Birth Defects vs. Chromosomal Abnormalities: What's the Difference?

Sara contacted us at the recommendation of her OB/GYN to talk about the paroxetine she takes for her depression. Sara wanted to see if taking paroxetine can increase the chance of having a baby with a heart defect. She was confused because her genetic screening result was normal. She asked: "Is it possible for my baby to have a heart defect if my non-invasive prenatal testing (NIPT) was negative?"

There are many tests that can check on the development of your baby during pregnancy. It can be difficult to keep track of what they are looking for. Knowing the differences between a birth defect and a chromosomal condition can help you understand these tests.

What are birth defects and how can you look for them?

Out of all babies born each year, about 3 out of 100 (3%) will have a birth defect. Certain exposures, like medications, can increase the chance of birth defects. Most birth defects happen during the first trimester when the baby's organs (body parts) are developing. A certain organ might look or work differently. You may have heard of a baby being born with a hole in the heart. This is a common birth defect. Ultrasounds during pregnancy can look for birth defects. Read more about ultrasounds through our blog here. Most women will have an anatomy ultrasound between 18 and 22 weeks of pregnancy. You might know this as the time when you can learn the sex of the baby. The goal of this ultrasound is to take a detailed look at the baby's organs to check for birth defects. It will also check the growth of the baby. While this is an important screening test, it is not perfect. A normal anatomy ultrasound does not rule out the baby having a birth defect or other pregnancy-related problems.

A birth defect can be an isolated finding, meaning that there are no other related health problems. A birth defect might be caused by a change in the baby's genetic information, such as a chromosomal condition.

What are chromosomal conditions and how can you look for them?

Chromosomal conditions are genetic, meaning they are a change in the baby's genetic information. Chromosomal conditions do not usually run in families and are not inherited. Chromosomes are like a recipe book that holds all the recipes for the development of the baby. If a certain recipe calls for one stick of butter, but the book accidentally says two sticks of butter, the recipe will turn out differently. In the same way, sometimes people have extra or missing chromosomes that can cause changes in development. For example, people who have a chromosomal condition known as Down syndrome have three copies of chromosome 21 rather than two. This extra amount of genetic information causes developmental differences. Babies that have Down syndrome sometimes also have birth defects that are seen on ultrasound. Other babies that have Down syndrome can have normal ultrasounds. Therefore, ultrasound is not the best way to screen for chromosomal conditions.

Your healthcare provider will offer you screening for the most common chromosomal conditions. Testing for a chromosomal condition includes a screen called NIPT or diagnostic testing through procedures called CVS (chorionic villi sampling) or amniocentesis. NIPT involves drawing blood from you as early as 8-10 weeks of pregnancy. NIPT can tell you if you have a high or low chance of having a pregnancy with certain chromosomal conditions. Diagnostic

testing can tell you “Yes” or “No” about your baby having a chromosomal condition. Diagnostic testing involves a procedure that might come with small risks to the pregnancy. These screens and tests are optional. Your healthcare provider might suggest you meet with a genetic counselor. Genetic counselors can talk with you about the pros and cons of these tests, help with decision making and interpret the results.

During our conversation, Sara asked: “So because my NIPT results were low risk, the baby is unlikely to have a chromosomal condition. However, a birth defect could have happened in the first trimester, and I need to wait until my anatomy scan to get those results. Is that right?” That is exactly right! Although some studies show an increased chance of heart defects with paroxetine use in pregnancy, most other studies do not show an increased risk. If there is an increased chance of birth defects with paroxetine use in the first trimester, it is expected to be small. See our factsheet on Paxil® for more detail.

Sara shared that she was feeling more knowledgeable about the difference between a birth defect and a chromosomal condition. She was relieved that her NIPT results showed low risk. She also felt more confident heading to her anatomy scan knowing that this test, while not perfect, would be the best way to identify birth defects during pregnancy.

If you have any questions about exposures during pregnancy, contact MotherToBaby. To find a genetic counselor in your area, visit the Find a Genetic Counselor page on the National Society of Genetic Counselors website.

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Birth Defects vs. Chromosomal Abnormalities: What's the Difference?

The arrival of spring and summer warmth spurs many of us to spend more time outdoors and make travel plans, be it for a honeymoon, babymoon, or other seasonal getaway. Such was the case with Yesenia, who contacted MotherToBaby with questions about her upcoming summer vacation after finding out she is pregnant. Yesenia knew that with warmer temperatures comes an increased chance of mosquito bites and the infections they can carry. Like Yesenia, as you engage in more outdoor activity and plan getaway time, you may also have questions about exposure to these infections and their possible effects on a pregnancy. Understanding the potential risks of these illnesses and ways to protect yourself may help you make better-informed decisions before you decide to travel. This month, MotherToBaby turns to our partner the Society for Maternal-Fetal Medicine (SMFM) to help answer some of Yesenia's questions:

What are vectors and vector-borne illnesses?

