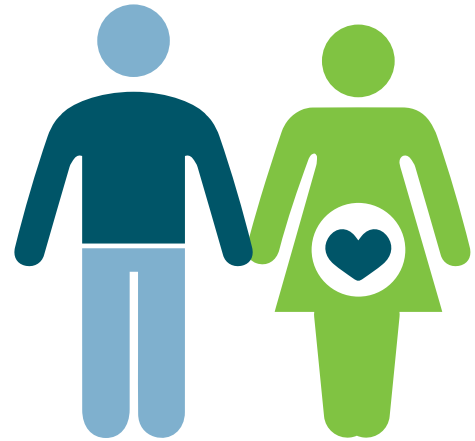




WOMEN WITH RARE DISEASE: THE REPRODUCTIVE YEARS

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Introduction

As a woman, you will experience distinct biological stages as part of your life cycle: childhood through puberty, the reproductive years, and menopause. Choosing whether or not to have a family is one of the most significant decisions you will make and either choice will affect your life in untold ways. For every woman, the choice is unique and highly personal.

The decision to have a child involves emotional, physical, psychological, financial and lifestyle considerations. You may experience a biological urge to have a child or feel influenced by societal norms or your religion. Every pregnancy comes with some degree of risk and deciding to have a child is a leap of faith for anyone.

As a woman living with a rare disorder, you have the additional implications of considering

how your condition may impact a pregnancy and the degree of risk of passing on your genetic disorder to your children. There is no right or wrong answer. This toolkit was designed to help you consider this important choice by providing questions and resources to guide your decision. If you decide to have a child, it will help you plan for pregnancy and adapt to life as a mother who must also manage her own rare condition.

To further complicate matters some women find themselves responsible for the care of both a child and an aging parent who may or may not have the same genetic disease. The toolkit also touches on the unique challenges faced by “The Sandwich Generation,” of individuals who are serving as caregivers for both their children and parents at the same time.

SECTION 1:

CHALLENGES OF FAMILY PLANNING WITH A GENETIC DISEASE



Advances in reproductive technology have made it possible for many more women to have children. Increased information and technology presents more options. Genetic information has become integral to pre-conception and prenatal care.

Discussing your decision with your spouse or partner and the doctor who helps to manage your condition is a first step. Your doctor will be able to discuss your personal medical history, your family history as well as your partner's. It will also be important for you to see a genetic counselor and to undergo genetic screening to determine the probability of passing on your condition to your child. You may be able to get a referral from your primary care physician or obstetrician or gynecologist. The National Society of Genetic Counselors (www.nsgc.com) also maintains a "Find a Genetic Counselor" directory that you can search.

A genetic counselor is a clinician with specialized training in genetics and counseling who can help you unravel the answers to your questions and concerns. They are trained to help you understand genetic risks, test results and to consider the medical, psychological and ethical considerations of having a child. They will prove particularly helpful if tests reveal a strong likelihood that your child will be born with a medical condition. Many women choose to have a child even if the tests reveal the

existence of a genetic condition. Others may choose to terminate. The decision of whether or not to have a child or to abort following a diagnosis is ultimately yours and once you will make with your husband or partner, but one that may be helpful to discuss prior to planning a pregnancy.

"For women approaching family planning/pre-conceptual 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

A genetic counselor can also provide information about starting a family with genetic technologies that might be available to minimize risks in certain cases. Most importantly, a genetic counselor can provide support and resources for you and your family, regardless of your choices surrounding a potential pregnancy. Everyone perceives risks differently and each woman has unique values and circumstances that contribute to her decisions.

SECTION 1: CHALLENGES OF FAMILY PLANNING WITH A GENETIC DISEASE

Rachael Jones, XLH Patient /Parent Advocate XLH Network Inc, Board Member

“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.”

