

MEDICAL AND GENETIC ASPECTS OF PLANNING PREGNANCY AMONG MARRIED COUPLES

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Annotation. Relevance. The decision to start a family is a significant milestone for many married couples. However, with advancements in medical genetics, there's a growing awareness of the importance of considering genetic factors when planning pregnancy. Understanding medical and genetic aspects can help couples make informed decisions to minimize the risk of hereditary diseases and optimize the health of future generations. **Purpose of the study.** The aim of this article is to explore the medical and genetic aspects that married couples should consider when planning pregnancy. It seeks to provide comprehensive information on genetic counseling, carrier screening, and preconception genetic testing to empower couples to make informed choices about their reproductive health. **Materials and methods:** an extensive review of current literature on medical and genetic aspects in pregnancy planning, including studies on genetic counseling, carrier screening programs, and preconception genetic testing; interviews of medical geneticists, obstetricians, genetic counselors, and reproductive specialists to gather insights into the latest developments and best practices in medical and genetics for pregnancy planning; relevant clinical guidelines and recommendations from professional organizations to provide evidence-based recommendations for couples planning pregnancy. **Results end conclusion.** Understanding the medical and genetic aspects of pregnancy planning is critical for couples to make informed decisions that promote healthy outcomes for both parents and their unborn children.

Key words: inheritance, genetic counseling, family history, genetic risks, prenatal testing, PGT, X-linked, multifactorial.

For citation:

Tuychibaeva N. M., Tursunbaeva D. B. Medical and genetic aspects of planning pregnancy among married couples. *Advanced Ophthalmology*. 2023;6(6):54-57

TURMUSH QURGAN JUFTLIKLAR O'RTASIDA HOMILADORLIKNI REJALASHTIRISHDAGI TIBBIY VA GENETIK JIHATLAR

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Annotatsiya. Dolzarbligi. Farzandli bo'lish qarori ko'plab turmush qurgan juftliklar uchun muhim bosqichdir. Hozirda tibbiy genetik sohasidagi yutuqlar bilan homiladorlikni rejalashtirishda irsiy omillarni hisobga olish muhimligi haqidagi xabarlar ortib bormoqda. Tibbiy va genetik jihatlarni tushunish juftliklarga irsiy kasalliklar xavfini kamaytirish va kelajak avlodlar salomatligini optimallashtirish uchun ongli qarorlar qabul qilishga yordam bermoqda. **Tadqiqot maqsadi.** Ushbu maqolaning maqsadi turmush qurgan juftliklar homiladorlikni rejalashtirishda e'tiborga olishlari kerak bo'lgan tibbiy va genetik jihatlarni o'rganish, shuningdek ularga o'zlarining reproduktiv salomatligi bo'yicha ongli tanlov qilishlariga imkon berish maqsadida genetik maslahat, tashuvchi skriningi va homiladorlikdan oldingi genetik testlar haqida to'liq ma'lumot berishdir. **Materiallar va usullar:** homiladorlikni rejalashtirishda tibbiy va genetik jihatlarni bo'yicha joriy adabiyotlarni keng ko'lami tahlil qilish, shu jumladan, genetik maslahatlar, tashuvchilar skrining dasturlari va homiladorlikdan oldingi genetik testlar bo'yicha tadqiqotlar; tibbiy genetiklar, genetik maslahatchilar va reproduktiv mutaxassislar homiladorlikni rejalashtirish uchun tibbiyot genetikasining so'nggi ishlanmalari va ilg'or tajribalari to'g'risidagi ma'lumotlar; homiladorlikni rejalashtirayotgan juftliklar uchun dalillarga asoslangan tavsiyalar berish uchun tegishli klinik ko'rsatmalar va professional tashkilotlarning tavsiyalari. **Natijalar va xulosa.** Homiladorlikni rejalashtirishda tibbiy va genetik jihatlarni tushunish, turmush qurgan juftliklar va ularning tug'ilgan bolalari uchun sog'lom natijalarga yordam beradigan ongli qarorlar qabul qilishlari uchun juda muhimdir.

