XLH TRANSITIONS TOOLKIT

Your guide to navigating changes in health and life with **X-linked hypophosphatemia**

WELCOME!

Everyone goes through transitions, or changes, at certain times in their life. During these times your day-to-day life, routine, priorities, or responsibilities may change. For someone living with a chronic and progressive condition like XLH, these times can also come with unique or specific challenges.

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WHAT ARE TRANSITIONS?

The word "transition" in healthcare was first used to describe the time when patients move from a childhood doctor to an adult doctor. Since then, the concept of transitions has expanded to include several types of life changes such as:

- Being diagnosed with a rare or chronic condition
- Becoming a teenager/young adult
- Starting to manage your own care
- Moving out of the house and becoming independent
- Getting married and/or starting a family
- Entering the workforce, changing jobs, or leaving the workforce

This toolkit is designed to provide information and resources to help young adults with XLH and their parents/caregivers prepare to transition to adult healthcare. It will also spark some thoughts about other parts of adulthood with XLH – from dating to managing mental health.

"The knowledge that XLH is a progressive, lifelong disease has helped me understand that I need to be on top of things at every stage in my life."

FOCUS ON: MEDICAL TRANSITIONS

XLH is a lifelong, progressive disease, so it is important for young adults with XLH to transition from a pediatrician to an adult care doctor to continue their disease management. This can be a difficult process because:

- Patients and families often develop close relationships with pediatric doctors.
- It can be hard to say goodbye to someone who has cared for a family or child for many years, helped them through difficult experiences, or cheered them on.
- It's hard to find doctors who have experience with XLH and who are willing and able to care for new patients.

Preparing far in advance for this situation through gradual steps and conversations will help make sure everyone is ready when it's time. This toolkit contains tips and advice to help make the transition to an adult care doctor easier and more seamless.

MAKING THE SWITCH

The American Academy of Pediatrics suggests that parents and doctors begin to plan for transition as early as age 12. The goal is to make the change to adult healthcare sometime between age 18 and early 20s.[1]

Finding the right doctor is a key part of the medical transition, but it's also important for young patients to:

- Be aware of changes in the healthcare system, laws, insurance, and self-care.
- Understand how the symptoms of XLH may change over time.
- Learn how to communicate effectively with healthcare providers.
- Know how to make a medical appointment and access their medication.

Ultimately, the medical transition should be a team effort among young patients, parents/caregivers, pediatric specialists, and adult healthcare providers.

FOCUS ON: SELF-ADVOCACY

As you strike out on your own, learning self-advocacy skills will help you gain confidence in educating others about XLH and communicating your needs. Think about the people with whom you interact frequently – family members, healthcare providers, employers, or others. What do they need to know? Do they need to know every detail, or would a broader explanation be enough? How might the emphasis of your story or communication differ from one person to the next? This toolkit, and the resources below, can help you practice these skills.

- Share Your Rare Journey
  ultrarareadvocacy.com/wp-content/uploads/2018/08/Ultragenyx_ShareYourRareJourney_080218_v7.pdf
- Communicating with Healthcare Providers
  https://ultrarareadvocacy.com/wp-content/uploads/2018/08/Ultragenyx _DosAndDonts_080918_v3.pdf

FOCUS ON: DISEASE MANAGEMENT

There are many facets to managing your own healthcare. Sticking to your XLH treatment plan and making sure you keep up with medical appointments may be harder now that you're on your own juggling work, school, family, or personal schedules. Resources included in this toolkit can help you.

- The **XLH Educational Brochure** provides in-depth information on the condition, including how it causes symptoms within the body and how it's passed down in families.
- The **Self-Care Assessments** will help you understand where your current comfort level is with different types of disease management activities.
- The **Doctor Discussion Guide** will help you make sure you get the most out of your appointments.

DID YOU KNOW?

Privacy and consent laws prevent parents from making decisions for their children after they turn 18, and they aren't allowed to be in the doctor's office without their child's permission.

