Global Genes®
RARE Toolkits

DECEMBER 2022

Family Planning and Financial Considerations

About **Global Genes®**

Global Genes is a 501(c)(3) non-profit organization dedicated to eliminating the burdens and challenges of rare diseases for patients and families globally. In pursuit of our mission we connect, empower, and inspire the rare disease community to stand up, stand out, and become more effective on their own behalf—helping to spur innovation, meet essential needs, build capacity and knowledge, and drive progress within and across rare diseases. We serve the more than 400 million people around the globe and nearly 1 in 10 Americans affected by rare diseases. If you or someone you care about has a rare disease or is searching for a diagnosis, you can *contact us by submitting our confidential form*. A Patient Services Guide will provide you with a personalized response within 2-3 business days that will include information, resources, and connections that address your specific needs.

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Acknowledgments

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Introduction

"When my husband and I were discussing whether or not to have children, we definitely discussed the possibility of passing XLH onto our children. We decided to go ahead and have children. We knew that it was a risk to have a child with XLH, but we also believed that we would love our children no matter what and that we would be responsible in taking our children to specialists and getting them the medical care that they need."

– RACHAEL JONES, XLH PATIENT /PARENT ADVOCATE,
XLH NETWORK INC, BOARD MEMBER

Choosing to start or expand a family is one of the most significant decisions couples will make in their lives. It is a highly personal one that may be influenced by a person's religion, societal norms, a biological urge to have a child, or even a desire to fill your home and world with more children.

The decision to have a child is filled with hopes, dreams, love, and longing. Yet it is also a decision that involves emotional, physical, psychological, lifestyle, and financial factors that must be carefully considered. This is especially true for people living with a rare disease, and those who are carriers. ***Always keep in mind that your decisions are uniquely your own, highly personal, and there are no right or wrong answers.***

This toolkit is designed to help you and your partner explore and prepare for genetic and rare disease tests and family planning options and their associated financial considerations. It is intended for couples where one or both people:

- are living with a rare disease
- are a carrier of a rare disease
- know there is a family history of a genetic disorder

The toolkit also features tips and advice from people living with a rare disease, carriers of a rare medical condition, and experts who can help you choose different family planning paths. Their insights and shared experiences, together with the information and resources provided, will help you

choose the family planning approach that is healthiest, safest, and best for your family and your financial situation.

Consider this a high-level guidebook with links to credible resources for you to dive into a wide range of topics and questions.

And, of course, Global Genes is always here to help you in any way we can. To contact us, *fill out our confidential Patient Services form*. Within 2-3 business days, you will receive a personalized response that will include information, resources, and connections that address your specific needs.

Family Planning and Rare Inherited Conditions

When you and your partner make the decision to add a branch to your family tree it is important to do some advanced family planning to help choose the best path to grow your family. For couples living with a rare disease or where one or both partners are carriers of a rare disease, family planning can help to understand:

- the genetic inheritance of their disease
- potential risks and symptoms that could be experienced during pregnancy and/or childbirth that could make it difficult, dangerous, or not feasible for a woman to experience
- options and costs for fertility preservation

Family Health History

Knowledge is key. To begin your family planning discussions, meet with your doctor or healthcare team who manages your condition so that you and your partner can discuss medical and family histories. This is important because it is one of the tools your doctor can use to keep you and your future children healthy.

Talking about your family's health history can feel overwhelming to some people. To get you started, here are some questions to consider as you gather your family history:

- Do multiple people in my family have similar health problems and have not had a formal diagnosis?
- Do certain health issues seem to be passed on to the next generation of my family?
- Have there been multiple miscarriages in my family?
- Have women in my family had problems during pregnancy or birth, or have not been able to have children?

Once you have talked to your relatives and gotten information about what conditions run in your family, you are now ready to share this information with your doctor. They can determine how to best take care of you, particularly as you plan your family.

Deciding To Have Children When You Have A Rare Disease

In *this video* Maria Della Rocca, Genetic Counselor and Senior Director of Patient Services and Programs, Global Genes, reminds us how personal the decision is to have a child and discusses what to consider when you start family planning.

Global Genes Rare Disease Concierge Patient Services

Global Genes RARE Concierge
Global Genes offers a free patient information and referral service to people living with rare diseases and those who support them including, partners, caregivers, care partners, families, friends, and healthcare providers and teams.

