



## RESEARCH ARTICLE

### FAMILY PLANNING AND GENETIC COUNSELLING IN MACEDONIA

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#### ABSTRACT

The family planning becomes very important issue in every society. It is linked with many different aspects. In this article are presented the purpose and the components of the genetic counselling. The main goal of genetic counselling is to set specific and accurate diagnosis with information regarding the aetiology and recovery risk, manifestations, treatment and reproductive alternatives. Also the components of genetic counselling and the need of collecting genetic information about the health of each child are emphasized. The presentation for every day practices and activities in Genetic counselling institution in the Republic of Macedonia is explained. Also are explained the indications for genetic counselling and the process of how the genetic counsellor should talk to people about their actions and possible alternatives. The forms of prevention which exist in some countries, and also in the Republic of Macedonia, are expressed as relationship for the protection of healthy children and to avoid hereditary diseases. The system of prevention and care should be regulated especially in environments where there are health, social and other services and institutions that monitor these aspects of the health and planning of the family.

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#### INTRODUCTION

Family planning is a conscious activity of the people of reproductive age who want not only to regulate the number and time of birth, but to have a healthy child who will meet expectations throughout the life cycle. Family planning involves different aspects - biological, medical, demographic, sociological, psychological, economic, ethical, political and other. In terms of the carriers occurs on three levels: as individual practice, as a movement of social groups and as a program for family planning at the national level. Basic right of all couples and individuals is to decide freely and responsibly about the number and timing of births of children and to be informed and educated about it. The concept of "family planning" has replaced the concept of "birth control" as a modern approach to the development of the family and the population in a country. The preparations for a healthy pregnancy are result of the general progress of modern society. The reason for this is the increased birth control, family planning, and thereby increasing concern about the birth of a live and healthy child. Preparations for a healthy pregnancy may have prophylactic, diagnostic or therapeutic nature. The goal is to determine the health status of the mother, to anticipate possible complications during pregnancy and ensure

the healthy development of the fetus. The Genetic Counselling makes up the overall process of exchange of information relating to medical and genetic prediction. Genetic advice is applicable to all possible phenotypic manifestations mutant genomes, regardless on category affiliation of respondents' backlog development. It is actually a set of measures which are implemented in primary prevention as a result of lag in mental and physical development, whose causes are genetically determined. Preventive activity is commonly used in mental backlog due to the frequent presence of mental disorders across the various syndromes and diseases. The genetic counselling is defined by many authors as a process of giving answers by the doctors' geneticists who in the role of counsellors answer numerous questions about diseases that are, or may be of genetic origin. In communication with the patient who is interested in genetic counselling through his first hearing and through specific research establishes the diagnosis of hereditary disease. Mostly through the understanding of the risk of disease on hereditary base the relationship of doctor and patient are established by the process of genetic counselling. With doing research on the family tree and the changes that have been observed are endeavouring to come to the history of the emergence of a possible hereditary disease. Also Genetic counseling is defined (Definitions of Genetic Testing, 2008-09) as the process by which the patients or relatives at risk of an inherited disorder are advised of the consequences and nature of the disorder, the probability of developing or

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transmitting it, and the options open to them in management and family planning. This complex process can be separated into diagnostic (the actual estimation of risk) and supportive aspects. The scenario-based decision on the advice or suggestion (Clark, 2014) for the couples will be driven in a position to decide on the creation of future descendants, which is essential suggestion for making the decision. The conflict that could be caused to the effect of that decision in the consciousness of patients (especially the mother) may assist in the determination to accept genetic counselling as a reality and to break on time the risk pregnancies that could result with hereditary anomalies. Conversely if the patient despite the given genetic council has accepted the risk of genetic disease findings, knowingly accepted to regret and pity for years because it chose. The decision to abort the pregnancy, which is stated sex chromosome anomaly (which may be with minor importance compared with Down syndrome), is influential on the history of the person who is addressing for genetic counselling.