Vectors are living organisms, such as mosquitoes, ticks, biting flies or fleas, that are capable of spreading diseases to humans through bites and other forms of contact. Some common examples of these diseases, called vector-borne illnesses, include Chikungunya, dengue, malaria, Oropouche, and Zika virus. While disease-carrying insects can bite year-round, they are most active during warm weather. Many vector-borne illnesses have no cure, and treatment is usually limited to just managing the symptoms. This is why it's important to protect yourself from bites!

How can vector-borne illnesses affect a pregnancy?

There can be serious health complications for the mother with some vector-borne infections during pregnancy, such as severe anemia, bleeding, and the need for hospitalization. If the mother develops serious complications, it increases the chance of problems for the baby, such as low birth weight, preterm delivery, or stillbirth. Some vector-borne illnesses may be passed from mother to baby during pregnancy, which can lead to serious birth defects, such as microcephaly (a condition where the head is smaller than normal), other structural defects of the brain and eyes, and problems with long-term development and cognition (such as learning, understanding, and memory). For more information, see the MotherToBaby fact sheets on dengue, malaria, Oropouche, and Zika. The CDC also has information about chikungunya and other vector-borne diseases. Additionally, SMFM has fact sheets on vector-borne illnesses and pregnancy (also available in Arabic, Chinese, Korean, Spanish, Tagalog, and Vietnamese).

I am pregnant and I am planning to travel. How can I protect myself?

Here are some quick tips:

- If traveling, check your destination(s) on CDC's Travelers' Health page before booking your trip. There you will find recommended vaccines and other tips to prevent travel-related illness at each destination. Avoid

nonessential travel to any areas that have a Travel Health Notice of Level 2 or higher.

- Apply Environmental Protection Agency (EPA) registered insect repellents to exposed skin and clothing.
- Avoid outdoor activity during dusk and dawn when mosquitoes are most active.
- Minimize exposure to open water sources, such as pools and lakes, which are breeding grounds for vectors.
- Keep windows and doors closed or use fans, mosquito nets, and mesh screens on windows and doors.
- Use the CDC's Pregnant Travelers' Health Guide for other general travel tips.

Which insect repellents are EPA-registered? And are they safe for me to use during pregnancy?

EPA-registered insect repellents include:

- N, N-diethyl-meta-toluamide (DEET)
- Picaridin
- IR3535
- Oil of lemon eucalyptus
- Para-menthane-diol

The EPA has studied these insect repellents for their effectiveness and safety profile. When used as directed on the product label, there is no evidence to suggest that these insect repellents increase the chance of birth defects or other pregnancy-related complications. For additional information, please see the MotherToBaby fact sheet on insect repellents.

Are there alternative insect repellents I can use?

Other essential-oil-based insect repellent products, including disposable wristbands and patches, are often advertised as safer alternatives to EPA-registered products, but there is no scientific evidence that they effectively protect you and your baby against disease-carrying insects. In addition, the essential oils in these products, such as peppermint, citronella, and lemongrass, are not regulated by the EPA as pesticides. It's better to steer clear of these products in favor of EPA-registered insect repellents.

I've read about "parasite cleanses." Do they work on vector-borne illnesses?

Parasites are living organisms that live on or inside a host and can cause harm by damaging tissues or triggering severe immune responses. Vector-borne illnesses are different from parasitic infections. But you may have seen social media posts touting home parasite cleanses that claim to "detox the body" and "kill off parasites" and wonder whether they could help prevent these diseases. The answer is a decided "no."

These cleanses, which contain a blend of herbs, have no proven record of efficacy (doing what the label claims they do) or safety (that they will not cause unwanted symptoms), especially for use during pregnancy. Although many of these products claim to target a wide range of parasites, they can have dangerous and unknown side effects for you and your baby.

Parasitic and vector-borne illnesses are serious conditions requiring proper medical attention. They cannot be cured by these products. If you suspect that you have a parasitic infection—or any other serious illness—it's important to speak with a doctor as soon as possible, especially during pregnancy.

The bottom line

So, what's SMFM's advice to Yesenia for her upcoming travel? The bottom line for protecting herself this spring and summer from vector-borne illnesses: stay away from higher-risk areas, stick with proven, safe repellents recommended by the EPA, and get in touch with her healthcare provider if she has any questions. Safe travels to Yesenia and to you!