- Our patient services guides are individuals dedicated to improving the quality of life for all those with rare diseases, including those who are undiagnosed. Information provided is based on unique individual needs and experiences.
- You will be connected with a wide range of resources such as locating a genetic counselor, healthcare specialists, offering financial, mental health, and disability resources, and more.
- Visit *Global Genes Concierge - Patient Services* to start getting help today.

Learn about Global Genes RARE Concierge

Additional Resources

CDC's Family Health History and Planning for Pregnancy

The Family Medical History as a Tool in Preconception Consultation - PMC

Genetic Risks to the Mother and the Infant: Assessment, Counseling, and Management - PMC

Genetic Counseling

Genetic information has become integral to preconception and prenatal care planning. Seeking guidance from a genetic counselor and undergoing genetic screening are important steps in your family planning process as they help you and your partner determine the best approach to grow your family. Each family's decision is personal and reflects their unique situation. What is the best choice and plan for one family is not necessarily best for another.

Genetic counselors have specialized training in both medical genetics and counseling. They can help you unravel the answers to your questions and concerns. They help you and your partner understand genetic risks and test results and consider the medical, psychological, and ethical considerations of having a child.

What Genetic Counselors Can Do:

- help to identify families at possible risk of a genetic disorder
- provide information and support to families affected by or at risk of a genetic disorder
- gather and analyze family history and inheritance patterns
- determine if genetic testing is right for you
- share options you have in terms of testing and ways to pay for testing
- help translate genetic test results and calculate risks of recurrence
- provide support for the psychological/emotional impact of testing

Genetic Counseling and Shared Decision Making

Genetic counselors will not make decisions for you, but they will listen, share guidance and talk through test results, learnings and options. You can expect your genetic counselor to:

- listen to your thoughts and feelings
- help you think through your values
- answer any questions you have
- arm you with information, and
- support you in making the best choice for you, your future child and family.

Genetic counselors are particularly helpful if tests reveal a strong likelihood that your future child may inherit or potentially be born with a rare medical condition. Some couples may choose to pursue natural conception with or without prenatal testing, and others may choose to pursue a different family planning route. If you do decide to have prenatal testing, it's important to discuss with your partner, prior to pregnancy, whether to continue with the pregnancy if the child is likely to either have or be a carrier of a genetic mutation. Genetic counselors can talk to you about your options and also help you understand what inconclusive test results mean for you.

Should you choose to not continue with your pregnancy, you can discuss this with your care team. Contact your local *Planned Parenthood* to learn options available in your state.

The Questions to Ask Your Genetic Counselor

- Does the disease in question run in families?
- If my family member has a disease, might I get it?
- If I have a disease, are my family members at risk of getting it?
- Is any kind of genetic testing available? If so, what are the benefits and limitations of the testing? What are the costs and are the tests covered by health insurance? or, How will I pay for it?
- What kind of information can genetic testing give me?
- What does the genetic testing process involve?
- If I decide to have genetic testing for myself or my child, when can I expect to hear about the results? Will the results be given to me over the phone or in person?
- How can knowing more about a genetic risk help me?
- Could I be exposing myself or my family to discrimination based on genetic information?

Source: *Making Sense of Your Genes: A Guide to Genetic Counseling published by Genetic Alliance*

How Can A Genetic Counselor Help?

Maria Della Rocca, Genetic Counselor and Senior Director of Patient Services and Programs, Global Genes, *highlights the role genetic counselors can play* and how they can support you during your family planning process.

RARE TIP

"For women approaching family planning/preconception decisions, it can be very helpful to meet with a genetic counselor. A genetic counselor can help to discuss all available options and provide support through often challenging decisions. I always encourage families to start the conversation early so they have time to consider all of their available options in the context of their own personal experiences, resources, values, and beliefs."

–JODIE M. VENTO, MGC, LCGC, CHILDREN'S HOSPITAL OF PITTSBURGH OF UPMC

Learn More

American Board of Genetic Counseling Inc.

Global Genes Resource: Making Informed and Shared Decisions About Genetic Testing and Clinical Trial Participation

Global Genes Resource: Gene-Based Diagnosis 101: How to Successfully Navigate the Diagnostic Journey

National Society of Genetic Counselors (also maintains a "Find a Genetic Counselor" directory to help you locate a genetic counselor near you)

Preconception and Prenatal Genetic Testing

Genetic tests are medical tests that can help determine changes in genes, chromosomes and proteins. They can be performed before conception and during pregnancy to help families understand the chance that an existing rare condition could be passed down or that a genetic disorder could develop.