Kalit so'zlar: irsiyat, genetik maslahat, oila tarixi, genetik xavflar, prenatal test, PGT, X-bog'langan, multifaktorial.

Iqtibos uchun:

Tuychibayeva N. M., Tursunbayeva D. B. Turmush qurgan juftliklar o'rtasida homiladorlikni rejalashtirishdagi tibbiy va genetik jihatlarni. *Ilg'or oftalmologiya*. 2023;6(6):54-57

МЕДИЦИНСКИЕ И ГЕНЕТИЧЕСКИЕ АСПЕКТЫ ПРИ ПЛАНИРОВАНИИ БЕРЕМЕННОСТИ У СУПРУЖЕСКИХ ПАР

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Аннотация. Актуальность. Решение о создании семьи является важной вехой для многих семейных пар. Однако с развитием медицинской генетики растет понимание важности учета генетических факторов при планировании беременности. Понимание медицинских и генетических аспектов может помочь парам принять обоснованные решения, чтобы минимизировать риск наследственных заболеваний и оптимизировать здоровье будущих поколений. **Цель исследования.** Целью данной статьи является изучение медицинских и генетических аспектов, которые супружеским парам следует учитывать при планировании беременности. Целью проекта является предоставление исчерпывающей информации о генетическом консультировании, скрининге носителей и генетическом тестировании до зачатия, чтобы дать парам возможность сделать осознанный выбор в отношении своего репродуктивного здоровья. **Материалы и методы:** обширный обзор современной литературы по медико-генетическим аспектам планирования беременности, включая исследование по генетическому консультированию, программам скрининга носителей и генетическому тестированию до зачатия; интервью с медицинскими генетиками, акушерами, генетическими консультантами и специалистами по репродукции для сбора информации о последних разработках и передовом опыте медико-генетики при планировании беременности; соответствующие клинические руководства и рекомендации профессиональных организаций для предоставления научно обоснованных рекомендаций парам, планирующим беременность. **Результаты и заключение.** Понимание медицинских и генетических аспектов планирования беременности имеет решающее значение для супружеских пар, позволяющих принимать обоснованные решения, способствующие здоровому исходу как для родителей, так и для их будущих детей.

Ключевые слова: наследование, генетическое консультирование, семейный анамнез, генетические риски, пренатальное тестирование, ПГТ, X-сцепленный, многофакторный.

Для цитирования:

Туйчибаева Н. М., Турсунбаева Д. Б. Медицинские и генетические аспекты при планировании беременности у супружеских пар. Передовая офтальмология. 2023;6(6):54-57

Planning for a pregnancy is a pivotal moment in the lives of married couples, marked by excitement, anticipation, and careful consideration. In addition to factors such as financial stability and emotional readiness, medical and genetic aspects play a crucial role in ensuring a healthy pregnancy and the well-being of the future child. Understanding the genetic factors that can impact pregnancy outcomes is essential for couples embarking on this journey.

Genetic counseling plays a pivotal role in assisting married couples in navigating the complexities of medical and genetic aspects when planning pregnancy. It involves a collaborative process between trained genetic counselors and couples to assess their genetic risks, understand inheritance patterns, and make informed decisions regarding reproductive options.

Genetic counselors conduct a comprehensive evaluation of the couple's medical history, family history, and ethnic background to identify potential genetic risks and hereditary conditions. This assessment helps tailor counseling sessions to address specific concerns and provide personalized recommendations. Genetic counselors educate couples about the genetic basis of inherited disorders, including autosomal recessive, autosomal dominant, X-linked, and multifactorial conditions. They assess the likelihood of passing on genetic conditions based on family history, carrier status, and ethnic predispositions. Couples are

empowered with knowledge about the inheritance patterns, recurrence risks, and available testing options.

Couples may undergo carrier screening to determine if they carry gene mutations associated with hereditary conditions, such as cystic fibrosis, sickle cell disease, or Tay-Sachs disease. Genetic counselors interpret screening results, discuss implications for future pregnancies, and offer support in decision-making regarding reproductive options, such as prenatal testing or preimplantation genetic diagnosis.