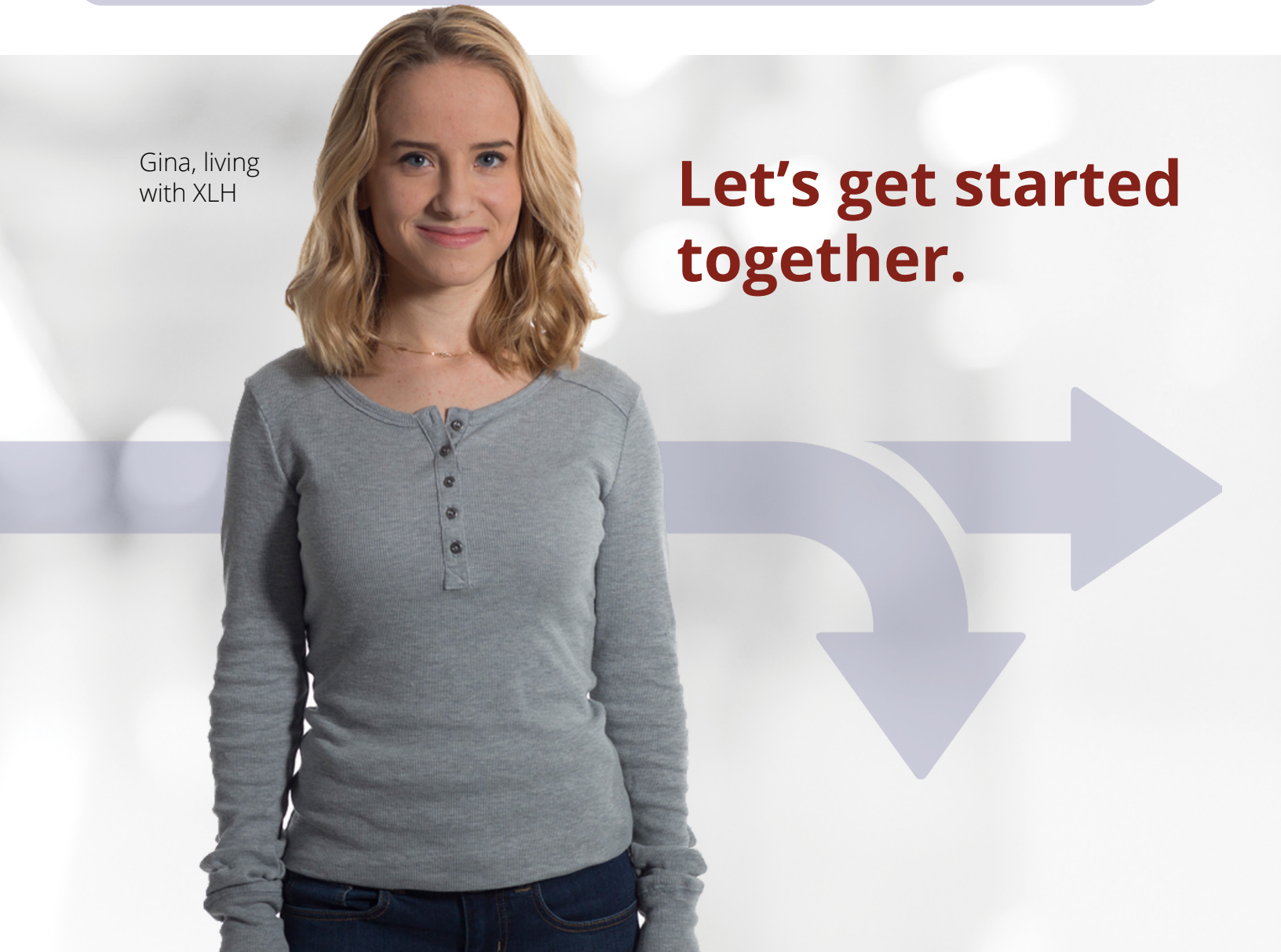
FOCUS ON: SOCIAL AND EMOTIONAL NEEDS

Meeting the social and emotional needs of someone living with XLH can be critical for their overall well-being, and these needs will change over time. The medical transition happens during "emerging adulthood" when young adults can feel uncertain about the future.[2] That uncertainty can be compounded by living with a chronic and progressive condition.

As with many rare and chronic illnesses, people with XLH may:

- Struggle with depression and/or feelings of isolation.[3]
- Experience bullying due to their physical appearance and symptoms.
- Need resources for speaking with friends, potential romantic partners, and/or employers.

"Sometimes XLH feels so overwhelming and you can feel helpless. But there are things you can do to feel empowered from an early age."



Gina, living with XLH

Let's get started together.

TOOLKIT CONTENTS

Making the Medical Transition

- Self-Care Assessments *(8-11, 12-14, 15-18, 19+)*
- Transfer of Medical Care Letter Template

Managing Your Health

- Understanding XLH
- Doctor Discussion Guide
- Tips for Communicating with Healthcare Providers
- Understanding Health Insurance

Taking Care of Social and Emotional Needs

- Addressing Mental Health
- Heading to College
- Navigating the Workplace
- Dating and Family Planning

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Download the full **XLH Transitions Toolkit** at **XLHlink.com**