The costs for genetic tests can range from a few hundred to more than $2,000. Be sure to contact your health insurance company to confirm coverage and out-of-pocket costs. In addition, talk with your genetic counselor who can help you find ways to pay for testing, including low/no cost options.

Preconception or Carrier Testing – Carrier testing can determine if you or your partner carry certain genes that can lead to a genetic disorder. If you both carry a certain gene there is a greater chance that your child could inherit the genetic disorder.

Preimplantation Genetic (PGT) Testing – If you or your partner are using alternative reproductive technology (ART), such as IVF, to conceive a pregnancy, this test is performed using cells from the embryo to identify one or more specific inherited conditions.

Prenatal Genetic Testing – Prenatal genetic tests are performed on the fetus during the first and second trimesters to diagnose genetic disorders.

Genetic Inheritance Patterns: How Rare Conditions are Passed Down

What are Genes, DNA, and Chromosomes?

- Genes are found within every cell of the body.
- Typically, each person has two sets

of genes in every cell. One set from a female and one set from a male.

- Every gene contains sections of DNA or instructions that tell the cell how to function and grow and what sex the person will be.
- The DNA is made of four different chemicals which are paired in different ways. These pairings create genetic code.
- DNA is tightly wrapped together in structures called chromosomes. Every normal cell has 23 pairs of chromosomes (for a total of 46).

Genetic Inheritance and Genetic Disorders

Genetic inheritance refers to a trait or variants encoded in the DNA and passed from the genes of males and females to babies during reproduction. The risk of a genetic disorder being passed down is dependent on the inheritance pattern of that specific condition. For some inheritance patterns, both male and female genes must be a carrier of the genetic disorder. For other disorders, only one person needs to be a carrier or have the disorder to be passed on to a baby.

As you and your partner move through the family planning process and decide whether or not to have genetic testing and which tests to have, it will be important to understand the type of genetic condition and the gene inheritance pattern of the disorder you and/or your partner have or carry. According to the *American Board of Genetic Counselors*, there are three types of inherited conditions:

- **Monogenic disorders** are caused by a variant or mutation in a single gene. The variant may be present on one or both chromosomes (one chromosome inherited from each parent). Examples of monogenic disorders are: cystic fibrosis, Huntington's disease, and sickle cell disease.
- **Multifactorial disorders** are caused by a combination of variations in genes, often acting together with environmental factors. Behaviors are also multifactorial, involving multiple genes that are affected by a variety of other factors. Examples of multifactorial disorders are: Alzheimer's disease and congenital heart disease. Note - Since these diseases are caused by variants in a large number of genes (and more causative genes are being discovered everyday), these diseases are harder to test for prenatally.
- **Chromosome disorders** are diseases that are caused by either changes to the number of chromosomes (such as adding or missing chromosome), or when certain areas within the chromosome are deleted, duplicated, or incorrectly added onto a different chromosome. This results in the disruption of genes that are located on these chromosomes so that the genes can no longer do their normal function. Even very small changes to the structure of chromosomes can have significant effects. An example of a chromosome disorder is Down syndrome, also known as Trisomy 21.

Monogenic Disorder Inheritance Patterns

Most rare diseases are monogenic or single gene disorders. The gene mutation is usually inherited in a simple pattern, depending on the location of the gene and whether one or two normal copies of the gene are needed. This is often referred to as Mendelian inheritance because Gregor Mendel first observed these patterns in garden pea plants.

The table below briefly describes the gene inheritance patterns for single gene disorders and includes examples of rare diseases that have these patterns. You can also view this *slideshow on how genetic disorders are inherited* created by the *Mayo Clinic* for a visual representation of how these patterns are created.