Tonia Sina, Founder, The Beauty in Illness

“I do not have children. I had kidney failure at the age of 18 and didn't know at the time that it was caused by a rare disease, atypical hemolytic uremic syndrome (aHUS). I have had two kidney transplants. My husband and I decided early on that having children was not going to happen. It would be too risky to my health, and I didn't want to risk passing on my aHUS to a child. Personally, it would be totally unacceptable to me, and their future children, and I really wouldn't be able to live with myself for the possible outcomes. There are also days that I'm too sick to get out of bed and I feel it would be extremely selfish to put a child and my husband through that. I's 35 now and I understand the urge; I hear the biological clock ticking. But the truth is my husband and I are very happy with our lives and with our decision not to have children.”

Tara Voogel, Patient Advocate, Co-Founder, GNE Myopathy

“When my spouse and I decided on having children, we knew that two of my sisters had some kind of muscle disease. We did not know what it was or whether or not it was a genetic disease. They were living outside the U.S.A. where the appropriate testing and diagnostic tools were not available to determine what my sisters were suffering from. Nevertheless, I consulted two neurologists at the Good Samaritan Hospital in Los Angeles. They took a biopsy from my quadriceps muscle, the results of which were declared as “inconclusive.” This biopsy was done in 1982 at which time I had not yet been definitively diagnosed as having GNE Myopathy that was discovered in 2011. At that time (1982) I did not know that I had a genetic disorder so my spouse and I decided to start a family. Although there was a kind of perpetual underlying fear that I may have contracted the same disease my two elder sisters suffer from, I did not have any fear that my children would likewise be affected. My two children are now healthy adults, and I feel very fortunate to have them in my life.

GNE Myopathy is a recessive genetic disorder. My illness did not affect my physical interactions while my son was a child, and I showed no signs at that time of having GNE Myopathy. By the time my daughter was in middle school I had declined physically that I was unable to do many things with her such as going to the mall with her or attending events at school.

My advice to would be parents who have a genetic disorder, yet would like to start a family should consider getting genetic testing as now there are advances in technology. One such procedure is called “Pre Implantation Genetic Diagnosis” or (PGD)”.

SECTION 2: THE UNIQUE JOURNEY THROUGH PREGNANCY AND DELIVERY



Your pregnancy will bring a multitude of physical and emotional changes for you and your family. Each day may feel different than the next as you adapt to the physical and emotional changes. This is true for any pregnancy, but your genetic condition or predisposition may pose additional concerns related to your health and the health of your baby. It is normal to feel anxiety from the tests, risks and information provided. Although you likely have a partner, family and friends to support you, they may not always agree with your values and perceptions of risks. Just as you did when you were making the decision to conceive, it is important to make informed decisions as you navigate your pregnancy.

It is important to speak with your healthcare provider(s) who will help to manage your prenatal care; as well as, balance your medical needs with the needs of your baby. They will also help to identify any risks and plan for any special precautions needed at the time of delivery.

Your prenatal care will likely include different types of prenatal genetic testing options. Each has its own benefits and limitations. Some testing provides only an estimate of risk while **amniocentesis** or **chorionic villi** sampling are diagnostic and convey specific results but do not test for every potential birth defect. Tests also have risks so it is important to fully understand your options. The American Pregnancy Association has a chart (<http://americanpregnancy.org/prenatal-testing>) that describes some of the various testing options that you may be offered. A genetic counselor can also help you to decide whether to test and which test is best for your circumstances.

It is normal to feel overwhelmed. Your clinicians and genetic counselors should strive to empower you with the information, support, and tools you need to make the best decisions for you and your family. Be sure to ask questions and discuss your concerns with your healthcare provider and partner.

Normally a woman's pregnancy is managed by her obstetrician, midwife or doula. Because of your genetic condition you will also continue to see your primary doctor and may have concerns with continuity of care among providers. You will need to be your own advocate and keep careful records of your medical history, test results and any symptoms you experience from either the pregnancy or your illness. The Global Genes Building a Care Notebook toolkit describes how to coordinate medical records and healthcare information. (<https://globalgenes.org/toolkits/building-a-care-notebook/carenotebook>).

Most mothers experience some anxiety over the health of their growing baby. Although no physician can guarantee a healthy baby, your doctor is trained to monitor the fetus's growth and detect any potential concerns. Focus on being diligent about prenatal care, proper nutrition and rest.

