Reproductive Options for People with NF1, NF2 and Schwannomatosis

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People who themselves have been diagnosed with NF1, NF2 and schwannomatosis, or who have a child with one of these diagnoses, often wonder whether future children of theirs will be affected with the same condition. There can be deeply-held beliefs and sensitivities that are accessed in the process of first talking about a genetic risk with a partner, considering having children, passing on a genetic condition, and exploring options for preventing future children from being affected with a genetic condition. Genetic counselors are trained to work with individuals and families to provide accurate and detailed information about this risk, as well as to discuss options for family planning and to support people in whatever choices they make.

This article is written to provide basic information for people with NF1, NF2 and schwannomatosis about genetic testing during a pregnancy and options for having a baby other than getting pregnant naturally. It is not a full discussion about the issues related to these choices. Also, this article does not discuss the choice to proceed with a natural pregnancy and not do genetic testing, which is a choice that many individuals and couples make. The information provided here can familiarize you with choices prior to receiving personalized and much more detailed information through the process of individual genetic counseling.

Prenatal Diagnosis

After a couple gets pregnant, they can choose to do testing to learn if the unborn baby has the gene change for NF1, NF2 or schwannomatosis. Prior to doing this, the gene change in the family must be known meaning that the person who is affected must have genetic testing. It is easiest to coordinate genetic testing for the affected parent prior to becoming pregnant, but it can be done during the pregnancy as well. Most of the time, prenatal diagnosis is at least partially covered by insurance.

The earliest prenatal test available can be done around 10 weeks of pregnancy and is called chorionic villus sampling, or CVS. The next available test can be done around 16 weeks of pregnancy and is called amniocentesis, or amnio. The tests are done differently, though both are very accurate. It is important to know that both tests have a small chance of causing a miscarriage. Many families feel that they are willing to take this risk in order to get the information that is available from doing the test.

A newer method of learning whether an unborn baby has inherited a gene change for NF1, NF2, or schwannomatosis is called cell-free fetal DNA (cfDNA) testing. This test is considered "non-invasive" because it only requires a blood draw from the pregnant woman so involves no risk of miscarriage. DNA from the baby that has crossed through the placenta and into the pregnant woman's bloodstream can be used to predict whether the baby has likely inherited a DNA change in the family. cfDNA testing is usually completed after 10 weeks of pregnancy. It is almost as accurate as CVS or amnio testing mentioned above but not quite, so some families choose to follow up after cfDNA testing with one of these procedures.

Pregnancy Termination

There are a variety of reasons that a person or couple might choose not to continue a pregnancy and this choice is a deeply personal one. It is important to afford yourself and those involved in these choices a great deal of respect and understanding, as we know that there are complex feelings and beliefs that underlie these decisions. Pregnancy termination is available in most states through 24 weeks of pregnancy and is often covered by insurance in the setting of a genetic diagnosis. People who make use of support provided to them from many sources (*i.e.,*

friends, family, church, individual and group counseling) through this process tend to do better incorporating their decision in the long term.

Preimplantation Genetic Testing for Monogenic Disorders (PGT-M)

Some couples prefer to consider testing prior to conceiving a pregnancy. In order to do this, couples undergo *in vitro* fertilization (IVF) combined with genetic testing. Eggs from the mother and sperm from the father are harvested separately and then combined in a specialized lab to create many fertilized eggs at one time, called embryos. The embryos are then tested to determine which carry the gene change being inherited in the family. Prior to doing this, the gene change in the family must be known meaning that the person who is affected must have genetic testing. Only embryos without the gene change are implanted. This process does not guarantee that a pregnancy will occur. It can also be very expensive and is often not covered by insurance.

Adoption/Gamete Donation

There are several types of adoption/donation, including traditional adoption, embryo adoption, sperm donation and egg donation. Some options allow for the baby to be related to one of the parents and some allow for the woman to carry a pregnancy. Other considerations include the cost and time needed to pursue these methods. The prices for these options can vary widely, and consideration is typically given to what other medical or health concerns a child might have given the common lack of detailed family history that is provided through the adoption process.

Not Having Children

Many people in our society choose not to raise children. Often this decision is based on a number of considerations, and having a genetic condition is only one of them. Some people worry that their condition may get worse over time and make it difficult for them to care for their children. Other people worry that pregnancy itself may make their disease worse or make it more difficult for them to have children. Some people also worry about having a child with NF or schwannomatosis and do not want to consider the options mentioned above.

Below is a table that summarizes available options at this time. Prices are rough estimates to provide a sense of scale, but may not be accurate for each case depending on circumstances.

	Biologically related	Natural conception	Financial Cost	Genetic testing needed
Natural conception ("take our chances")	Yes	Yes	$0	No
Prenatal diagnosis (CVS, amnio)	Yes	Yes	$1000-$3000 (usually covered by insurance)	Yes
Preimplantation genetic diagnosis	Yes	No	$15,000-$20,000 (small portion covered sometimes)	Yes
Egg/sperm donation	Yes (to one parent)	No	Egg: $15,000-$20,000 Sperm: $200-$3000	No
Embryo adoption	No	No	$6000-$17,000	No
Adoption	No	No	$5000-$40,000	No

No children	**N/A**	**N/A**	**$0**	**No**

The choice to have a child after you or a family member has been diagnosed with NF or schwannomatosis can bring up many different feelings and thoughts, which is normal. It is important to discuss these with your NF specialists to give you a chance to talk more about your symptoms, what pregnancy might be like for you and what options are available if you are interested in learning more. It is best to work with providers who have a lot of experience with NF and schwannomatosis when you are making these choices. To find a genetic counselor near you, go to www.nsgc.org and use their search feature. You can also contact your local NF/Schwannomatosis provider and ask for a referral to a genetic counselor with experience working with people and families with these diagnoses.