Inheritance Pattern	Characteristics	Disease Examples
Autosomal Dominant	• Can be passed down by one partner. • Partner can be either male or female. • Occurs in every generation.	• Huntington's disease • neurofibromatosis • achondroplasia • familial hypercholesterolemia
Autosomal Recessive	• A person can be a carrier and never show symptoms and never know he/she has the trait. • Both partners need to carry the trait to pass it on to the baby. The child can inherit the genetic condition or be a carrier. • One quarter of children will get an autosomal recessive gene if both parents have it. • Not typically seen in every generation.	• Tay-Sachs disease • sickle cell anemia • cystic fibrosis • phenylketonuria (PKU)
X-linked Dominant	• Both males and females are affected, but more females are affected. • Females are less severely affected. • Males cannot pass down to males. • Affected females can transmit the disorder to males and females. • Can be present in both males and females in the same generation if the mother carries the trait.	• Hypophatemic rickets (vitamin D resistant rickets) • ornithine transcarbamylase deficiency
X-linked Recessive	• Mostly affects males, but female carriers may have symptoms. • Transmitted through symptomatic carrier females or asymptomatic carrier females to males or females. No male-to-male transmission. • Affected males often present in each generation.	• Hemophilia A • Duchenne muscular dystrophy
Mitochondrial	• Can affect both males and females, but can only be passed on by females because all mitochondria of all children come from the mother. • Can appear in every generation.	• Leber's hereditary optic neuropathy • Kearns-Sayre syndrome

A Special Note about De Novo Genetic Disorders and Prenatal Screenings: A de novo genetic disorder is a genetic alteration that appears for the first time in one family member as a result of a variant (or mutation) in a germ cell (egg or sperm) of one of the parents, or a variant that arises in the fertilized egg itself during early embryogenesis. Currently screening for many dominant monogenic disorders associated with de novo mutations is not available.

Learn More

ACOG's Carrier Screening FAQ

Financing of Genetic Testing and Screening Services

Medline Plus: What Are The Different Ways A Genetic Condition Can Be Inherited

What is Fertility Preservation and Who Benefits From It?

Fertility preservation is a process where you extract, save, and protect egg, sperm, or reproductive tissue in order to use them to have biological children in the future. You may benefit from fertility preservation if you have one of these conditions or before undergoing a treatment that could affect your reproductive health:

- Have endometriosis
- Have uterine fibroids
- Are about to undergo a stem cell transplant or cancer treatment
- Are about to be treated for an autoimmune disease, such as lupus
- Have a genetic disease that affects future fertility preservation

Note: If you are a parent of a child who needs to undergo cancer treatments, talk to your pediatric oncologist and pediatrician about the potential risks and effects on your child's reproductive health and options for fertilization preservation.

What are my fertility preservation options?

- **Ovarian Transposition** - ovaries are surgically moved out of the pelvis and preserved.
- **Egg and/or Embryo Banking** - freeze a woman's eggs or embryos for future fertilization/ implantation.
- **Sperm Banking** - freeze sperm to be used for fertilizing a woman's egg in the future.
- **Tissue Banking** - the ovarian cortex or testicular tissue is surgically removed and frozen and is later transplanted back into the body and hormonally stimulated to produce eggs and sperm.

Navigating the Financial Aspects of Fertility Preservation

Costs for different fertility preservation treatments vary by storage bank facility, clinic, and geography. If you do not have insurance coverage, you will probably have to pay out of pocket for these services and will want to research and compare your options for your specific treatment and yearly storage fees. For more information or to begin your

research, visit the *Alliance for Fertility Preservation* web page on estimated costs.

While most health insurance companies do not cover fertility preservation costs, do check with your insurer to learn if any portion is covered. You can also contact organizations that can find ways to offset costs. Start your research with these *organizations*.

RARE TIP

If you are a cancer survivor or about to undergo cancer treatment check with your oncologist or cancer treatment center about financial assistance programs and if your state has mandated health insurance coverage for fertility preservation for people living with cancer.

According to the *National Academy of State Health Policy* since 2017, 15 states have introduced legislation that would mandate coverage of fertility preservation; five states – Connecticut, Rhode Island, Maryland, Delaware, and Illinois — have enacted this legislation, and New Jersey has a bill pending.

What is Oncofertility?

With advances in the treatment of many cancers, the survival rates for adolescents and young adults with cancer has increased. Individuals who have survived childhood/young adult cancer may decide to start a family and often assume that their reproductive health will be preserved after treatment. However, cancer treatments including chemotherapy, radiation, and surgeries that affect reproductive organs can have an impact on fertility.

The need to address cancer and fertility issues has given rise to the field of oncofertility. Oncofertility encompasses the care from medical professionals specializing in both cancer and fertility. Oncofertility specialists work with men and women to:

- Address concerns related to risks to fertility, fertility preservation options, and management of fertility complications related to cancer treatment
- Provide psychological support and counsel on options such as donor egg and sperm, gestational surrogacy, and adoption

RARE TIP

"At a time when so much can feel overwhelming and uncertain, it is important that we, as people living through the diagnosis, have the opportunity to keep options open and available for the future. All fertility complications and preservation options must be shared with the patient because it is their right to have the ability to make a decision that is best for them and their family."