### **The purpose of genetic counselling**

Diagnosis and risk assessment reappearing in family planning seems to express the purpose of genetic counselling. Genetic counselling is a process that emphasizes the emotional aspect of the counselling and support. As with all processes representing pronounced emotional implications for the health and lifestyle, it is necessary to take account of a multitude of important factors such as:

- Possibility the advised person to accept and understand complex information, often amid difficult and emotional burden and confusion;
- Necessity the information to adjust the level of education of the advised and understandable to it;
- Possibility to conduct an appearance of conflict of social, religious, cultural, economic and psychological point of view and ways of their prevention;

The advised should be unceasingly through the whole process of counselling to be encouraged to freely and openly express his emotions and conflicts. Genetic counselling can be defined as a communicative process that is undertaken in order to assist the affected and his family:

- They accept and understand the medical facts (diagnosis, prognosis and treatment);
- Has understand the contribution of hereditary component to the disease and the disease reappearing in relatives;
- They reveal the possible options for eliminating the obvious risk (prenatal diagnosis, alternative reproductive procedures);
- Appropriately adapted to the new situation and the possible implications.

### **Components of the genetic counselling**

#### **Collecting Information**

The interview begins at the moment of reference, and the atmosphere which is set can determine the success or failure

later in the further process. The demographic and basic historical information can be successfully obtained by means of well-trained administrative staff. It takes trust and communication to get to the point of access to genetic counselling. In the beginning it is needed to gain an understanding of the family of the patient and for the reasons to access and request testing or advice. Unless there is incontrovertible evidence of accurate diagnosis at the time of the referral, it is better to leave little hope that genetic evaluation can show that the patient has a genetic disorder. It is necessary to extensively describe what types of testing will require the consultation. Preparing the family for the fact that it is not always possible to determine the true aetiology and diagnosis will help to establish realistic expectations for the results. Adequate response at the time of the referral can facilitate triage and staffing of personnel in the specific case. If the family is in panic from the information received about the possibilities of diagnosis and potential consequences of urgent medical problems should be kept until the information is relevant and one hundred percent sure. The medical records of family that is affected to be completed in order to contribute to a productive and efficient meet with them. The meeting should be scheduled when the entire team led by geneticist physician will be completed. The interaction will allow activating the family as an active part of the counselling. It is good to give them the responsibility to collect information and other tasks that can increase the sense of control over the situation and its understanding. It makes understanding what information is relevant and which are not relevant to the case. Further medical aspects of diagnostic evaluation are performed by clinical approach. People are rarely familiar with the procedures involved in the process of testing and counselling. The nature of inherited diseases, even for people who attended the genetic counselling can make them to abstain to provide information, as well as the experience that has been gained. It is required of each individual people to take data on their view of the events and determine their contribution to the process.

### **Diagnosis**

The main goal of genetic counselling is to set specific and accurate diagnosis with information regarding the aetiology and recovery risk, manifestations, treatment and reproductive alternatives. Where appropriate, the geneticist manage to find grounds for disease that has existed for years, and was unknown to the family, or perhaps for the miscarriage was not revealed but the real reason was charged incorrectly exogenous cause (such as prenatal infection or trauma at birth). Starting the diagnostic process is with medical history. This is followed by clinical examination, laboratory analysis, ultrasound diagnostics, various imaging, and cytology, cytogenetic and molecular diagnostics that such procedures lead towards a final diagnosis. The identifying of the genetic disease when there is some preliminary diagnosis has a more specific course unlike any other disease because it is inherent. The process is personal and cannot be depersonalised. Although symptoms can be treated successfully, but genetic changes will not disappear, which means that the condition cannot be treated like any other disease. The accurate diagnosis is the first and main parameter as a starting point in genetic counselling. It must be taken a detailed history of the family and making

heredograma. If the patient is a child it is necessary to obtain data on the pregnancy of the mother, and over possible problems in pregnancy and childbirth. Medical findings, documents, reports of autopsies of infected relatives are helpful for diagnosis. In many cases it will be useful clinical, cytogenetic and specialist to inspected parents, brothers, sisters of the probates because of the possibility of having latent forms of the disease and undercover symptoms.

### Communications

Even when the diagnosis is clear and certain, problems can occur if the disorder is causally heterogeneous, for example deafness and mental retardation, which causes can be, combined genetic and environmental factors. Genetically heterogeneous disorder may be caused by multiple genetic mechanisms (autosomal dominant, autosomal recessive and J.-linked), which makes genetic counselling very difficult. Therefore, this process is complex and depends on the skill of one who implements, communication skills and expertise. Person giving advice is called genetic counsellor or genetic adviser. For the informative interview the genetic counsellor should be prepared to discuss not only the findings that are specific but also for the financial, social and for the part to remove the malformation (if any) and other issues that may be important for decision-making. In fact through communication he helps to quantify the risk and in determine the readiness for acceptance or rejection (Zergollern, 1994).