REFERENCES

1. Preparing for the transition from pediatric to adult health care: Parent guide. Got Transition; 2019. https://www.gottransition.org/resourceGet.cfm?id=497. Accessed June 17, 2019.
2. Arnett, J. *Emerging adulthood: The winding road from the late teens through the twenties*. Oxford University Press, USA; 2004.
3. Uhlenbusch N, Lowe B, Harter M, Schramm C, Weiler-Normann C, Depping MK. Depression and anxiety in patients with different rare chronic diseases: A cross-sectional study. *PLoS ONE.* 2019; 14(2): e0211343. https://doi.org/10.1371/journal.pone.0211343

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X-Linked Hypophosphatemia (XLH) Self-Care Assessment

Ages 8 to 11

Fill out this form with your parent or caregiver. Be as honest as you can. There are no right or wrong answers. Your answers will help you create a self-care plan for when you get older. You can fill out this form once a year and see how your answers change.

My Condition: XLH

	My Answers			Parent/Caregiver Assessment		
	Yes	No	Not Sure	Yes	No	Not Sure
I know what XLH is and I can explain it to my friends.	☐	☐	☐	☐	☐	☐
I understand that I can't give XLH to others.	☐	☐	☐	☐	☐	☐
I can describe how XLH makes my body feel.	☐	☐	☐	☐	☐	☐

Write down how you would explain or describe what XLH is to a friend or family member.

Taking Care of Myself

	My Answers			Parent/Caregiver Assessment		
	Yes	No	Not Sure	Yes	No	Not Sure
I can explain to my teacher or others what I need to do for XLH.	☐	☐	☐	☐	☐	☐
I know what my teachers and school nurses need to do to help me with XLH.	☐	☐	☐	☐	☐	☐
I know what medicines I take for XLH.	☐	☐	☐	☐	☐	☐
I know what days and times I take my XLH medicine.	☐	☐	☐	☐	☐	☐
I know how to take my XLH medicine.	☐	☐	☐	☐	☐	☐

Write down the medicines you take for XLH.

When do you take them?

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MY DOCTOR APPOINTMENTS

	My Answers			Parent/Caregiver Assessment		
	Yes	No	Not Sure	Yes	No	Not Sure
I know my doctors' names and how they care for me.	☐	☐	☐	☐	☐	☐
I understand why I have special doctors for XLH.	☐	☐	☐	☐	☐	☐
I know I can ask my doctor questions.	☐	☐	☐	☐	☐	☐
I ask my doctor to explain when I don't understand something they say.	☐	☐	☐	☐	☐	☐
I want to talk to my doctors more about XLH.	☐	☐	☐	☐	☐	☐

When I think about XLH, I wonder:

_____

_____

_____

_____

I want to ask my doctor about:

_____

_____

_____

_____

NEXT STEPS

Bring this self-care assessment to your next appointment with your child's primary care provider (pediatrician) and their XLH specialist (endocrinologist, nephrologist, or other). You can start the transitions discussion by asking:

When do you recommend my child starts to see you alone for a portion of their appointment?

Does your office have a template for a medical transition plan?

How will the transition plan be updated as my child grows?

Can we review our transition plan with you on a yearly basis?

Download the full **XLH Transitions Toolkit** at **XLHlink.com**

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X-Linked Hypophosphatemia (XLH) Self-Care Assessment

Ages 12 to 14

Fill out this form with your parent or caregiver. Be as honest as you can. There are no right or wrong answers. Your answers will help you create a self-care plan for when you get older. You can fill out this form once a year and see how your answers change.

XLH Knowledge	My Answers			Parent/Caregiver Assessment		
	I know this	I am still learning this	I don't know/do this yet	They know this	They are still learning this	They don't know/do this yet
I know what XLH is and can explain it to my friends.	☐	☐	☐	☐	☐	☐
I can describe how XLH makes my body feel, including the things that hurt me.	☐	☐	☐	☐	☐	☐
I know whether or not my parents have XLH.	☐	☐	☐	☐	☐	☐
I understand that I will need to change my doctor to a different doctor as an adult.	☐	☐	☐	☐	☐	☐
I understand that the ways XLH affects my body will change over time.	☐	☐	☐	☐	☐	☐

Write down how you would explain or describe what XLH is to a friend or family member.