- ROB LONG, EXECUTIVE DIRECTOR OF UPLIFTING ATHLETES

Family Planning Options

If genetic testing shows that you and/or your partner are at increased risk of having a child with an inherited condition, or if you are impacted by a rare condition that can make pregnancy or childbirth challenging, there are several options you can consider. Having open and honest conversations with your partner about the medical, emotional, and financial impact of each option is essential.

Your physician and genetic counselor can help you think through these options:

- Go ahead with a natural pregnancy, understanding that your future child could be affected by the genetic condition.
- Schedule prenatal diagnostic testing (PND) early in your pregnancy, with the option of terminating the pregnancy, or go forward using the information you receive to prepare for any necessary specialized care for mother and baby.
- Reduce the risk of an inherited condition by going through In Vitro Fertilization (IVF) to retrieve and fertilize eggs, then use Preimplantation Genetic testing (PGT) to identify unaffected embryos that can be implanted in the mother's uterus. IVF can be done using your own and your partner's eggs and sperm, or a donor's eggs and sperm.
- Find a surrogate to carry your child if you have a condition that makes it unsafe to go through pregnancy and birth.
- Look into adoption, locally or internationally.
- Choose not to pursue growing your family.

RARE TIP

When you are confronted with a choice of healthcare options, the Australian General Practice Network recommends asking yourself three questions:

1. What are my options?
2. What are the possible risks and benefits?
3. How likely are these risks and benefits?

Find more strategies for choosing what's best for you and your partner in Global Genes' toolkit on *Shared Decision Making*

Prenatal Genetic Tests (PNGT)

As noted in the Preconception and Prenatal Genetic Testing summary section there are two main types of prenatal genetic tests:

1. **Prenatal Screening Tests**
   a. Carrier tests involve analyzing blood or tissue samples (from inside the cheek) to determine if you or your partner carry a mutation for a certain genetic disorder
   b. Cell free DNA is a blood test that screens for certain conditions caused by an abnormal number of chromosomes
2. **Prenatal Diagnostic Tests**
   1. Chorionic villus sampling (CVS): Usually carried out between 10-13 weeks by taking a small sample of cells from the placenta and analyzing cells for certain genetic disorders
   2. Amniocentesis: Usually performed in the second trimester (15-20 weeks) by inserting a very thin needle in the uterus and collecting a small sample of amniotic fluid which is analyzed for certain genetic disorders

RARE TIP

The American College of Obstetrics and Gynecology provides more in-depth information in their *FAQs on Prenatal Genetic Diagnostic Tests* and *Prenatal Screening Tests* and in this *infographic of prenatal screening and diagnostic tests.*

5 Things to Know About PNGT

1. Even if you know in advance that you would not choose to terminate a pregnancy, testing can help relieve anxiety and/or prepare in advance for a high-risk birth.
2. Changes in abortion laws may have an impact on whether or not you have the option to terminate a pregnancy by the time test results are in.
3. There is a small risk of miscarriage from invasive procedures (1/200 for Chorionic Villus Sampling (CVS) and 1/900 for Amniocentesis.
4. Check with your insurance carrier to see which of these tests will be covered.
5. For both CVS and Amnio, you need to know the genetic condition you are at risk for, either through carrier testing or previous genetic testing.

Reducing the Risk of Genetic Disease with IVF and PGT

With In Vitro Fertilization (IVF), you will receive hormone treatments to trigger ripening and release of multiple eggs. The eggs are retrieved from your ovaries using a very small needle, then fertilized in the lab using your partner's (or a donor's) sperm. Fertilized eggs are allowed to grow into a five-day embryo. Then the embryos are tested for chromosomal problems and your and/or your partner's specific genetic mutation related to the disease for which you are testing. Only an embryo that is free of known conditions will be selected for implantation where it continues to grow.

Learn About IVF and PGT

4 Questions to Consider

Alison Weisman, MS, CGC, Genetic Counselor, Lurie Children's Hospital Assistant Professor of Pediatrics, Northwestern University Feinberg School of Medicine, recommends asking yourself these questions as you consider reproductive options:

- How would I feel if my child had the condition?
- Would I want to know if a pregnancy is affected by the condition? So I could:
  - Plan and prepare
  - Consider termination
  - Decrease my anxiety
- Do I want to consider alternative ways my child will not have the condition?
  - In Vitro fertilization with Preimplantation Genetic Testing
  - Donor egg or sperm
  - Adoption
- Should I consider alternative ways to have a family, such as surrogacy, due to my medical needs?