### Setting of the options

When the diagnosis is clear such as chromosomal abnormality, then for the characteristics of the condition should be informed the family. Some parents cannot accept the existence of hereditary diseases and need accurate diagnoses for their existence and occurrence. The choice of future trials belongs to the parents who should make the final decision while the role of the genetic counsellor is non-directive. Geneticist should avoid directivity, its role should be fair and neutral, leaving space for the decision of the parents to be establish and unaffected by side. In helping to set preferences genetic counsellor talk to people about their actions and possible alternatives. For example, despite the phenotypic characteristics of the fetus in womb for a couple are important and other information such as:

- Possibilities for fetus to die or to be born prematurely;
- Whether the fetal diagnosis increases the risk for health of the mother?
- What could be the child's conditions after the birth?
- What decisions should be made about the treatment of child after the birth?
- What assistance can be expected at growing child?
- What options have if they decide that they cannot care for the child?
- If they decide to terminate the pregnancy they will need information on:
- What alternatives they have, i.e. for the type of abortion?
- What are the strengths and weaknesses of each method?

- What is the relatively economic compensation for each method?
- What are the procedures and the actions to be explained?

Although genetic counselling for specific problems is of limited importance, however many authors hope that it will in future play one of the stoutest roles not only in medicine but also in the overall destiny of mankind. Knowing of the structure of DNA, the main carrier of hereditary traits to certain principle that in the future could consciously be changed some primitive organisms (for example pneumococcal without capsule and pneumococcal with capsules are produced by means of suitable extract of DNA).

### Indications for genetic counselling

Although there are indications on a large scale for various phenomena and events, just like basis and list some of them (as an example) are listed in the following conditions compulsorily implies the process of genetic counselling:

- Concerns for possible reappearing of a family disease;
- Noticeable defects in the newborn;
- Mental Retardation or delayed development;
- Chronic, infantile neurological and neuromuscular diseases;
- Short stature in child or other forms of retardation of the growth;
- Metabolic diseases;
- Dysmorphia on the face, head and body;
- Non distinctive genitals or abnormal sexual development;
- Status of the carrier of certain genetic diseases which are characteristic of certain population groups: sickle cell anaemia, thalassemia, cystic fibrosis, Tay-Sachs disease, etc;
- Infertility, Sterility, miscarriages;
- Exposure on potent mutagenic or teratogenic agents;
- Pregnancy on the age of 35 or later;
- Consanguinity;
- Expression of the capacity in adolescent-adult life;
- Behavioural disorders;
- Cancer disease, heart disease, hypertension, and other common diseases with known hereditary basis.

The outcome of the genetic counselling depends on a number of factors, from family, social, educational, religious, cultural, influence of the environment, etc. Depending on the impact of these factors the decision that the couple will make as a result of genetic counselling will be shaped or the assistance which they could asked is linked with certain specific circumstances.

### Activities of the Medical Genetic Counselling in Macedonia

The genetic counselling can be reviewed over and over and to be presented by many aspects to understand the different medical and ethical issues. The dilemmas which are raised by genetic counselling is necessary for practical reasons to be highlight on certain functions that are performed within the

medical genetic counselling. The experience in Macedonia, especially in the Medical Genetic Counselling with Cytogenetic laboratory which for many years worked within the Institute of Mental Health of Children and Youth "Youth" in Skopje, as well as other institutions in the country provide the basis for the modernization and improvement of the activity of these counselling offices. On the prevention of the acquired diseases in the past have been paying more attention. The well known eugenic counselling, which could provide information on specific hereditary diseases later are called genetic counselling centres. Their work, among other things is based on the principle of informing patients about weight, importance and prognosis of certain hereditary disease. This activity, which is the ultimate achievement in medical genetics (Zergollern, 1994) can be called genetic information, which is very important for the proper treatment of each patient. The exact etiological diagnosis is an important precondition for providing genetic information for the risk assessment, possible treatments, and for prenatal diagnosis or prevention of congenital malformations. The genetic consultative activity as a daily practice and working activity in the Medical Genetic Counselling with Cytogenetic laboratory in Macedonia. The following methodologies are implemented:

- Determining the risk of hereditary anomalies.
- Explanation and evaluation of certain risk in particular family situation.
- Provision of adequate assistance regarding the proper and practical risk assessment and preparation of a rational plan for further action and decision.
- Continuous monitoring of the conditions and evaluation of the achieved results.