Each baby arrives in his or her own time and way, and it is not predictable. Some deliveries are smooth and others require more intervention. If you have any special needs for delivery your doctor will have prepared for them.

SECTION 2: THE UNIQUE JOURNEY THROUGH PREGNANCY AND DELIVERY

Rachael Jones, XLH Patient /Parent Advocate XLH Network Inc, Board Member

“When my husband and I first got married we wanted to wait awhile before considering children. After 3 years, we began to discuss having children vs. adopting. After about six months of talking and research we decided to have children because I believed they would have more resources than I did growing up. It took me almost a year to get pregnant, and I had a miscarriage at about 7-8 weeks, but they didn’t know if it was related to my illness. I waited about three months before trying again and then got pregnant right away. It was considered a high risk pregnancy. I had my endocrinologist, an ob-gyn and a high-risk ob-gyn and I had more ultrasounds, and monthly blood tests. My phosphorous levels were extremely low and my endocrinologist was over medicating me which made me very ill; but they were finally able to stabilize with alternative medication. My doctor decided that the baby’s head was too large for a safe delivery given the lack of flexibility in my hips and scheduled a C-section. The delivery went perfectly fine with the exception of some lingering side effects of anesthesia.”

Adriana Venegas , Patient Advocate, Huntington’s Disease, President, Huntington’s Disease (HD) Society of America (HDSA), Greater Los Angeles Chapter

My mother was diagnosed with HD in 2009 and we became involved with the Centers of Excellence at UCLA and the local chapter of Huntington’s Disease Society of America. I was not ready to be tested initially. My sister did not inherit Huntington’s, but my brother and I both did. I am six months pregnant. It’s far different than what I imagined. I had planned to have in vitro fertilization. I elected not to have an **amniocentesis** because abortion is not an option for me. I think if my mother had been tested, maybe she wouldn’t have had me and I can’t imagine that. My pregnancy is going well, but there is a lot of guilt and a lot of fear. Even in our HDSA chapter, this is a hot topic with very strong opinions on both sides of the fence in regards to having a child. But, I know I was given this beautiful boy for a reason. I will introduce him to HDSA at an early age. We will talk about Huntington’s so it is comfortable for him. The average age of onset is 30 so he will have a healthy and happy childhood.”

Maya Doyle, Ph.D., LCSW

“While there is sometimes a sense of guilt when a child has an inherited genetic disease (whether parents were aware of it or not before the child is born and/or diagnosed), families who have previously lived with a disease themselves or witnessed it in another family member may have strong and varied feelings. They may not want a child to “suffer”. Some may want the disease to “stop in their generation”. Families who already have an affected child may be hesitant to do so again, or may feel willing to take on potential risk since they are particularly capable of managing the illness. Individuals who have lived with a condition themselves or witnessed it in a family member may feel strongly about valuing a child with a disease or disability as the unique individual that they are. Women may face stigma for having a child with a disability or rare disease...or for choosing NOT to have that child if genetic and prenatal testing reveals abnormalities. They may also face difficult decisions about terminating a pregnancy.”

SECTION 3: MOTHERHOOD



Your baby will undoubtedly fill your life with joy and love. At the same time, adjusting to a newborn that depends on you 24/7 will prove physically and emotionally demanding. You may be exhausted from delivery, experience hormonal shifts from childbirth especially if you are breastfeeding. And like all new mothers, you are unlikely to get much sleep. It can be overwhelming for anyone. If you or your child has additional complications from a genetic condition, it may be even more difficult.

It is important to rest. If your baby is sleeping—SLEEP! Accept all offers of help from friends and family and if you need more—ask. If you are experiencing unusual anxiety or depression, speak to your doctor. It could be exhaustion or it could be **post-partum depression**.

It is also important to have your primary doctor and pediatrician to monitor your health and that of your baby for routine wellness and any issues either of you may have specific to your genetic condition. Keep your care notebook and start one for your baby to record medical visits, symptoms and other information.