Adoption

Families are created in many different ways, including adoption. If this is an option you are drawn to, there are multiple pathways to consider, including private or public agencies, adoption attorneys, and domestic and international adoption.

This *Fact Sheet from the Child Welfare Information Gateway* is a good resource to help you locate service providers and learn about the process of adoption.

One Family's Story

Jessica Fein, a rare disease caregiver and MERFF Advocate, *shares her experience with international adoption*.

Planning for Baby - Financial Considerations

As you plan for the safest and healthiest way to grow your family, take time to also consider your anticipated costs. From natural childbirth, to the use of reproductive assistive technologies, to adoption, the costs can vary widely. Some may be covered by your health insurance and others will have high out-of-pocket costs. Planning in advance from a financial perspective, allows you time to explore financing options that may be available to you.

Creating a Family - Pregnancy and Non-Pregnancy Options - A Financial Overview*

Use the table below to jump start your financial research of pregnancy and non-pregnancy options.

Option	Covered by insurance?	Cost range (without insurance)
In vitro fertilization with preimplantation genetic testing (IVF with PGT)	Rarely	$16,800-$26,000 *This is the cost for one cycle
Amniocentesis	Often	Without Termination for Medical Reasons (TFMR): $1,000-$7,000
Chorionic villus sampling (CVS)	Often	Without TMFR: $1,400-$5,000
Egg donor	Rarely	$14,000-$40,000
Embryo donor	Rarely	$2,000-$7,000
Sperm donor	Rarely	$1,000-$4,000
Sperm banking	Rarely	$1,000 for the first year (+$150-$300 per year after the first year for storage)
Egg freezing	Rarely	$10,000-$15,000 (+$300-$500 per year for storage)
Embryo freezing	Rarely	$11,000-$15,000 (+$400-$600 per year for storage)
Ovarian tissue freezing	Rarely	$10,000-$12,000 (+$300-$500 per year for storage)
Surrogacy	Rarely	$100,000-$150,000
Adoption	N/A	$20,000-$50,000 (depending on the type of adoption) *Adoptive families may be eligible for tax credits and/or reimbursements

*Costs and health insurance coverage are applicable only in the United States.
*For other countries, the availability and cost depends on their healthcare system. For example, in England, three rounds of IVF with PGT are covered *for couples who meet certain criteria*.

Sources:
https://www.forbes.com/health/family/how-much-does-ivf-cost/
https://www.allianceforfertilitypreservation.org/paying-for-treatments/
https://healthcareinsider.com/maternity-costs-310937
https://www.childwelfare.gov/pubpdfs/s_costs.pdf
https://www.healthline.com/health/fertility/freezing-sperm#cost

Nine Strategies to Help Finance Your Family Planning Options

Depending on your personal situation, financial resources, and the family planning option or options you choose, financing your family planning can quickly become challenging. Here are 10 strategies to help you get organized, discover potential savings and funding sources, and make the experience less stressful.

1. Create a Financial Plan that could include:

- action steps you need to take - who to contact, when, and their contact info
- current financial resources and savings
- incurred and anticipated expenses such as planning for multiple egg retrieval cycles when undergoing IVF or fertility preservation. (an *online* templated tracker can help you track your expenses and savings goals)
- anticipated additional healthcare and health provider costs should you choose to pursue childbirth and your rare disease puts you in the category of a high-risk pregnancy, which may or may not be covered by your health insurance plan
- Note: If you need to undergo an immediate treatment that may affect your reproductive organs and do not have time to plan, contact a genetic counselor about financial assistance programs you can quickly access
- potential healthcare expenditures in the case your child will need genetic testing, treatments, or interventions after birth

2. Contact your insurance provider and employee benefits manager (if employed) to:

- understand all treatment and family option costs that are fully or partially covered, and individual or family deductibles

3. Compare the costs across different medical facilities or adoption agencies.

- Costs for hormone medications, treatments, and procedures such as IVF with PGT, and egg and embryo freezing can widely vary across clinics.

4. Maximize your Health Savings Account (HSA) or Flexible Spending Account (FSA),

- If your employer offers them, fund these accounts with pre-tax dollars to pay for medical treatments such as IVF.