The early application of these methodologies is consists in setting an exact diagnosis of genetic disease while continuously thinking about possible occurrences of phenocopies and genocopies, the possible emergence of new mutations and variability in expression and penetration of the mutant gene. Also a precise and detailed family history and making a family tree (hierogram) is necessary. In addition a complete clinical examination of the child is conducting and laboratory tests (for chromosomal and biochemical substrate), prenatal and postnatal, then electroencephalography, computerized tomography and also analysis of the dermatoglyphic modalities. For the detection of the risk of recurrence of the disease is very important to make a diagnosis, to take history from the person seeking advice because then and only when sufficient heredogram is ready could set proper diagnosis and possibilities of its recurrence. Also additional laboratory and clinical trials are needed, which confirms that teamwork of specialist is very important. Within the one counselling office worked: doctor paediatrician, geneticist; biochemist; gynaecologist population geneticist; endocrinologist, general practitioner, ophthalmologist, human embryologist, lawyer haematologist, immunologist, sociologist, educator, etc. For the genetic information the obtaining of the diagnosis is an essential postulate. Because of the given limited laboratory facilities, especially in the diagnosis of genetic diseases, sometime it is necessary to use empirical findings that give only probable, but not certain, correct diagnosis (risk is probable that needs to indicate it to

the person who receives information). In the Medical Genetic Counselling with Cytogenetic laboratory in Macedonia the patients who were carriers of the disease, or are in the states of altered phenotypic characteristics are classified (due to practical reasons) in the following categories:

- monosymptom anomalies (Hydrocephaly, Microcephaly, Polidactylia etc.);
- polysymptom anomalies, without syndrome identifications (Syndactyly+mental retardation, Arachondactyly + mental retardation, etc.);
- confirmed syndromes which are with discrete symptoms (Syndrome Sturge-Weber, Apert syndrome, etc.)
- identified syndromes with clear phenotypic expression (tuberosc Sclerosis, Neurofibromatosis, Fenylcetonuria, Hunter syndrome, etc.);
- verified syndromes with chromosomal aberrations: Down Syndrome, Syndrome Turner, Klinefelter Syndrome etc;
- changes in the human genome because of the mutagenic effect, caused by various factors and agents (physical, chemical mutagens, viruses, ageing, some inherited genetic diseases, etc.).

The respondents in which the chromosomal aberrations and gene mutations were found usually have: trisomy of specified autosomal chromosome, tetraploid, mixploid, termination of chromosome, partial monosomy, ring chromosomes, duplications, isochromosomes, bicentric chromosomes, inversions, translocations, insertions, numerical aberrations gonosomes etc. The largest number of respondents with chromosomal aberrations is autosomal trisomy, i.e. with Down syndrome which is most pronounced where the phenotypic effect of mental retardation and congenital abnormalities of the heart are expressed. Most cases of Down syndrome (93% in our specimens) have a simple Trisomy of chromosome 21. In 1-2% of patients occurs mosaicism, and in about 5% of cases trisomy 21. This trisomy 21 occurs because when a parent is a carrier of balanced translocation (chromosome 21 translocation or arising de novo in the meiosis of one parent). The Trisomy 18, also known as Edwards's syndrome, is a condition which is caused by an error in cell division, known as meiotic disjunction. When this happens, instead of the normal pair, an extra chromosome 18 results (a triple) in the developing baby and disrupts the normal pattern of development in significant ways that can be life-threatening, even before birth. Besides these kinds of investigations the dermatoglyphics screenings are conducting (which in clinical researches mainly is used as auxiliary diagnostic method in conjunction with other clinical methods and techniques, because changes vary more or less). In our patients the diagnosed find out changes in 112 of them (only 8 entities changes are present 100%, and it is known that changes in dermatoglyphics present even in 143 types of diseases). The aetiology of congenital anomalies should be noted, when it is analysed as an important aspect in the medical practice. It is a very complex and heterogeneous, and sometimes it is very difficult to set the correct etiological diagnosis, especially those malformations and diseases in which there is a marked genetic heterogeneity. The exact etiological diagnosis is an important precondition for giving

proper genetic information, risk assessment, eventual treatment, as well as for prenatal diagnosis or prevention of congenital malformations. It has to consider the fact that congenital diseases are becoming more common cause of morbidity and mortality in childhood. Many studies show that morbidity in 8.5% of the investigated cases is due to genetic defects in 2.5% due to chromosomal abnormalities, 31% due to complex genetic reasons (multi factor analysis). Mortality in paediatric wards, at 40-50% can be explained to be a consequence of genetic diseases, including flattening and most common are congenital anomalies.

