. Early diagnosis allows for actions that can improve the baby's prognosis, such as surgical interventions in the womb. Currently, research is being conducted on the treatment of some genetic diseases while still in the womb. Although this technology is in the experimental phase, it already gives hope for effective intervention before the birth of a child.

Due to the fact that the topic of hereditary diseases in the foetus is a very sensitive topic, many centers and clinics provide genetic counseling to support parents who find out during pregnancy that their child may have a serious genetic disease (e.g. Crouzon's, Down's, Edwards' syndrome, etc.).

Genetic counseling is a specialized medical consultation, conducted by a clinical geneticist (sometimes in cooperation with a psychologist), aimed at:

1. explanation of the nature of the genetic disease,
2. determining the risk of its occurrence in the child and future siblings,
3. presentation of possible medical, diagnostic and therapeutic options,
4. providing emotional support and helping to make decisions about further pregnancy or child care in the future.

In general, it aims to prepare parents for the birth of a sick child, the consequences of a given disease for the family's lifestyle, ways of caring for and coping with this problem.

Counselling is not 'evaluative' – it is not intended to suggest to parents what to do, but to help them understand the situation and make informed decisions.

4.1. Analysis of the literature on the subject

The article "Psychological and social consequences of non-invasive prenatal testing (NIPT): a scoping review" by Valérie Labonté, Dima Alsaid, Britta Lang, Joerg J. Meerpohl (Labonté et.al., 2019) describes the results of research on the psychological and social consequences of NIPT. Research shows the need for appropriate psychological support for women who receive positive results to help them make informed decisions about how to proceed.

In turn, Steffensen, et al., in the article "Impact of a prenatal screening program on the Down syndrome phenotype: An interrupted time series analysis (Steffensen et al., 2023) describes the research conducted on the impact of the introduction of a screening program on the phenotype of Down syndrome. The results indicate a decrease in the number of births of children with Down syndrome and an increase in the number of prenatal diagnoses, which suggests the effectiveness of the program in identifying cases before birth.

The article "*Recent trends in prenatal genetic screening and testing*" by Ondrej Pös, Jaroslav Budiš, Tomáš Szemes (Pös, Budiš, Szemes, 2019) discusses the latest trends in prenatal genetic testing, with a focus on NIPT. The authors highlight the increasing availability and acceptance of these tests in clinical practice, as well as their potential to identify chromosomal aneuploidies. The tests also have an impact on the safety of the mother and the child, because early detection of problems allows for the prevention of pregnancy complications (e.g. pre-eclampsia, serological conflict).

They serve to protect the health and life of the child and mother through early diagnosis. There are studies on the impact of prenatal testing on the safety of mother and baby, especially in the context of early detection of pregnancy complications such as pre-eclampsia. They confirm that early diagnosis of these problems allows for more effective pregnancy management and reduced risk of complications (Vikraman, Elayedatt, 2022, p.1808).

A novelty in the development of prenatal medicine is CRISPR (Redman et.al. 2016). Currently, experimental research (on animal models) is underway on the use of CRISPR to treat serious genetic diseases even before

birth. potential gene therapy in utero (in the womb). This applies to, m.in others, diseases such as: beta-thalassemia, cystic fibrosis, DMD (Duchenne muscular dystrophy).

Photo 4. CRISPR Cas9 proteins and gene editing

Source: https://www.sigmaaldrich.com/PL/pl/products/molecular-biology-and-functional-genomics/gene-editing-and-functional-genomics/crispr-gene-e [Access: 6.06.2025]

These therapies are at a very early stage of development, but their results are extremely promising – especially in the treatment of severe diseases of the brain, lungs and blood. It is assumed that it will be available to humans in about 10 years. CRISPR and gene therapies are just entering the clinical phase, but they show great potential, especially in immunotherapy and genetic correction. However, they still need to be improved to meet the requirements of safety and precision.

5. RESULTS AND DISCUSSIONS

Genetics as a field of medicine has been developing since the second half of the nineteenth century. Its precursors are considered to be G. Mendel, W. Bateson, R. G. Punnett, T. H. Morgan, W. Johannsen, R. Franklin, R. Gosling, J. Watson and F. Crick.

A hundred years later (l955-56), prenatal diagnosis was initiated, when amniocentesis was used for the first time to assess the sex of the fetus (in cases of risk of X-linked diseases). In 1966 Prof. Fritz Fuchs (Germany) and Dr. Povl Riis (Denmark) were the first to perform amniocentesis to diagnose a genetic disease (Down syndrome), and since 1997 non-invasive tests have been performed. This gave rise to modern prenatal diagnostics, which is still developing today.

Table 5. Summary of the genesis of prenatal tests

Year	Event	Place
1956	First use of amniocentesis	Denmark/Germany
1966	First genetic diagnosis of the fetus (amniocentesis + karyotype)	Denmark
1972	The beginnings of ultrasound in pregnancy	Sweden / Great Britain
1980+	Biochemical tests (AFP)	USA, Europe
1992–1995	Genetic ultrasound (NT) as a screening test	Great Britain, Germany
1987	CRISPR	Japan
2011	First NIPT (Fetal DNA from Maternal Blood) tests	USA, China

Source: Own elaboration

In the 21st century, genetic diagnostics began to develop from invasive tests (amniocentesis, chorionic villus sampling) to non-invasive (NIPT) – from the mother's blood (safe, modern).

To sum up, it should be emphasized that prenatal tests are about identifying mutations, not about repairing them. Therefore, methods such as Sanger sequencing, NGS (Next Generation Sequencing), karyotype, MLPA, WES (Whole Exome Sequencing) are still used in prenatal diagnostics. These methods detect mutations rather than correct them.

Prenatal genetics are developing rapidly, and genetic testing in pregnancy has great potential – both medically and ethically. But it is worth using them consciously and responsibly. For this reason, I want to point out :

1. There must be widespread availability of screening tests.
2. They should be included in standard medical care during pregnancy, at least for women at higher risk.
3. There should be a staged diagnostic approach – first non-invasive tests (more safe), and only then invasive tests (if necessary).

4. There must be access to reliable information and genetic counseling.

Prenatal testing is an incredibly valuable tool, but its strength lies not just in the technology, but in how it's used — with knowledge, empathy, and respect for parents' decisions.

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