CHILDREN'S TUMOR FOUNDATION
ENDING NF THROUGH RESEARCH

Family Planning and Reproductive Options In NF

This resource reviews family planning and reproductive options for individuals with a diagnosis of NF, which refers to neurofibromatosis type 1 (NF1) and all types of schwannomatosis (SWN), including *NF2*-related schwannomatosis (*NF2*-SWN), formerly known as neurofibromatosis type 2 (NF2). This resource is for informational purposes only and should not be considered medical advice. It is important to make individual reproductive decisions with the help of a genetic counselor or other knowledgeable healthcare provider.

DECIDING WHETHER OR NOT TO HAVE CHILDREN

Becoming a parent is a major life decision with many new responsibilities. There are often medical, financial, career, and lifestyle considerations. Personal experiences and perspectives influence our views of becoming a parent. Discussing feelings and concerns with a spouse or partner is important to make an informed decision together. The optimal time to start these discussions is before one becomes pregnant.

For many couples, having a genetic condition does not affect whether they choose to have children. However, for others, a diagnosis may have a role in their choices for the future. Having a medical condition like NF may contribute to the considerations of starting a family. Potential factors include concerns about having a healthy pregnancy, future health, how it impacts raising a family, and risks to a child inheriting the condition.

REPRODUCTIVE AND FAMILY OPTIONS

Couples have many choices when considering family planning and reproductive options.

NO CHILDREN

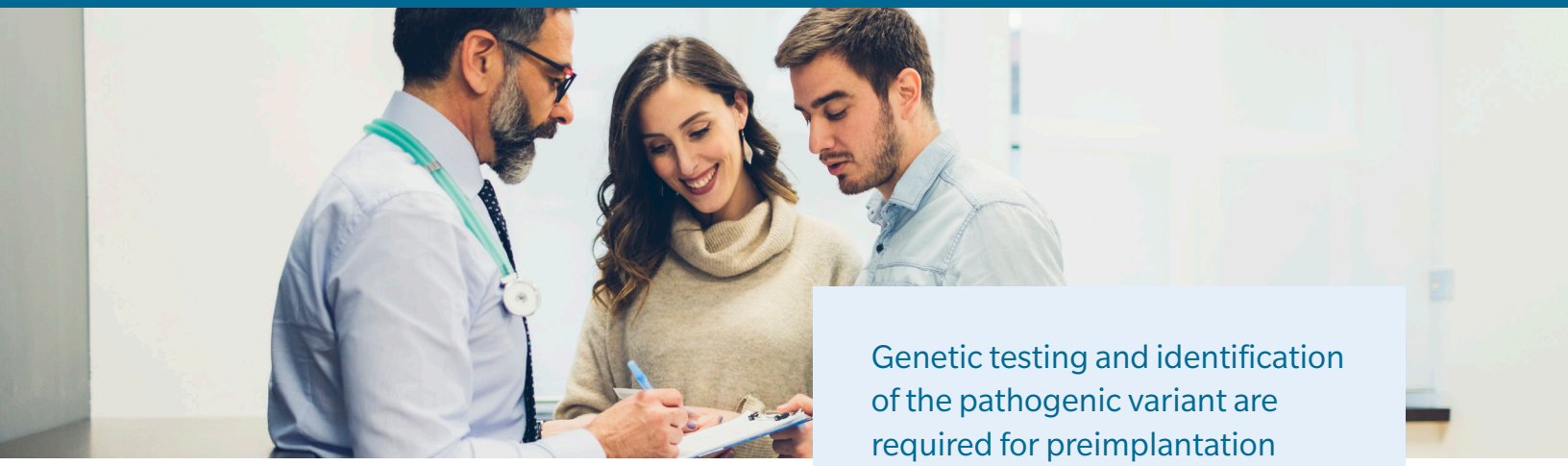
Some people may decide not to have children or to wait until a later stage in life. In these situations, abstinence (not having sexual intercourse) is the only way to prevent pregnancy with 100% certainty. A tubal ligation or vasectomy is also highly effective in preventing pregnancy. There are several methods of contraception available, including pills, implants, patches, and barrier methods. Although conclusive studies do not exist, when hormonal contraceptives are used, it is generally recommended that women with NF avoid high levels of progesterone.

ADOPTION

Some families choose not to conceive naturally and pursue adoption as an alternative. Adoption is a process whereby an individual or couple assumes the legal rights and responsibilities of parenting a child.

REPRODUCTION USING DONORS

When a couple is concerned about the potential risk of having a child with a genetic condition in the family, they may choose to use a donor reproductive cell (egg or sperm). Instead of using the affected parent's cell, a donor egg or sperm is used so the condition is not passed



> Genetic testing and identification of the pathogenic variant are required for preimplantation genetic testing and prenatal diagnosis. If genetic testing has been completed but has not revealed a confirmed genetic variant, these test options will not be useful.

to a child. This is done using in vitro fertilization (IVF) technology, which combines an egg and sperm to create an embryo in a laboratory. The embryo is then implanted into a woman for pregnancy. Another option is to use a donor embryo, which is not biologically related to either parent but can be implanted into a woman to carry the pregnancy to term.