Based on these principles can be noted that social responsibility for the reproduction of the population in the context of avoiding hereditary abnormalities in early prenatal diagnostics his stronghold can only be based on genetic counselling. It is feasible both in Medical Genetic Counselling with Cytogenetic laboratory and in institutions which have cytogenetic laboratories and are equipped with experts in this field. It is known that in the country such opportunities exist in the Academy, Clinic of Paediatrics and other institutions. Such engagement allows some degree of prevention of the occurrence of handicapped persons that increases the concern of parents for healthy children. To achieve this biological function, society must ensure all requirements for the functioning of medical genetic counselling that will indicate the possibilities for matching defects and genetic diseases, as well as the way of avoiding them. With this activity of genetic counselling offices within the social framework can give impetus in the creation of a healthy and useful workable population and in terms of prevention of major expenses for assistance, education and training of mentally retarded persons. But it is not only the material effects that are measurable, but possibilities to avoid trauma and psychological consequences that are living with the curse of mental illness in the families facing them and who are struggling to overcome.

## Prevention

Some of these forms of prevention exist in other countries, and also in the Republic of Macedonia, which expresses the relationship to the protection of healthy children which is a priority in every society. The system of prevention and care in the country is regulated by laws and other acts which in part regulate the issue, especially in environments where there are health, social and other services and institutions that monitor these phenomena. Starting from the possibilities of prenatal monitoring, childbirth in hospital with contemporary conditions, vaccination, visit the home of the newborn revaccinations and similar forms of medical care are part of the endeavours at an early stage to be detected forms of mental subnormality that are possible in all social groups and to be given them proper treatment and care. But as with any system there are various limitations and obstacles that raises the question of building a more effective system of prevention, early detection, care and treatment of persons with mental subnormality. Constantly worldwide are developed new recommendations for the treatment and prevention of mental retardation and developmental disorders. Seeks to improve the quality of services for the mentally subnormality and to find ways to better meet their specific needs. From insulating

access to these persons, on the beginning of the 20th century, the attitude and understanding for their involvement and for normalization within society and institutions are changed. Most developed countries adopt this approach (especially Canada, UK, USA and Scandinavian countries)(6,7,8,9). For this purpose are driven insights to improve the prevention of mental subnormality, deinstitutionalization in treatment of persons, education and employment of persons with mental disabilities, taking legal and other measures to improve their standing in society. Based on observations made in the aforementioned literature and on practical knowledge and experience for the emergence of mental subnormality can give a very general assessment, which is:

- Prevention is possible and it can be conducted if there is timely information about the factors that affect this phenomenon.
- If there is widespread knowledge among the population about the effect of certain factors on the expression of the emergence of mental subnormality, which means health education and training within the system of the country.
- If there is a well developed network of health, social and other institutions within its jurisdiction to initiative and propose measures for more effective preventive action and prevention in terms of mental subnormality.

Prenatal observation is early prevention, but also all subsequent stages of development of a child is very important to detect diseases that can damage the central nervous system, and thus to cause mental subnormality. According to the degree of mental subnormality depends on the type of prevention strategy and approach. Because of the heterogeneity of the situation of these people can be brought to the principles of prevention and mental subnormality as:

- primary prevention, which includes abstinence from alcohol, smoking of drugs and other substances during pregnancy; universal immunization (rubella, hepatitis, measles, etc.); then improve the nutritional status in the middle; universal salt iodization to eliminate iodine deficiency as a risk factor for mental retardation; avoiding exposure to harmful chemicals and substances; detection and care for high-risk pregnancies and protection from possible infections; health education etc;
- measures for early intervention (Rowitz (4), in order to minimize the functional disability, and to maximize the capabilities of the individual through education, raising the level of skills etc;
- specific treatment, if known the cause of the problems in order to minimize the possibility of stroke;
- wider medical attention to treat and prevent general medical conditions which are created as a result of complications of mental subnormality;
- treatment and prevention to be used for the psychosocial dysfunctions (9,10) in order to react positively on the person.
- Despite these measures the needs for prenatal diagnosis and screening are perceived in order to eliminate

abnormal conditions; neonatal screening; neonatal intensive care; genetic counselling.