For most, diagnostic tests or symptoms will reveal whether or not your baby has inherited your condition. The reality of having a child with a rare disease may be more daunting than you anticipated. In fact, you may be surprised to find that the emotions and challenges are much different than those you have experienced managing your own illness.

It will prove easier to navigate the challenges of parenthood by connecting with other families. You will likely form friendships with other mothers of babies and young children. You may also find that it is especially important to take advantage of support and advocacy groups specific to your condition if available or rare diseases in general.

Connecting with others who share similar experiences can provide much needed emotional support and a reliable source of information regarding services and resources. You have likely acquired expertise and amassed knowledge relative to your condition; but find that you need to fortify your arsenal to navigate parenting, pediatric and specialty care, child care, and other child specific needs. Those in the rare disease community may be able to provide a level of compassion, support and understanding that others simply do not have the experience to offer.

Most importantly, relax and enjoy the journey. There is no play book. Accept that you will make mistakes as a mother and that there will be difficult days and heartbreaking moments. But there will also be love, unlike anything you have experienced before.

SECTION 3: MOTHERHOOD

Becky Mock, XLH Patient/parent Advocate XLH Network Inc, Board Member, Past President

“In 2003, I was at Duke Medical Center with my son and daughter. We were seeking a doctor with knowledge and experience with XLH patients as a transition for my children from their retiring pediatrician to someone treating teens/adults. I randomly told a nurse at the office that I would like to start a support group for parents with the same disability. The nurse asked if I knew about the XLH Network. I didn’t know what XLH even stood for nor had I ever heard of the XLH Network. So this random remark led me to connect with the online XLH Network. I found them online, joined the listserv and participated as I had time over the next several years. I found so many answers and caring support from others with XLH. After I retired, I joined the Board, later became President of the XLH Network, and helped organize three XLH Day events bringing people with XLH together in different parts of the country. If this nurse hadn’t used the term XLH, I never would have known my disease was called X-Linked Hypophosphatemia or perhaps ever connected with the Network. So random!!

As a parent, I’ve had to learn about the American with Disabilities Act, IDEA and 504 plans for school assistance. Physicians may never see another person with a rare disease or XLH so it’s up to you as a parent to work closely with the doctor to find a correct diagnosis and appropriate treatment. For a young parent, it takes time before you realize that the doctor does not have all the answers and you must become your own expert, taking the lead to search for a proper diagnosis and care for your children. Partnering with your doctor is critical. Finding a doctor that has the time and interest in working with an exceptional patient is more difficult than you would imagine. Health care appointments are too brief, so extensive discussions are hard to arrange at times. Thank goodness for the internet and advocacy groups for rare diseases! On your own you can find comprehensive information on many rare diseases online. The next problem is to determine if the information is accurate or opinions of other parents. That takes a great deal of time and study on your own.

Rachael Jones, XLH Patient /Parent Advocate XLH Network Inc, Board Member

“Blood tests revealed that my son’s blood levels were normal but his legs were bowed. My father told me my brother had also tested negative at birth and suggested I continue to have my son tested. At about 13 months, he was diagnosed with XLH and put on medication which helped to straighten his legs. My daughter was born a year after my son. We didn’t have her tested the first year because my son’s blood tests were traumatic for him and revealed nothing for the first year. She also tested positive. Taking medication and going to the doctor is normal to my children. My husband and I coordinate their care, appointments and advocating for them. We want to give them as normal a life as possible.

It’s very different having XLH and being a mother of children with XLH. Growing up, my mom was not consistent in giving me medication because she was concerned with kidney damage and we didn’t really track my illness. As a mother, I’m hyper-vigilant because I believe it gives my children the best chance. But, I’m so busy taking care of them; I still don’t take care of myself like I should.

I guess what I would say to anyone with a genetic disorder who is considering having a child is to make sure you are ready for the time it will take to care for a child with an illness. And if you have a child who inherits your disorder, be their advocate, stay on top of the research, and communicate with and question doctors. And, involve yourself in support groups.”