5. Use a Health Reimbursement Account (HRA) to get reimbursed from a certain medical expenses

- If your employer offers an HRA, consider using it to cover the costs for certain medical expenses not covered by your health insurance benefits.

6. Contact financial assistance programs to see if you are eligible and become familiar with the application process.

- Note: when searching for IVF grants, many of them require a diagnosis of infertility. This means if you are pursuing IVF for reasons other than infertility, such as to utilize PGT, you may not be eligible for funding.
- **Financial assistance programs can be** international, national, or state-specific so be sure to check eligibility. Here are a few to consider:
  - *Alliance for Fertility Preservation*
  - *The Associates: Adoption Grant (Florida only)*
  - *Baby Quest Foundation*
  - *Chick Mission*
  - *HelpUsAdopt.org*
  - *Northern California Fertility Medical Center*
  - *RESOLVE: The National Infertility Association*

7. Talk to your accountant, financial advisor, or the Internal Revenue Service (IRS) about tax savings or deductions for your out-of-pocket medical expenses and/or adoption fees included in your yearly tax return.

8. Contact your financial advisor or loan officer at your bank or research online lenders who provide loans for high-cost medical procedures such as IVF or for supporting adoption fees and costs.

9. Consider creative and unconventional ways to engage your family's and friends' financial support.

- Directly ask family or friends for donations
- Conduct a crowdfunding effort with a GoFundMe fundraiser

Birth Plan

If you and your partner choose to have a pregnancy, it is recommended that you create a birth plan to help your labor and delivery experience go as smoothly and safely as possible. If you or your partner's rare condition could cause complications during labor and delivery be sure to include any special medical issues and complications that could arise and discuss with your obstetrician–gynecologist before your due date.

It is important to remember that a birth plan is an outline and cannot always go as planned, but it does give your obstetrician–gynecologist and labor and delivery care team good guidance, and plan in advance for any possible challenges.

Your birth plan should also include:

- requesting any information about genetic or other tests that the new baby should have while in the hospital following childbirth
- contacting the hospital's neonatal intensive care unit (NICU) should the baby need its care after delivery
- securing health insurance for your baby by contacting your health insurance company or *Children's Health Insurance Program (CHIP)* or *Medicaid* if you qualify for free or low-cost health insurance

You do not have to start a birth plan from scratch. There are many online templates and plan outlines. Use this *sample birth plan template* from the American College of Obstetricians and Gyneocologists to get started.

RARE TIP

"As a rare disease patient it is important you share with your doctor/medical team as soon as you know you plan on trying to have a baby. This is important because they can tell you if you should stop any of your medications. In addition to the specialist you see, you should also be offered genetic counseling, referral to a high risk OBGYN in addition to a normal OBGYN. I'd also suggest looking into a doula or midwife for added support during the pregnancy. These are all the things I did and had when I was pregnant with my now 1 year old son. Remember, pregnancy is a beautiful thing, even if it's high risk. Enjoy it!"

- SHAMONICA WIGGINS-MAYES, SOCIAL MEDIA MANAGER OF THE SICKLE CELL COMMUNITY CONSORTIUM AND PATIENT ADVOCATE

Learn More

CDC: Pregnancy Complications | Maternal and Infant Health

Children's Hospital of Philadelphia: How to Cope When Your Unborn Baby is Diagnosed with a Birth Defect

March of Dimes: Birth defects and your baby

Mayo Clinic: Postpartum complications: What you need to know

Nemours Kid Health: Birth Plans

Rare Stories

Knowing how other community members approach family planning decisions may help you think through your own options. In this panel discussion, you'll hear from two passionate rare disease advocates:

- **Taylor Kane**, founder of Remember the Girls, an organization dedicated to raising awareness of the issues faced by females with X-linked diseases, to advocate for greater access to reproductive options, genetic testing, clinical trials, and to offer communal support.
- **Carrie Ostrea**, Associate Director of Patient Advocacy at Passage Bio and Board Chair of Little Miss Hannah's Foundation, an organization with a mission to help enhance the quality of life for young children diagnosed with rare, life-limiting, or undiagnosed complex medical needs, as well as those in hospice or palliative care. The foundation was created to honor her 3-year-old daughter Hannah, who lost her battle with Gaucher Type 2/3, an ultra-rare genetic disorder.

Glossary

Carrier screening - is a type of genetic test that can tell whether you or your partner carry a gene for certain genetic disorders. When it is done before or during pregnancy, it allows you to find out your chances of having a child with a genetic disorder.