_____

_____

_____

_____

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	My Answers			Parent/Caregiver Assessment		
	I know this	**I am still learning this**	**I don't know/do this yet**	**They know this**	**They are still learning this**	**They don't know/do this yet**
I know my doctors' names and where to find their contact information.	☐	☐	☐	☐	☐	☐
I make my own doctor appointments.	☐	☐	☐	☐	☐	☐
I can talk to my doctor about my concerns or questions about how I manage XLH.	☐	☐	☐	☐	☐	☐
I know to ask for an explanation when I don't understand something my doctor says.	☐	☐	☐	☐	☐	☐
I can provide a summary of my medical history (such as dates and types of surgeries, family history, allergies, and medications).	☐	☐	☐	☐	☐	☐
I know how to fill out medical forms.	☐	☐	☐	☐	☐	☐
I know what health information I should carry with me.	☐	☐	☐	☐	☐	☐
I know my health insurance information (plan, coverage).	☐	☐	☐	☐	☐	☐

To assist in planning for your next doctor's visit, you can also reference this toolkit's *Doctor Discussion Guide* for advice on speaking with healthcare providers and getting more out of your appointments.

When I think about my health in the future, I wonder about:

_____

_____

_____

_____

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When I think about my future, these are the things I want to do:

NEXT STEPS

Bring this self-care assessment to your next appointment with your child's primary care provider (pediatrician) and their XLH specialist (endocrinologist, nephrologist, or other). You can discuss how best to teach your child these steps to self-care and start the transitions discussion with your doctor. Consider asking:

- When do you recommend my child starts to see you alone for a portion of their appointment?
- Does your office have a template for a medical transition plan?
- How will the transition plan be updated as my child grows?
- Can we review our transition plan with you on a yearly basis?

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NOTES

X-Linked Hypophosphatemia (XLH) Self-Care Assessment

Ages 15 to 18

Fill out this form before visiting your doctor and review it with your parents. Be as honest as you can. There are no right or wrong answers. Your answers will help you create a plan for when you turn 18 and take over responsibility for your own care. You can fill out this form once a year and see how your answers change. You may also want to bring it to your next doctor visit.

How important do you think it is to learn about managing your own healthcare, where 1 is least important and 10 is most important? Please circle one.

1 (least)	2	3	4	5	6	7	8	9	10 (most)

XLH Knowledge	I feel comfortable with this	I am still learning this	I don't know/ do this yet
I know what XLH is and how it affects my body.	☐	☐	☐
I know how to talk to my teachers or employer about needs I may have in the classroom or work environment.	☐	☐	☐
I know how I was diagnosed with XLH.	☐	☐	☐
I know the tests I should receive during my doctor visits and how often to monitor XLH symptoms.	☐	☐	☐
I know what kind of XLH I have (spontaneous or inherited).	☐	☐	☐
I understand how XLH can be passed down in families.	☐	☐	☐
I can find my medical history (dates and types of surgeries, family history, allergies, and medications).	☐	☐	☐
I understand how XLH symptoms may change over time.	☐	☐	☐

For more information about XLH, including symptoms, inheritance pattern, and management, read the "Learn more about X-linked hypophosphatemia (XLH)" educational brochure in this toolkit.

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SELF-CARE

	I feel comfortable with this	I am still learning this	I don't know/ do this yet
I am comfortable with my current XLH treatment plan.	☐	☐	☐
I can explain my treatment plan to a doctor or nurse.	☐	☐	☐
I know the consequences of not taking care of XLH.	☐	☐	☐
I know my medication schedule and what to do if I miss a dose.	☐	☐	☐
I know when I'm supposed to receive my medication (my medication schedule) without someone reminding me.	☐	☐	☐
I know the potential side effects of my medication.	☐	☐	☐
I know what medications I should not take and what medications I am allergic to.	☐	☐	☐
I can talk about my emotions with others.	☐	☐	☐
I understand where to go for help and support for my mental health, like if I feel depressed or alone.	☐	☐	☐
I understand what accommodations are available to me for my education (e.g., 504 Plan) and what that means.	☐	☐	☐
I know about assistive equipment like canes, crutches, and walkers, that might help me in the future, and where to find them if I need to.	☐	☐	☐
I understand what XLH means for starting a family.	☐	☐	☐
I have discussed my ability to make my own healthcare decisions with my parents or caregivers.	☐	☐	☐
I understand how my rights will change when I turn 18.	☐	☐	☐
I understand that I will need to change my pediatric (childhood) doctor to an adult doctor as a young adult.	☐	☐	☐

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	I feel comfortable with this	I am still learning this	I don't know/ do this yet
I know my doctors' names and contact information.	☐	☐	☐
I make my own doctor appointments and keep track of them on my calendar.	☐	☐	☐
I feel comfortable speaking to my doctor and asking questions without my parent or caregiver.	☐	☐	☐
I am preparing to make decisions about my own care.	☐	☐	☐
I know what to do in case of a medical emergency.	☐	☐	☐
I know how to get to my doctor(s) office(s) without help.	☐	☐	☐
I come to my doctor appointments prepared with questions and ready to talk about my current issues or concerns.	☐	☐	☐
I know to ask for an explanation when I don't understand something my doctor says.	☐	☐	☐
I understand that I may need to talk to other types of doctors for my symptoms as I get older.	☐	☐	☐
I can provide a summary of my medical history (dates and types of surgeries, family history, allergies, and medications).	☐	☐	☐
I know how to fill out medical forms or where to get help doing so.	☐	☐	☐
I carry my important health information with me.	☐	☐	☐
I know my health insurance information (plan, coverage).	☐	☐	☐
I know that I will eventually be unable to continue under my parents'/caregiver's health insurance.	☐	☐	☐

How confident do you feel about your ability to manage your own healthcare, where 1 is lowest confidence and 10 is highest confidence?