In certain cases, genetic counselors may recommend preconception genetic testing to identify individuals at risk of passing on genetic disorders. This proactive approach allows couples to make informed decisions before conception, such as pursuing assisted reproductive technologies or exploring alternative family planning options.

Genetic counselors discuss prenatal testing options, such as chorionic villus sampling and amniocentesis, for detecting chromosomal abnormalities or genetic disorders during pregnancy. They provide information about the benefits, limitations, and potential risks associated with each testing method, empowering couples to make choices aligned with their preferences and values.

Genetic counseling addresses not only the medical aspects but also the emotional and psychosocial implications of genetic risk assessment

and reproductive decision-making. Counselors offer emotional support, facilitate open communication between partners, and provide resources for coping with uncertainty, grief, or anxiety related to genetic concerns.

Genetic counseling is an ongoing process that may involve follow-up sessions to address evolving concerns or changes in circumstances. Genetic counselors collaborate with healthcare providers, including obstetricians, maternal-fetal medicine specialists, and fertility specialists, to ensure coordinated care and appropriate referrals for additional medical management or support services.

Genetic counseling serves as a cornerstone in empowering married couples to make informed decisions about pregnancy planning by providing education, risk assessment, and psychosocial support tailored to their unique genetic backgrounds and reproductive goals. By integrating genetic counseling into the preconception care continuum, couples can optimize their reproductive health outcomes and mitigate the risk of hereditary diseases in future generations.

Inherited conditions represent a significant aspect of medical and genetic considerations in planning pregnancy among married couples. These conditions encompass a wide spectrum of genetic disorders that can be passed down from parents to their offspring. Understanding the inheritance patterns, genetic risk factors, and potential implications of these conditions is crucial for informed family planning decisions.

Inherited conditions may arise from single gene mutations, chromosomal abnormalities, or complex interactions between multiple genetic and environmental factors. Examples of inherited conditions include autosomal recessive disorders like cystic fibrosis, sickle cell disease, and Tay-Sachs disease, which require both parents to carry a mutated gene for their child to inherit the condition. Autosomal dominant disorders, such as Huntington's disease and Marfan syndrome, only require one parent to pass on the mutated gene for the offspring to be affected.

Furthermore, X-linked disorders, such as hemophilia and Duchenne muscular dystrophy, are caused by mutations on the X chromosome and can exhibit different inheritance patterns depending on the sex of the parent and offspring. Understanding the mode of inheritance and the risk of transmission is essential for couples to assess their likelihood of having a child affected by an inherited condition.

Genetic counseling plays a crucial role in evaluating the risk of inherited conditions and guiding couples through the decision-making process. Through comprehensive genetic testing and counseling sessions, healthcare providers can assess the couple's family medical history, perform carrier screening, and discuss available reproductive options tailored to their specific circumstances.

In cases where one or both partners carry genetic mutations associated with inherited conditions, couples may explore various reproductive options, including adoption, gamete donation, or assisted reproductive

technologies (ART) coupled with preimplantation genetic testing (PGT). These approaches aim to minimize the risk of passing on genetic disorders to offspring while fulfilling the couple's desire to have children.

Overall, addressing inherited conditions in the context of family planning requires a multidisciplinary approach that integrates medical expertise, genetic counseling, and ethical considerations. By empowering couples with knowledge about their genetic risk factors and available reproductive options, healthcare providers can support them in making informed decisions that align with their values, preferences, and long-term healthcare goals.

Genetic testing also plays a crucial role in the comprehensive approach to family planning among married couples. By leveraging advancements in genomic medicine, individuals can assess their risk of hereditary conditions and make informed decisions regarding their reproductive health. This proactive approach empowers couples to mitigate potential genetic risks and optimize the health outcomes of their future offspring.

Genetic testing encompasses various modalities, including carrier screening, preconception testing, prenatal diagnosis, and preimplantation genetic testing. Carrier screening allows individuals to identify whether they carry genetic mutations associated with inherited disorders, providing insight into the likelihood of passing these conditions to their children. Preconception testing enables couples to assess their genetic compatibility and potential risk of transmitting genetic diseases before conception occurs.