SECTION 4:



MANAGING YOUR DISEASE WHILE CARING FOR YOUR CHILD(REN) AND AGING PARENTS

Motherhood may become the focal point of your life, but all of the other areas of your life will not cease to exist. If you have aging parents, you may find yourself among “The Sandwich Generation” torn between caregiving responsibilities for your child or children and one or both of your parents. At some point most of us have to deal with aging parents and potential illnesses or injury. Being a caregiver or securing care, helping to manage medical and living conditions and more can be a full-time job.

Being apart of the sandwich generation is becoming more common today than ever with longer life expectancy. More individuals today are growing older while caring for their parents and children. People are also finding themselves in the sandwich generation longer as many children are “boomeranging” back to their homes after moving away or choosing to live with their parents longer for financial reasons.

“Genetic testing and technology have improved so much that people are being diagnosed earlier and living longer resulting in multigenerational issues,”
 –Jodie M. Vento, MGC, LCGC, of Children’s Hospital of Pittsburgh of UPMC

The sandwich generation is something not everyone fully anticipates nor considers all the personal implications. Time, money,

housing, caregiving. Often, this generation unfortunately finds themselves with financial concerns. Caring for multiple generations and yourself may mean you have to work longer to financially support those you care for. The tradeoff then is not being as available for day-to-day care, so caregiving or day care facilities need to be looked at which in turn increases the financial burden.

Living in the sandwich generation can also be emotionally tasking and self-care is important to remember. In caring for your parents and children, you are on constant alert and at times are torn between support for each. If neglecting your self-care, feelings of guilt and anger and resentment can bubble up for a number of reasons. In general, caring for multiple generations is not something most people plan so when it occurs it may be an adjustment.

“Moms tend to let themselves and their health lag a lot. It’s important to encourage them to take care of themselves so they can be the best caregiver possible. Women taking care of both children and parents may deal with very real Mommy guilt; and at the same time perceive that their struggles are not the same as those of their parents.”
 –Jodie M. Vento, MGC, LCGC, of Children’s Hospital of Pittsburgh of UPMC

SECTION 4: MANAGING YOUR DISEASE WHILE CARING FOR YOUR CHILD(REN) AND AGING PARENTS

Now imagine all of this, but you, your parent, and child all have a genetic rare disorder. A difficult aspect of managing a rare disorder while living through the sandwich generation is the emotional toll it can take and the worry and grief you can feel. Caring for and watching your parents age with the same rare disorder as you and your child, can be like looking into the future. You will witness firsthand what the progression of the disorder may look like later in life for you and your child. If your parent is suffering a large decline in health and abilities, it is grief you have to encounter much sooner than later. Strategies for coping with this along with proper self care is absolutely vital to successfully navigating the sandwich generation.

As with each lifecycle stage, information, resources and support are critical to managing whatever you may face. The variables are too numerous to offer a one size fits all solution but utilizing the same skills you have acquired in managing your own condition and caring for your child or children will prove invaluable.

The Caregiver Bill of Rights (<http://www.aplaceformom.com/senior-care-resources/articles/caregiver-support>) may prove helpful in managing your emotions and decision-making. Depending on the situation you may have to make decisions regarding in home care, assisted living or a nursing home. You may need to have end of life discussions or talk about legal and financial matters. AARP (www.aarp.org) has a wealth of resources on this topic. You will need to communicate with your mother or father's physician and you will need to identify senior resources in your community.

GLOSSARY

Amniocentesis—a needle is inserted into the uterus to obtain a sample of amniotic fluid which is then used to screen for genetic abnormalities in a fetus.

Chorionic villus sampling (CVS) or chorionic villous sampling—a prenatal diagnostic test used to determine chromosomal or genetic disorders in the fetus.

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

Genetic predisposition or genetic susceptibility is a **genetic** characteristic which influences the possible phenotypic development of an individual organism within a species or population under the influence of environmental conditions.

Post-partum depression—mood swings, crying, anxiety are common the first few days to two weeks after pregnancy but some new mothers experience a long-lasting form of depression known as postpartum depression.

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

RESOURCE GUIDE



The American Congress of Obstetricians and Gynecologists (ACOG) (<http://www.acog.org/>) The American College of Obstetricians and Gynecologists is a professional association of physicians specializing in obstetrics and gynecology in the United States. Their website has a “For Patients” section and a FAQ Preconception Carrier Screening that helps explain what it is, how it’s done and what types of results can be expected.