1 (lowest)	2	3	4	5	6	7	8	9	10 (highest)

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When I think about my health in the future, I wonder about:

When I think about future, these are the things I want to do:

If you answered "I don't know / do this yet" to any of these questions, review them with your parents, other family members, or your doctor. You can also refer to the **Doctor Discussion Guide** in this toolkit to help you better discuss your health and symptoms during your doctor appointments.

NEXT STEPS

It is likely that when you turn 18 you will have to change to an adult doctor. You will also have to give your parents' permission to be in the room during your doctor visits and to hear your personal health information starting when you turn 18. You can begin or continue the transitions discussion with your doctor by asking:

- When do you recommend I start to see you alone for a portion of these appointments?
- Does your office have a template for a medical transition plan?
- Can we review our transition plan with you on a yearly basis?
- Can you recommend any adult specialists who might be knowledgeable and willing to care for patients? If not, are you aware of any primary care providers who specialize in caring for adults with complex health conditions such as XLH.
- Are you open to having a shared visit with the new adult doctor? If not, are you willing to contact the new adult doctor before our first visit? See the *Transfer of Medical Care Letter Template in this toolkit as a reference.*
- How will my medical records be sent to the new doctor?

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X-Linked Hypophosphatemia (XLH) Self-Care Assessment

Ages 19+

Fill out this form before visiting your doctor. Be as honest as you can. There are no right or wrong answers. This will help you understand your level of preparedness for taking over your own healthcare as an adult. If you answer "I don't know / do this yet" to any of these questions, review them with your doctor.

How important do you think it is to learn about managing your own healthcare, where 1 is least important and 10 is most important? Please circle one.

1 (least)	2	3	4	5	6	7	8	9	10 (most)

XLH Knowledge

	I feel comfortable with this	I am still learning this	I don't know/ do this yet
I know what XLH is and how it affects my body.	☐	☐	☐
I can explain the common symptoms of XLH to another person.	☐	☐	☐
I understand what "phosphate-wasting" means.	☐	☐	☐
I know what kind of XLH I have (spontaneous, inherited).	☐	☐	☐
I know how I was diagnosed with XLH.	☐	☐	☐
I know the tests I should receive and how often to monitor XLH.	☐	☐	☐
I know how to read and interpret my lab test results.	☐	☐	☐
I understand how XLH can be passed down in families.	☐	☐	☐
I can find my medical history (such as dates and types of surgeries, family history, allergies, and medications).	☐	☐	☐
I understand how XLH symptoms may change over time.	☐	☐	☐

This is how I explain how XLH is passed down in families:_____

_____

_____

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For more information about XLH, including symptoms, inheritance pattern, and management, read the "Learn more about X-linked hypophosphatemia (XLH)" educational brochure in this toolkit.

SELF-CARE	I feel comfortable with this	I am still learning this	I don't know/ do this yet
I know my current XLH treatment plan.	☐	☐	☐
I can explain my treatment plan to a doctor or nurse.	☐	☐	☐
I know the consequences of not taking care of XLH.	☐	☐	☐
I know my medication schedule and what to do if I miss a dose.	☐	☐	☐
I know how to recognize and talk to my doctor about new symptoms.	☐	☐	☐
I stick to my medication schedule without someone reminding me.	☐	☐	☐
I know the potential side effects of my medication.	☐	☐	☐
I know what medications I should not take and what medications I am allergic to.	☐	☐	☐
I can talk about my emotions with others.	☐	☐	☐
I understand where to go for help and support for my mental health, like if I feel depressed or alone.	☐	☐	☐
I understand what accommodations might be available to me at college or in the workplace.	☐	☐	☐
I know about assistive equipment that might help me in the future and where to find it.	☐	☐	☐
I understand what XLH means for my reproductive health and starting a family.	☐	☐	☐