During pregnancy, prenatal diagnosis techniques such as amniocentesis and chorionic villus sampling offer the opportunity to detect chromosomal abnormalities and genetic disorders in the developing fetus. This information allows parents to make informed decisions about pregnancy continuation, medical interventions, and preparations for the child's care needs.

Moreover, preimplantation genetic testing (PGT) offers a proactive approach for couples undergoing in vitro fertilization (IVF). PGT involves screening embryos for genetic abnormalities before implantation, thereby reducing the risk of transmitting inherited conditions to offspring and increasing the likelihood of a successful pregnancy.

Genetic testing not only informs reproductive decision-making but also facilitates personalized counseling and medical management tailored to each couple's genetic profile. By integrating genetic information into family planning discussions, healthcare providers can support couples in making informed choices that align with their values, preferences, and healthcare goals.

Overall, genetic testing is a foundation in the medical and genetic framework for planning pregnancy among married couples, promoting reproductive autonomy, informed decision-making, and the prevention of hereditary diseases across generations.

While genetic factors play a significant role in

pregnancy outcomes, lifestyle factors also have a substantial impact on reproductive health. Prioritize a balanced diet rich in fruits, vegetables, lean proteins, and whole grains to ensure adequate nutrients for both partners. Regular exercise not only improves physical health but also enhances fertility and reduces stress. Avoiding tobacco, excessive alcohol, and illicit drugs is crucial to optimize reproductive health. Adequate sleep and stress management techniques contribute to overall well-being and fertility. Consulting healthcare professionals for personalized advice and genetic counseling can further optimize pregnancy planning.

In addition to dietary and exercise habits, maintaining a healthy weight is essential for fertility optimization. Both underweight and overweight conditions can negatively impact fertility, so striving for a body mass index within the healthy range is recommended. Managing chronic health conditions such as diabetes, hypertension, and thyroid disorders is crucial, as uncontrolled conditions can affect fertility and pregnancy outcomes. Prioritizing regular medical check-ups and screenings can help identify and address any potential health concerns early on. Finally, fostering open communication and mutual support between partners is key to navigating the complexities of pregnancy planning and medical and genetic considerations.

In conclusion, the integration of medical and genetic considerations into the planning of pregnancy among married couples is paramount in ensuring the health and well-being of both parents and offspring. By understanding the genetic risk factors and potential hereditary conditions, individuals can make informed decisions, seek appropriate medical guidance, and undertake necessary precautions to mitigate risks. Embracing advancements in medical genetics allows for personalized reproductive counseling, fostering empowered decision-making and the promotion of healthy pregnancies. As we navigate the complexities of family planning, it is imperative to recognize the

invaluable role of medical and genetic insights in safeguarding the future generations' health and vitality.

In addition to safeguarding the health of future generations, integrating medical and genetic aspects into pregnancy planning offers profound societal benefits. By identifying genetic predispositions to inheritable diseases and disorders, couples can take proactive measures to reduce the transmission of such conditions, ultimately alleviating the burden on healthcare systems and enhancing the quality of life for affected individuals and their families.

Furthermore, the consideration of medical and genetic factors in pregnancy planning facilitates the advancement of personalized medicine. Tailoring reproductive guidance based on individuals' genetic profiles enables healthcare professionals to deliver more precise and effective interventions, optimizing maternal and fetal outcomes. This personalized approach not only enhances reproductive health but also contributes to the broader evolution of healthcare strategies, emphasizing prevention and early intervention.

Moreover, by fostering open dialogue and education surrounding medical and genetic aspects, we empower couples to make informed choices that align with their values, preferences, and cultural beliefs. Through accessible and comprehensive genetic counseling services, individuals can navigate the complexities of genetic inheritance with confidence, promoting autonomy and autonomy in reproductive decision-making.

In essence, the integration of medical and genetic considerations into pregnancy planning represents a pivotal advancement in reproductive healthcare. By harnessing the insights of medical genetics, we can cultivate a future where every pregnancy is approached with knowledge, compassion, and the utmost commitment to the well-being of both current and future generations.

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Нет финансовой заинтересованности.