Chromosomes - tightly wrapped structures of DNA found in each cell. Every normal cell has 23 pairs of chromosomes (for a total of 46).

DNA - Deoxyribonucleic acid is found in genes and made up of four chemicals that form pairs in different combinations. These combinations create codes for different genes.

Fertility preservation - a process that extracts, saves, and protects egg, sperm, or reproductive tissue in order to use them to have biological children in the future.

Genes - Sections of DNA that are found inside every human cell and carry information about different traits such as eye color, body type, male or female sex, etc. that get passed down or inherited from one generation to the next.

Genetic counseling -professional services to assess and advise individuals about the risk of an inherited disorder, its consequences, and available options.

Genetic inheritance - to a trait or variants encoded in the DNA and passed from the genes of males and females to babies during reproduction. The risk of a genetic disorder being passed down is dependent on the inheritance pattern of that specific condition.

Genetic tests - medical tests that can help determine changes in genes, chromosomes, and proteins. These changes may be a sign of a disease or condition, such as cancer. They may also be a sign that a person has an increased risk of developing a specific disease or condition, or of having a child or other family member with the disease or condition.

Invitro Fertilization (IVF) - a hormone treatment used to help with fertility or prevent genetic problems and assist with the conception of a child. These hormone treatments trigger ripening and release of multiple eggs. The eggs are retrieved from a woman's ovaries using a very small needle, then fertilized in the lab using the partner's (or a donor's) sperm.

Monogenic disorders - genetic disorders caused by a variant or mutation in a single gene. The variant may be present on one or both chromosomes (one chromosome inherited from each parent). Examples of monogenic disorders are: cystic fibrosis, Huntington's disease, and sickle cell disease.

Preimplantation genetic (PGTD) Testing – screening test performed on embryos created using alternative reproductive technology (ART), such as IVF, to analyze the embryos and identify one or more specific inherited conditions prior to the embryo transfer to the person carrying the pregnancy. Prenatal diagnostic tests: These tests can tell you whether your fetus actually has certain disorders. These tests are done on cells from the fetus or placenta obtained through amniocentesis or chorionic villus sampling (CVS).

Prenatal screening test - Test that can tell the chances that a fetus has an aneuploidy and a few other disorders.

Preimplantation genetic diagnosis (PGD) - a screening test used to test embryos produced through in vitro fertilization (IVF) to detect genetic or chromosomal disorders.

Resources

Family Health History

CDC's Family Health History and Planning for Pregnancy

The Family Medical History as a Tool in Preconception

Genetic Risks to the Mother and the Infant: Assessment, Counseling, and Management - PM

Genetic Counseling

Global Genes Resource: How a Genetic Counselor Can Help You

National Society of Genetic Counselors: Pregnancy and Family Planning

Preconception and Genetic Testing

American Board of Genetic Counseling, Inc.: Family Planning

American College of Obstetrics and Gynecology: FAQs on Prenatal Genetic Diagnostic Tests

NCBI Bookshelf: Inheritance Patterns - Understanding Genetics

Cleveland Clinic: Autosomal Dominant & Autosomal Recessive Inheritance & Disorders

NCBI Bookshelf: Genetics, X-Linked Inheritance - StatPearls

Fertility Preservation

Eunice Kennedy Shriver National Institute of Child Health and Human Development: What is Fertility Preservation?

Genetic Inheritance Disorders

Cleveland Clinic: Autosomal Dominant and Autosomal Recessive

Understanding Genetics: A New York, Mid-Atlantic Guide for Patients and Health Professionals

American Board of Genetic Counselors: Inherited Conditions

Family Planning Options

American Board of Genetic Counselors: Family Planning

American College of Obstetricians and Gynecologists: Prenatal Diagnostic Tests

American College of Obstetricians and Gynecologists: Prenatal Screening Tests

Centers for Disease Control and Prevention (CDC): Assisted Reproductive Technology (ART)

What is Assisted Reproductive Technology?

Financial Considerations

A Child Waits Foundation

Adoptions Together: Convinced You Can't Afford Adoption? 6 Ways to Offset Adoption Costs

Fertility Space Blog: How to Write Off IVF on Your Taxes

Fertility Space Blog: Tips on Saving to Afford IVF

Genetic Alliance UK: How Can I Access Preimplantation Genetic Diagnosis

HSA Store: Fertility Treatment: HSA Eligibility

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