This is where I can go for help and support:_____

_____

_____

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	I feel comfortable with this	I am still learning this	I don't know/ do this yet
I know my doctors' names and contact information.	☐	☐	☐
I know how frequently I should be seen, I make my own doctor appointments, and keep track of them on my calendar.	☐	☐	☐
I go to my appointments without my parent or caregiver.	☐	☐	☐
I am prepared to make decisions about my own care.	☐	☐	☐
I come to my doctor appointments prepared with questions and ready to talk about my current issues or concerns.	☐	☐	☐
I know to ask for an explanation when I don't understand something my doctor says.	☐	☐	☐
I understand that I may need to seek out specialty healthcare providers for new or different symptoms.	☐	☐	☐
I know what to do in case I have a medical emergency.	☐	☐	☐
I know how to fill and refill prescriptions.	☐	☐	☐
I can provide a summary of my medical history (such as dates and types of surgeries, family history, allergies, and medications).	☐	☐	☐
I know how to fill out medical forms or where to get help.	☐	☐	☐
I carry my important health information with me.	☐	☐	☐
I know my health insurance information (plan, coverage).	☐	☐	☐
I know when I may have to get my own health insurance and where to find it.	☐	☐	☐

This is where I would look for health insurance:_____

_____

_____

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How confident do you feel about your ability to manage your own healthcare, where 1 is lowest confidence and 10 is highest confidence?

1 (lowest)	2	3	4	5	6	7	8	9	10 (highest)

When I think about my health in the future, I wonder about:_____

_____

_____

_____

When I think about my future, these are the things I want to do: _____

_____

_____

_____

When preparing for your next doctor visit, keep the following questions in mind:

- When I meet with my doctor, what do I want to discuss?
- What do I want to get out of this appointment?
- What do I really need or want to know?

You can also refer to this toolkit's *Doctor Discussion Guide* for more advice on speaking with healthcare providers and getting more out of appointments.

NEXT STEPS

You can start or continue the pediatric to adult care transition discussion with your doctor by asking:

- Can you recommend any adult specialists who might be knowledgeable and willing to care for XLH patients? If not, are you aware of any primary care providers who specialize in caring for adults with complex health conditions such as XLH?
- Are you open to having a shared visit with the new adult doctor? If not, are you willing to contact the new adult doctor before our first visit? *See the Transfer of Medical Care Letter Template in this toolkit as a reference.*
- How will my medical records be sent to the new doctor?

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Sample Transfer of Medical Care Letter

(For Pediatric to Adult Care)

You can share this letter with your primary XLH doctor as an example of information that can be provided to your new doctor during the transition of care.

Download the full **XLH Transitions Toolkit** at **XLHlink.com**

January 12, 2020

Dr. John Smith
123 Main Street
Anytown, New York 01234

Dear Dr. Smith,

Jane Davis is a 21-year-old patient of our pediatric practice who will be transferring to your care. The expected date of transfer to your practice is February 20, 2020.

Jane's primary condition is **X-linked hypophosphatemia (XLH)** – a rare, hereditary, progressive, and lifelong condition characterized by chronic hypophosphatemia due to increased fibroblast growth factor (FGF23) activity. Increased FGF23 and resultant phosphate-wasting may lead to musculoskeletal defects, muscular dysfunction, and dental abscesses.[1,2] Adults with XLH can experience pain, restrictions in range of motion, arthritis, fractures, hearing loss, tinnitus, and gait disturbances, which may limit or restrict physical mobility.[3] The XLH Network (xlhnetwork.org) is the advocacy organization for this community and has additional information and resources on the condition.

Jane's secondary conditions are: pain, bowing of her legs, enthesopathy, history of fractures, and dental abscesses. Please find attached a list of current medications and specialists involved in her care. I am also attaching a medical history form from my records and her current treatment plan.

I have cared for Jane since age three and am familiar with her condition and medical history. I would be happy to provide any assistance to you during the transition to adult care. Please do not hesitate to contact me by phone or email if you would like to talk or if you have questions.

Thank you very much for your willingness to assume the care of this patient. We will follow up to confirm that your office has received this information.

Sincerely,

Dr. Mary Jones
(555) 123-4567
mjones@mail.com

SAMPLE

REFERENCES
1. Carpenter TO, Imel EA, Holm IA, Jan de Beur SM, Insogna KL. A clinician's guide to X-linked hypophosphatemia. J Bone Miner Res. 2011;26(7):1381-1388.
2. Ruppe MD. X-linked hypophosphatemia. In: Adam MP, Ardinger HH, Pagon RA, et al, eds. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2017. https://www.ncbi.nlm.nih.gov/books/NBK83985/. Accessed December 9, 2019.
3. Skrinar A, Dvorak-Ewell, M, Evins, A, et al. The lifelong impact of X-linked hypophosphatemia: Results from a burden of disease survey. J Endocr Soc. 2019;3(7):1321–1334.