According to the recommendations of the American Academy of Pediatrics it is recommended to perform routine inspections on 1, 2, 4, 6, 9, 12 and 18 months and 2 years in order to consider any adverse change that would lead to mental subnormality. Children should have an annual review between 2 and 6 years. Despite the general immunization a number of other measures aimed at preventive action within the family and society are proposed. The procedures assume that a careful anamnesis should be done, by examining the family conditions and diseases of genetic basis as a cause of mental subnormality which is also crucial in the early detection of any possibility of occurrence of these conditions. Therefore genetic consultation gets crucial importance together with cytogenetic studies of hereditary and other diseases. Given that genetic factors show a great diversity in the way of emergence, in severity and timing of expression and duration, it is necessary to conduct extensive prevention of diseases caused by damaged genes and chromosomes. Their prophylaxis for families at risk can be significant, using various forms and methods of such prophylaxis.

In this connection it is necessary in the country to form and organize a network and structure of genetic laboratory facilities and genetic consultative service that will be interconnected organizational, structural and information related to the order to be established knowledge of these phenomena at the level the entire country. In this respect it is very important genetic counselling in the state to be designed and equipped to be able to exercise the function in preventing the occurrence of mental subnormality. With the disclosure of the impact of different etiologic factors as causes of mental subnormality and get to know the trends of these events and opportunities for the organization and efficient operation in the prevention of mental health. The data that exist and are upgrading constantly, represent a solid stock of knowledge in this area of which the starting point in the development of rational health care and efficient health care system in Macedonia. Besides the genetic consultation should be applied and amniocentesis, which will provide data on enzyme activity, a finding of chromosomal aberration, determination of sex, and thus the possibility of perceiving the related sexually transmitted diseases and based on the findings to decide possible interventions as early as the prenatal stage. In terms of having a family syndrome (Down, Trisomy 21, etc.) should take special care in all forms and methods that track the occurrence, depending on the type of the syndrome. Besides prenatal care for mothers is particularly important in the function of protecting the fetus that assumes avoiding radiation, consumption of alcohol and drugs, viral diseases etc. Especially important is timely vaccination against rubella to prevent congenital anomalies on the fetus. The perinatal care is consists in avoiding the consequences of birth trauma and prompt treatment of hyperbilirubinemia and kernicterus, and other actions that may be forced at this stage depending on etiological factors that may be expressed. The postnatal prevention includes: early detection of genetic metabolic encephalopathy, investigation of tyrosinemia, adequate and timely immunization therapy for preventing the sequelae that can lead to mental subnormality.

Given that the genetic diseases show a great diversity as a way of inheritance and in severity, time of occurrence and duration, it is necessary to conduct extensive prevention of diseases caused by damaged genes and chromosomes. The benefits to the community of each promptly warned case is constant. The mode of production and dissemination of the etiological factors for mental subnormality, and their impact on the incidence of mental subnormality is very important for prospective parents; they to be informed about the possible consequences of the action of these factors. It is possible if the forms and methods of health education are improved on the importance of heredity, the consequences of infectious diseases, various toxins, radiation and other external influences that can lead to prenatal or postnatal negative impact on children. In addition it is necessary to introduce a complete system of health records, monitoring and care, with modern technological information and opportunities with trained staff that will promptly detect and treat people with disabilities (11). This will enable the diagnostic process continues in continuity with the rehabilitation process and permanent monitoring of persons with disabilities.

## Conclusion

In conclusion: A healthy child is the goal of every parent. Therefore it is important for prospective parents on time and on the right way to learn and to plan their offspring. It can point out that genetic diseases show a great diversity as a way of inheritance and in severity, time of occurrence and duration. Although certain hereditary diseases are rare, their prophylaxis for families at increased risk may be both significant and prophylaxis of severe infectious diseases. It is therefore necessary monitoring, recording and prevention of these diseases in the population. By introducing a single register of hereditary diseases in the form of data bank as individual chromosomal structures and for family genetic conditions, it would enable the social framework to take immediate measures to influence in all areas, both through education of the population for the effects of these diseases, and in terms of possibilities for their prevention. It will improve the efficiency of services for genetic counselling, and patients' trust in these institutions

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