Caregiver Action Network (www.rarecaregivers.org)

The Caregiver Action Network (CAN) works to improve the quality of life for the than 90 million Americans who care for loved ones with chronic conditions, disabilities, disease, or the frailties of old age. CAN (formerly the National Family Caregivers Association) is a non-profit organization providing education, peer support, and resources to family caregivers across the country.

Centers for Disease Control and Prevention: Planning for Pregnancy

(<http://www.cdc.gov/preconception/planning.html>)

Family Tree Templates (<http://www.familytreetemplates.net/>)

Free family tree templates that can help you start recording information about your family and extended family medical history that might be relevant when meeting with your clinician.

Global Genes (www.globalgenes.org)

Global Genes™ is one of the leading rare disease patient advocacy organizations in the world. They work to build awareness, educate the global community, and provide critical connections and resources that equip advocates to become activists for their disease. It produces fact sheets, toolkits, podcasts, webinars, documentaries and other resources for patients and their families.

Genetic Alliance (www.geneticalliance.org)

Genetic Alliance is one of the world’s leading nonprofit health advocacy organizations. Its network consists of more than 1,200 disease-specific advocacy organizations, universities, private companies, government agencies and public policy organization.

How to Care for Aging Parents (<http://careforagingparents.com/>)

This book by Virginia Morris has been called indispensable by AARP and “the Bible of Elder Care” by ABC World News.

March of Dimes (www.marchofdimes.org)

The March of Dimes has led the way to discover the genetic causes of birth defects, to promote newborn screening, and to educate medical professionals and the public about best practices for healthy pregnancy. The March of Dimes works to end premature birth and other problems that threaten babies. They provide information and support to families.

RESOURCE GUIDE



Medline Plus (<https://www.nlm.nih.gov/medlineplus/ency/article/000816.htm>)

MedlinePlus directs you to information to help answer health questions. MedlinePlus brings together authoritative information from the National Library of Medicine (NLM), the National Institutes of Health (NIH), and other government agencies and health-related organizations. MedlinePlus also contains extensive information about drugs and supplements, an illustrated medical encyclopedia, the latest health news, and surgery videos. Written for patients, their families and friends, MEDLINE PLUS contains information about diseases and conditions, treatments, drugs, supplements, medical definitions and terminology, view videos and illustrations and links to the latest medical research and clinical trials.

National Society of Genetic Counselors (www.nsgc.org)

The National Society of Genetic Counselors (NSGC) promotes the professional interests of genetic counselors and provides a network for professional communications. NSGC's Find a Genetic Counselor directory has been developed to assist physicians, patients and other genetic counselors in locating genetic counseling services.

National Health Council (www.nationalhealthcouncil.org)

The National Health Council (NHC) brings together all segments of the health community to provide a united voice for the more than 133 million people with chronic diseases and disabilities and their family caregivers.

National Organization for Rare Disorders (NORD) (<http://rarediseases.org/>)

NORD provides a unified voice 30 million people living with rare diseases, their families and caregivers through patient advocacy, patient and professional education, mentorship for patient organizations, research support, and a patient assistance program.

PubMed (<http://www.ncbi.nlm.nih.gov/pubmed>)

PubMed comprises more than 25 million citations for biomedical literature from MEDLINE, life science journals and online books. Citations may include links to full-text content from PubMed Central and publisher websites.

Rare Diseases Clinical Research Network (www.rarediseasesnetwork.org)

The **Rare Diseases Clinical Research Network (RDCRN)** an initiative of the Office of Rare Diseases Research, made up of [22 research consortia](#) and a [Data Management and Coordinating Center](#) that are working together to improve availability of rare disease information, treatment, clinical studies, and general awareness for both patients and the medical community. The RDCRN provides up-to-date information for patients and assists in connecting patients with advocacy groups, expert doctors, and clinical research opportunities.



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<http://globalgenes.org/toolkits>

June 2016