More about Our Guest Co-Authors from SMFM

Naima T. Joseph, MD, is Vice Chair of the Society for Maternal-Fetal Medicine (SMFM) Committee on Infectious Diseases and Emerging Threats. Dr. Joseph is an MFM subspecialist in the Department of Obstetrics and Gynecology at Boston Medical Center and an Assistant Professor at Boston University School of Medicine.

Hazel Salvador is an intern at the Society for Maternal-Fetal Medicine and a Master of Public Health (MPH) student at George Washington University. She is interested in health policy, maternal and child health, and epidemiology, and she hopes to pursue a Doctor of Public Health (DrPH) degree in the future. In her free time, she enjoys traveling and playing with her two parrots.

Birth Defects vs. Chromosomal Abnormalities: What's the Difference?

You are pregnant. You ask, "Can I take my medication?" If you ever heard, ***"it depends…",*** you are not alone. Things affect people differently.

So, what is going on?

This has to do with genetics. Genetics is the study of genes. Genes are a part of our DNA. Genes tell your cells what to do. Genes tell us how to process the world around us.

What Do We Mean by "Exposures"?

Anything that comes into contact with your body is an exposure. Food, drinks, and drugs are exposures. Getting sick is an exposure. Stress is an exposure too.

Exposure questions can feel like a lot!

Why Can the Same Exposure Affect People Differently?

People can have the exact same exposure. Their bodies might handle it differently. That is because of metabolization. Metabolization is how fast our bodies break down an exposure. This affects how long an exposure stays in your body.

Genes affect how your body breaks down something. Genes shape how the body responds to an exposure.

What Are Some Examples?

- **Caffeine and Pregnancy**

Let us say that you got coffee with a friend at 3:00 pm. You might fall asleep easily that night. Your friend might feel jittery after half a cup. You and your friend have different responses to the same amount of caffeine. That difference comes from your genetics. Your genes might metabolize caffeine faster. This is why you fell asleep easily. Your friend

might metabolize caffeine more slowly. This is why your friend felt jittery.

Genes affect how fast caffeine is broken down. This is why caffeine during pregnancy rules are careful. It is suggested that you get 200 mg of caffeine (or less) per day during pregnancy. The rules are there to protect those that process caffeine more slowly.

The end message? There are rules about pregnancy. These rules are broad. Your experience might be different than what the rules say. For more information on caffeine, click here:
https://mothertobaby.org/fact-sheets/caffeine-pregnancy/

- **General Anesthesia and Breastfeeding**

Let us say that your dentist tells you to get your wisdom teeth removed. This is a surgery. For this, you will get general anesthesia. General anesthesia is a mix of medication. This is given so that you can fall asleep for a procedure.

General anesthesia is broken down fast. Your friend got her wisdom teeth removed last year. She woke up 2 minutes after her surgery was done. You woke up 5 minutes after your surgery was done.

It takes some time for you to feel normal after general anesthesia. You might feel a little loopy right after general anesthesia. Your friend felt normal about 1 hour after her surgery. You felt normal about 3 hours after your surgery. You and your friend react differently to general anesthesia.

Genes affect how fast something is broken down by the body. This is why the rules for getting general anesthesia while breastfeeding are careful. It is suggested to wait to breastfeed if you still feel loopy. This means waiting to breastfeed until you feel “back to normal.”

The end message? Everyone needs a different amount of time to break down general anesthesia. The best time to breastfeed is different for everyone. For more information on general anesthesia, click here:
https://mothertobaby.org/fact-sheets/general-anesthesia-pregnancy/

What Does This Mean for Pregnancy and Breastfeeding Advice?

People are different. People have different genes. This means that people break down exposures in a different way.

Most recommendations try to:

- Lower the chance of any possible problems;
- Help people who might break things down more slowly;
- Balance safety with needs.

Final Thoughts

What happens to your body during pregnancy and breastfeeding can feel so hard. It can feel harder when your body reacts differently than what is expected. If you compare yourself to others, you should know you did nothing wrong. You did not need to know better. You did not fail.

You might feel overwhelmed or worried about an exposure. You might feel frustrated if you get advice that clashes. Talking to MotherToBaby can help you get information without judgment.

Birth Defects vs. Chromosomal Abnormalities: What's the Difference?

Do you enjoy protein shakes, adding protein powder to your smoothies, or using pre-workout mixes with protein? My friend, Emily, is currently in her first trimester and loves getting extra protein. Before she got pregnant, she started power lifting, which also came with extra protein supplementation. Once she got pregnant, Emily asked me, like so many others in her position, if she could continue her protein supplementation during pregnancy and when breastfeeding.