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NOTES

Talking to Your Doctor about XLH

Download the full **XLH Transitions Toolkit** at **XLHlink.com**

Talking to your doctor isn't always easy. You may not have a lot of time during the appointment, and it can be hard to explain exactly what's wrong. If you're not prepared or unsure where to start, you may leave the appointment dissatisfied and without the answers you need. Filling out this Doctor Discussion Guide prior to your appointment can help you get the most from your doctor visits. It is designed to help you start the conversation about symptoms, challenges, questions, and concerns you may face on a daily basis as someone living with XLH.

Think about why you scheduled your doctor's appointment. Then ask yourself:

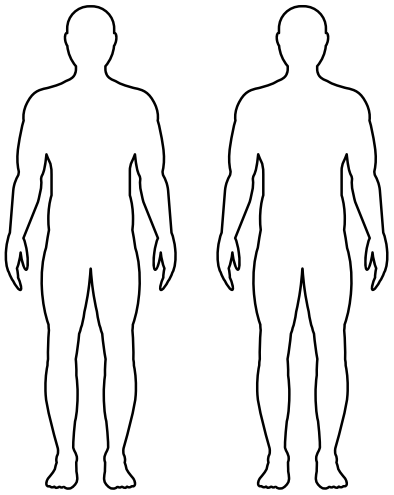
- When I meet with my doctor, what do I want to discuss?
- What do I want to get out of this appointment?
- What do I really need or want to know?

My XLH Experience

I am experiencing the following symptoms: (check all that apply)

- ☐ Bone pain
- ☐ Joint pain
- ☐ Muscle pain
- ☐ Pain in a specific area or body part(s): _____
- ☐ I have pain, but I'm not sure what kind
- ☐ Joint stiffness
- ☐ Limited range of motion
- ☐ General muscle weakness: _____
- ☐ Muscle weakness in a specific area: _____
- ☐ Tooth abscesses/dental issues
- ☐ Fatigue/exhaustion
- ☐ Hearing impairment/ringing in ears (tinnitus)
- ☐ Balance changes/feeling dizzy or off-balance (vertigo)

Mark on diagram where symptoms occur:



Front Back

I experience pain: (circle one)

Most of the day Some of the day During activities

My pain is typically worse: (circle one)

Mornings Mid-day Evenings During sleep It lasts all day

How would you rate your worst pain level recently? *Below is a typical pain scale doctors may use. Pain can be difficult to explain. It might be helpful to ask your doctor for a referral to a pain specialist.*

0	1	2	3	4	5	6	7	8	9	10
No Pain	Very Mild	Minor	Noticeable	Strong	Piercing	Intense	Very Intese	Horrible	Excruciating	Unimaginable

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Some of these symptoms are new or have recently changed: (provide an explanation below)

When I walk, I find myself needing support, or assistive equipment: (circle one)

Not at all Sometimes Most of the time All of the time

I typically use (e.g., a cane, crutches, the wall, hand rail):

I'm struggling with: (check all that apply)

- ☐ Doing daily activities
- ☐ Walking up and down stairs
- ☐ Sleeping
- ☐ Exercising
- ☐ Feeling depressed
- ☐ Feeling anxious
- ☐ Other: _____

I am concerned about: (check all that apply)

- ☐ Getting older
- ☐ The progression of my disease
- ☐ Starting a new job
- ☐ Going to college
- ☐ Starting a relationship
- ☐ Starting a family
- ☐ Getting health insurance
- ☐ What my health insurance covers
- ☐ How to find a specialist
- ☐ Other: _____

I would like more help with, or information about:

When I think about my health in the future, I am most concerned about:

ACTION PLAN

My doctor and I decided on these next steps:

Download additional copies of this form at XLHLink.com
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Addressing Your Mental Health

Download the full **XLH Transitions Toolkit** at **XLHlink.com**

The complexity of living with a rare condition, coupled with the physical aspects of XLH, can be overwhelming. Feeling different or not being able to do everything that your peers do can be frustrating and isolating. That's why it's important to pay attention to your mental health. There are many tools and supports that can assist you, including self-care, coping techniques, peer support, and reaching out for professional help. A mental health professional can be an important part of your care team.

Making Self-Care a Priority

Try to find a stress-reducing activity you enjoy and make it part of your regular routine, even when you're feeling well and life is not especially stressful. As you experience life's ups and downs, it's also important to know the **signs of depression** such as feeling sad, anxious, or "empty," having difficulty concentrating or sleeping, feeling restless and/or irritable, or feeling hopeless.[1]

These feelings are common and normal, but if they interfere with your daily activities, consider talking to your XLH specialist or primary care doctor. He/she may refer you to a mental health professional, such as a counselor, social worker, psychologist, or psychiatrist. After diagnosis, individual or family therapy, perhaps in combination with medication, can help you cope with symptoms of depression and anxiety.

