What You Might Want to Know Before Embarking on Your Parenting Journey

Our heartiest wishes to you, mom to be! During this special phase of your life, you must be so excited and look forward to meeting the youngest member of your family. But at the same time, many of you might have concerns about what is happening with you and your baby.

Today, medical advances offer new options for obtaining information about baby's health even before the birth. While more information sounds helpful, the truth is that it often leads to an information overload and causes confusion among patients. There are many optional tests that aren't included in regular checkups, which makes it even harder for parents to decide on their own.

Our professional mission is to assist you in order to make the best-informed choice. This leaflet includes an overview of your prenatal options, as well as a few resource suggestions to better meet your needs. Feel free to contact any of them if needed.

Following is a handy list of questions and answers that will help you in your learning of the basic concept of your prenatal options.

Search for accessible institutions. All the centers listed are the members of The Japan National Liaison Council for Clinical Sections of Medical Genetics.
http://www.Idenshiiryoubumon.org/search/

Available only in Japanese

Access by QR code

All About Your Options for

Pregnancy & Prenatal Care

The Project of Health Research on Children, Youth, and Families by Ministry of Health, Labor and Welfare, Japan (2014-2016)

Q1. What are prenatal genetic tests?

A. They are medical tests used to obtain genetic information about unborn babies. Certain tests done on prenatal check-ups are routine and almost all pregnant women are to get them, while prenatal testing we discuss here is completely optional. In this leaflet, we focus on the evaluation of the fetus to find out specific chromosomal abnormalities. You can have a safe delivery without these tests and it's up to you to choose whether or not to have them.

It is becoming increasingly complicated to make a decision, with there being more options for you to consider. Genetic counseling offers support in order for you to be comfortable with your pregnancy whether or not you decide to undergo the testing.

Q2. What are the common types of prenatal tests?

A. There are two types of prenatal tests: screening and diagnostic. The former includes NIPT, maternal serum tests, or special ultrasounds such as NT scans, none of which are definitive. When the screening test result shows a possible problem, you might consider diagnostic tests, including amino or CVS, which can carry a small chance of miscarriage.

In genetic counseling, specialists will outline all the options available for you and help you decide how to proceed.

Q3. What is genetic counseling? Should every expectant mom get it?

A. Genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease, and it involves specialists who identify families at risk, investigate the problem, interpret medical information, and review available options with the family. Anyone with unanswered questions about her baby should seek genetic counseling. It provides helpful information, education, support, and very often, a peace of mind.

Q4. Is a birth defect common? How much can a test find out?

A. One in every 20~33 babies are born with a birth defect and about 25% of them are affected by chromosomal abnormality. It's impossible to detect everything about a fetus, but proper prenatal care does help ensure things go right, as it enables your doctor monitor your/your baby's health and identify any problems before they become serious.

Q5. What is a "high risk" pregnancy?

A. A high risk pregnancy is any pregnancy that can result in health problems for the mother/baby. The term sounds scary, but it simply means that extra special attention must be paid. In the framework of prenatal testing, however, "high risk" means "higher chance" of the baby having a genetic condition. Both cases require special management, and again, regular prenatal visits are very important.

Q6 Is getting tested a common option?

A. Currently, about 10% of expectant mothers over 35 years old get prenatal testing in Japan. The decision to pursue the tests is yours. People choose to have or not to have them for different reasons. The decision must be made after a thorough discussion between you and your partner, along with genetic professionals.

Q7. What are my choices if the test result shows a problem?

A. There are always some options for you, and it is a hard choice to make, especially in such an anxious time. However, you don't have to go through it all alone. Your practitioner, as well as genetic specialists, will help you decide what to do and what life will be like for your child if he/she has a disorder.

Q8. Does a health problem in the family affect the baby?

A. Only very few types of diseases can be passed on, while it is possible to inherit some types of genetic disorders. Estimating the chance of inheriting the condition can be complex. You'll be provided a detailed explanation based on your profile during your genetic counseling.

Q9.Does a disorder found on a test mean another risk factor among family members?

A. A vast majority of the family are not affected at all, but there are a few exceptions. When a genetic disorder is diagnosed, family members often want to know the likelihood that they or their children will develop the condition. The answers vary and additional genetic counseling may prove useful.

Q10. When should I see a genetic professional?

A. Genetic counseling is helpful any time during —or even before— your pregnancy. You can get the support you need when you intend to start a family, you're expecting, or after the baby is born.

Q11. What can I discuss with my geneticist/genetic counselor?

A. You can talk about absolutely anything you need to be clarified: the consequences and nature of a disorder, the options and supports open to you, family planning and parenting. Make the most of the counseling for your own best interest. It serves as a continued resource for you.

Q12. Where do I start for genetic counseling?

A. To find the right institution for you, visit the website on the back cover.

You can talk to your practitioner about where to seek genetic services. Specially-trained professionals, such as board-certified geneticists/genetic counselors will be of your help.

*Patients with limited Japanese proficiency are expected to bring an interpreter.

Q13. How can I make an appointment?

A. You can always ask a referral from your health care provider, or locate a service and make an appointment yourself. Bring your partner with you to the session if possible. Your geneticist/counselor will always be happy to assist you.

図２　リーフレット英語版

産科医療施設の先生方へ

「妊娠がわかったみなさんへ～妊婦健診で行われないおなかの赤ちゃんの検査について～」
リーフレット活用の手引き

１．リーフレット作成の目的

本リーフレットは、産科診療の場において、妊婦が出生前検査に関する情報が得られ、必要な時には遺伝カウンセリングが受けられるようにする体制づくりのために作られました。

２．リーフレットの普及の目標

各地域において、リーフレットを介して産科医療施設と遺伝カウンセリング実施施設が連携することを通して、妊婦の不安への対応がなされ、妊婦が安心して妊娠期間を過ごすことができることを目標としています。

３．リーフレットの使用にあたって

１）リーフレットの裏面に、近隣の遺伝カウンセリング実施施設を記入してください。

妊婦に近隣の遺伝カウンセリング実施施設を知らせることを目的としています。

貴施設に通う妊婦が利用しやすい地域の施設を１か所または複数記入してください。

（研究班 HP の遺伝カウンセリング実施施設一覧をご参照ください。）

２）リーフレットは、妊婦やその家族がおなかの子への不安を抱えている場合や、出生前検査の情報を更に詳しく知りたいと考えている場合など、必要な時に適切な情報を得られるよう、ご活用ください。

貴施設の体制に応じて、配布方法をご検討ください。

例１：受付カウンターや、待合室内など、妊婦の目に留まりやすい場所に設置する。

例２：妊娠初期の保健指導や母子健康手帳の交付の案内などと一緒に渡す。

３）リーフレットを見た妊婦が相談できる機会を作ってください。

リーフレット内容についての質問、補足説明の希望、などの相談があった場合には妊婦への対応をお願いします。

例１：遺伝カウンセリング実施施設に紹介する。

例２：自施設の医師または助産師等によって最初の面談を行い、更に相談を希望した場合に遺伝カウンセリング実施施設に紹介する。

※紹介先の体制に応じて、遺伝カウンセリングの予約や紹介状の記載等をお願いします。

４）遺伝カウンセリング実施施設に紹介した後について

妊婦が安心して妊娠期間を過ごせるためには産科医療施設の皆様の暖かなサポートが欠かせません。必要があれば、遺伝カウンセリング実施施設ではいつでも相談に応じます。

図３　リーフレット活用の手引き