With any pregnancy, there is a 3% chance of developing a birth defect. So I looked into protein supplementation to see if it changes the risk for birth defects, side effects during breastfeeding, or any other issues. That way, Emily could make a well-informed decision for her health and the health of her family.

Many of these protein products contain types of protein that are also found in everyday foods and even in breast milk. Everyone needs a certain amount of protein every day as a part of a balanced diet. However, there are certain reasons why someone might have a specific daily protein intake goal. Like Emily, many people are interested in having more protein to build muscle. There are other health conditions, like phenylketonuria (PKU), that affect how much protein you should get in your diet. Ultimately, it is very important for you to review your personal daily protein intake goals with your healthcare team to make sure your health is supported.

Not every protein supplement has been well studied, so information on using them during pregnancy or breastfeeding is limited. In addition, many protein powders and drinks also include other ingredients, such as added vitamins or herbal products. For more information on herbal products, check out our fact sheet on here: https://mothertobaby.org/fact-sheets/herbal-products-pregnancy/.

Supplements are regulated differently than prescription drugs. Even though the United States Food and Drug Administration (FDA) regulates supplements, the FDA doesn't check every product for safety or effectiveness before it goes on the shelf. This means some protein powders might contain some unknown or unlisted ingredients.

So before adding that extra scoop of protein powder, take some time to figure out if protein supplementation is right for you.

How Much Protein Do You Need?

How much protein you need depends on different factors, such as age, activity level, and health. Some proteins have a daily recommended dietary allowance (RDA). The RDA depends on a person's weight. You, most likely, get some amount of protein from your diet naturally. It's important to take your diet into consideration when trying to figure out how much protein you should be having. Because there are so many things to consider, it's recommended to talk with your healthcare team to make sure you are meeting your nutritional needs. Getting the right amount of protein for you is not expected to increase the risk for issues during pregnancy or side effects during breastfeeding.

What are the typical amino acids in protein supplements?

Most protein powders contain many amino acids. Amino acids are the building blocks of protein. The list below may not include everything that might be found in your protein powder. Since not every amino acid has an RDA, here is a list of the known amino acids with an RDA. It's always a good idea to double check with your healthcare team on what your specific protein intake goals are.

- **Cysteine & Methionine***
  - RDA during pregnancy: 25 mg/kg
  - RDA during breastfeeding: 26 mg/kg
- **Isoleucine**
  - RDA during pregnancy: 25 mg/kg
  - RDA during breastfeeding: 30 mg/kg
- **Leucine**
  - RDA during pregnancy: 56 mg/kg
  - RDA during breastfeeding: 62 mg/kg
- **Lysine**
  - RDA for adults: 12 mg/kg
- **Threonine**
  - RDA during pregnancy: 26 mg/kg
  - RDA during breastfeeding: 30 mg/kg
- **Tryptophan**
  - RDA for adults: 3.5 mg/kg
- **Valine**
  - RDA for pregnancy: 31 mg/kg

- RDA for breastfeeding: 35 mg/kg

*Note: The RDA for methionine and cysteine is combined.

What About Other Ingredients?

Often, protein powders can have other ingredients in addition to the protein. Again, check the label on your specific products, as they can vary.

- **Caffeine**: For more information on caffeine, check out our fact sheet: https://mothertobaby.org/fact-sheets/caffeine-pregnancy/
- **Collagen:** There aren't any studies that look at the use of collagen as a supplement during human pregnancy or breastfeeding.
- **Creatine**: Creatine is naturally found in the body and in breastmilk. There aren't any studies that look at the use of creatine as a supplement during human pregnancy or breastfeeding.

So What Now?

Emily realized that since there is not a lot of information on protein supplementation during pregnancy and breastfeeding. The best next step for her was to chat with her healthcare team about what would best support her health and pregnancy. Some providers on her healthcare team include her primary care provider, her nurse-midwife, her nutritionist, and her endocrine specialist. Emily decided that she's even going to talk to a lactation consultant now so she can have time to prepare for breastfeeding. She knew how to measure how much protein she was getting in her protein powder, but she needed help estimating how much protein she naturally gets in her diet as well.

Have more questions about supplements or other exposures during pregnancy or breastfeeding? Talk to a teratogen-information specialist at MotherToBaby.org.

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Birth Defects vs. Chromosomal Abnormalities: What’s the Difference?

Thinking about pregnancy while also worrying about weight can feel stressful. You are not alone—about 6 out of 10 women in the U.S. are overweight or have obesity. Talking about weight can be hard, but it is an important part of planning for a healthy pregnancy.

This blog will explain how weight can affect pregnancy, what GLP-1 medications are, and what we know, so far, about their use before and during pregnancy.