PREIMPLANTATION GENETIC TESTING (TESTING PRIOR TO CONCEPTION)

Some couples consider testing prior to conceiving a pregnancy. Preimplantation genetic testing (PGT) involves the use of in vitro fertilization (IVF) combined with genetic testing. To pursue this option, genetic testing must have revealed a pathogenic variant in the individual in the family with NF. This procedure involves IVF to create embryos in a laboratory. Genetic testing is used to determine which embryos have the known variant and only embryos without the variant are implanted into the woman. This technology is only offered at specialized clinics throughout the country and may not be covered by health insurance plans.

PRENATAL DIAGNOSIS (TESTING PRIOR TO BIRTH)

After a couple becomes pregnant, they may choose to pursue testing to determine if the unborn baby has NF. In order to pursue this option, genetic testing must have revealed a pathogenic variant in the individual in the family with NF. There are two options for prenatal testing to allow for the diagnosis of NF during pregnancy.

Chorionic Villus Sampling (CVS) involves a biopsy of the placenta and typically is performed between 10-12 weeks of pregnancy.

Amniocentesis can be performed after 15 weeks of pregnancy by sampling the amniotic fluid surrounding the fetus.

Each procedure is performed using ultrasound guidance by an experienced obstetrician. The sample is sent to the laboratory for genetic analysis to determine whether the fetus inherited the known gene variant. As with any type of sampling procedure, these options each carry a risk for complications such as miscarriage. Both tests are very accurate, but specific risks and limitations should be discussed with a healthcare provider prior to any procedure.

Both of these procedures have been performed for various reasons in different settings for years and are available by many practitioners throughout the country. Couples may find prenatal testing helpful for a variety of reasons, including:

- If a test indicates that the fetus inherited the condition, it allows for the preparation of a child with NF and potential changes in labor and delivery plans. It also provides the option to consider interventions such as pregnancy termination. Different state laws affect the available options.
- If a test indicates that the fetus does not have the condition, it offers reassurance that the child will not have NF.

It should be noted that routine testing done on all pregnant women, such as ultrasound or blood testing, does not detect a diagnosis of NF during pregnancy except in very rare circumstances.

TESTING AFTER BIRTH

Parents may choose to test the baby once he or she is born instead of pursuing prenatal testing. This allows for early diagnosis and monitoring if a diagnosis is not possible based on clinical features alone.

PREGNANCY AND NF

A diagnosis of any type of NF does not affect an individual's ability to conceive a child (become pregnant). Many women with these conditions have normal, healthy pregnancies. However, there are some potential health complications for a woman with NF that may arise. Therefore, a referral to a high-risk obstetrician or maternal-fetal medicine provider is often recommended.

Although many women with NF1 do not have medical issues during pregnancy, some report an increase in the number or size of neurofibromas while pregnant, which may not resolve after pregnancy. They may also experience elevated blood pressure or pre-eclampsia.

Pregnancy risks for *NF2*-SWN and other types of SWN are unknown, but new and significant complications are generally not expected.

As with any pregnancy, one should consider existing health conditions, current medications, and family history risks so that specific recommendations can be made.

GENETICS AND INHERITANCE

NF1 and all forms of SWN are autosomal dominant genetic conditions. Therefore, if a parent has one of these conditions, there is a 50%, or 1 in 2, chance that they will have a child who inherits the affected gene. Knowing someone has the affected gene does not predict what features will develop or how severe the condition might be, except in a few situations.

If a patient has one of these conditions, there is a 50% chance that they will have a child who inherits the affected gene.

It is essential to talk to a healthcare provider about specific medical issues and concerns and to have a pregnancy management plan in place.

ADDITIONAL CONSIDERATIONS

In considering the options for family planning, some additional considerations may impact what options are available. Some options are costly and may not be covered by medical insurance plans. In addition, some options require genetic testing to identify the specific pathogenic variant (mutation) in the family member with NF before being used for reproductive purposes. For details about genetic testing in NF, see the CTF resource, "A Guide to Genetic Testing in NF and SWN." Lastly, not all options are available to every couple. Talk with an experienced provider who can review the available options with you.

PERSONAL CONSIDERATIONS OF THE OPTIONS

There are many options available for family planning and no one option is right for every couple. These are very personal decisions and involve many feelings based on a family's beliefs, values, and faith. A couple needs to feel supported in whatever choices they make. Factors to consider include:

1. Wishes to have children
2. Importance of having a biologically related child
3. Concerns for one's own health or the health of a child
4. Religious and cultural beliefs
5. Risks, benefits, and limitations of each option
6. Insurance coverage and costs of available options

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The Children's Tumor Foundation does not advocate for specific family planning or reproductive actions. We are committed to sharing this information so families know the possible options and can make informed decisions. Families should speak with a genetic counselor or other knowledgeable healthcare provider when making reproductive decisions.

Please find additional information and resources at ctf.org/genetics

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