Why Is Managing Weight Before Pregnancy So Important?

Being overweight or having obesity increases the chance for several pregnancy-related problems, including:

- Miscarriage
- Birth defects
- Preterm delivery (before 37 weeks)
- Gestational diabetes
- High blood pressure during pregnancy
- Stillbirth
- Cesarean delivery
- Thromboembolic events (blood clots)

You can read more about obesity and pregnancy in our factsheet here: https://mothertobaby.org/fact-sheets/obesity-pregnancy/

The good news is even a small weight loss—just 5–7% of your body weight—before pregnancy can improve health and pregnancy outcomes. Some people do this through healthy eating and exercise, while others may need surgery or medication.

What Are GLP-1 Medications?

GLP-1s are medicines that act like natural hormones in your body. They help control blood sugar, slow down digestion, and make you feel full longer. This can lead to weight loss. Most GLP-1s are given as shots. The best-known ones are liraglutide (Victoza®) and semaglutide (Ozempic®, Wegovy®, Rybelsus®). These are also the ones most studied in pregnancy to date.

Can I Use GLP-1s While Trying to Get Pregnant?

The current product labels recommend stopping GLP-1 medications at least 2 months before pregnancy. The time it takes the body to process medication is not the same for everyone. In healthy non-pregnant adults, it can take up to 6 weeks, on average, for most of the GLP-1s to be gone from the body.

Stopping the medication can sometimes cause weight gain, which can feel frustrating. Because of this, some people choose to continue until they know they are pregnant. It is best to talk with your healthcare providers about the risks and benefits for you.

What Do We Know About GLP-1s in Pregnancy?

Here is what the research tells us so far:

- Studies including over 4,000 women exposed to GLP-1s during the first trimester have not shown an increased chance of birth defects or miscarriages.
- The chance of other pregnancy-related problems, such as stillbirth, preterm delivery (birth before week 37) or low birth weight (weighing less than 5 pounds, 8 ounces [2500 grams] at birth) was also not increased with first trimester exposure to GLP-1s.

It is important to remember that every pregnancy has a baseline risk:

- Out of all babies born each year, about 3 out of 100 (3%) will have a birth defect.
- 15 to 20 out of every 100 (15-20%) pregnancies will end in miscarriage.

Birth defects and miscarriages typically occur in the first trimester — whether or not medication is used.

Why Are GLP-1s Not Recommended During Pregnancy?

At this time, continuing GLP-1s after pregnancy is confirmed is not recommended for 2 main reasons:

- Weight loss during pregnancy is not advised. Losing weight while pregnant might increase the chances of having a baby that is small for gestational age (SGA), which can lead to complications such as:
    - Low oxygen levels
    - Low Apgar scores (grading system in newborns to define their wellbeing)
    - Meconium aspiration (breathing in the first bowel movement)
    - Hypoglycemia (low blood sugar)
    - Difficulty maintaining body temperature
    - Polycythemia (too many red blood cells)
- We lack research on GLP-1s in the second and third trimesters. Without research studies on the use in the second and third trimester, we do not know if GLP-1s increase the chances of other pregnancy-related problems.

Finding the Path That is Right for You

Your journey is unique, and there is no simple answer. That is why it is important to talk with your healthcare providers about the best way to approach weight management before pregnancy. As Dr. Sarah Obican, a board-certified obstetrician/gynecologist specializing in Maternal-Fetal Medicine in Florida, so masterfully said in our ***Battling Obesity Ahead of Pregnancy*** blog, “Each of us are beautifully individual” — and our weight loss and pregnancy journeys are beautifully individual, too.”

Final Thoughts

Whether you are already on a weight loss journey or just starting to think about pregnancy, you deserve support and trusted information. We are here to help you every step of the way.

Helpful Links:

Factsheets:

- Obesity and Pregnancy:https://mothertobaby.org/fact-sheets/obesity-pregnancy/
- Semaglutide: https://mothertobaby.org/fact-sheets/semaglutide/
- Tirzepatide: https://mothertobaby.org/fact-sheets/tirzepatide/

Baby Blogs:

- Battling Obesity Ahead of Pregnancy is 'Beautifully Individual':https://mothertobaby.org/baby-blog/battling-obesity-ahead-of-pregnancy-is-beautifully-individual/

Podcasts:

Ep. 64: Weight Loss and Ozempic in Pregnancy: https://mothertobaby.org/podcast/ep-64-weight-loss-and-ozempic-in-pregnancy/

Ep. 84: GLP-1 Medications & Pregnancy: What We Know So Far: https://mothertobaby.org/podcast/ep-84-glp-1-medications-pregnancy-what-we-know-so-far/

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