Coping Techniques

Coping techniques are tools to help manage stress and anxiety. They can be **emotion-focused** or **solution-focused**.[2] You can find techniques you can use on your own, or you may work with a therapist who can support and educate you about new ways of coping and behavior.

Emotion-Focused Techniques

- Are used in situations that you can't (or don't want to) change
- Help people react less emotionally to stress

Solution-Focused Techniques

- Are used in situations you can change, or change the way you handle them
- Try to eliminate or change the source of stress

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Working Through Challenging Emotions

One tried and true emotion-focused technique is **deep breathing**.

1. Take a deep breath and hold it for five seconds before exhaling slowly.
2. Repeat this several times until you feel more peaceful.

You can also try using meditation to help detach yourself from stressful thoughts. There are several free and paid mobile apps that guide users through meditation.

You may want to try changing the way you view a stressful or negative situation. For example, if you are feeling discouraged by a setback at school or work, try to reframe it as an opportunity to improve and shine next time. You could also reframe the setback as an obstacle to overcome, which is something most people need to do at one time or another, and then visualize yourself overcoming it and feeling great about it in the future.

Another coping strategy is doing something relaxing, like taking a hot bath, spending time with your pet, crafting, doing light exercise, or connecting with friends or your support group.

Working Through Challenging Situations

Solution-focused skills encourage you to **think about your desired outcome, and then figure out the ways to get there.** You can think about the abilities and tools you've used successfully in the past to address a problem or consider a different way to approach it this time.[3] Try making a list of the things in your life that cause the greatest stress or biggest drain on your time and energy, and eliminate or change what you can.

Some situations, however, just can't be changed. You can't change the fact that you have XLH; nor can you lessen the amount of time it takes to get to class, or change the performance review process at work. But you can change how you handle these things. You can come up with creative ideas on your own, or work with a mental health professional to find solutions to a pressing problem.

Here are some examples of solution-focused coping techniques:

- When XLH makes it impractical to join your friends in their run training program, consider inviting them to go swimming or to join a tai chi group.
- Listen to podcasts or audiobooks or use driving time to do something else that makes you feel happy or productive – like catch up with friends or family on the phone. (Make sure you're using a hands-free device!)
- Talk to an occupational therapist about tips and tools you can use at home, work, or in a car to make those settings more usable, accessible, or comfortable for you.
- If you receive negative feedback about classwork or your performance at work, talk to your teacher or boss directly about what you can do differently. Remember that you have rights if you feel the negative feedback is connected to XLH or its physical limitations. *See the Navigating the Workplace section of this toolkit for more information.*

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MANAGING STIGMA AND BULLYING

Unfortunately, there can be still be stigma associated with disability and chronic illness. Often people make assumptions about what others with disabilities can or cannot do and **may not understand** the impact of a health condition like XLH. This misunderstanding may cause people to react negatively.

Here are some ways to combat bullying that occurs in-person at school or in the workplace:

- It's "old-but-good advice" – ignoring a bully is a good way to silence them. Bullies are looking for an emotional reaction. If you don't give them one, they may turn their attention elsewhere.
- Tell the bully to stop in a calm, non-emotional way – it can disarm them.
- There's safety in numbers. Sticking with a friend or group of friends can reduce the likelihood of being bullied.
- Educate them. Help reduce the likelihood of negative behavior by talking with your school or workplace about the best way to explain and educate your peers about what XLH is, and the impact it can have.

Cyberbullying – when someone repeatedly harasses, mistreats or mocks another person online or through text – is a serious problem for many people, whether they have a health condition or not. More than one-third of high school and middle school students report having been cyberbullied at some point in their lifetime, according to a national survey.[4]

Here are some strategies to combat cyberbullying:[5]

- Keep a record of what's happening. Take screenshots of the bully's attacks and hurtful posts whenever possible. When you're ready to report the bullying, it'll help to have documentation.
- Ask peers, friends, and mentors to intervene online by making positive posts to counteract a bully's negative or hurtful posts or shift the conversation in a more positive direction.
- Report it. Most social media platforms have clear policies for reporting cyberbullying and having offensive content removed. However, if you receive a threat of physical harm, report it directly to the police.

If you feel someone is bullying you, **ask for help.** When a student is being bullied at school, it may be necessary to speak to parents and/or school officials. When an employee is being discriminated against or exposed to bias at work, Human Resources may need to get involved. Even if you aren't ready to report, you should talk to someone you trust about what you're going through, so you don't have to face it alone. Also consider seeking support from a professional, such as a guidance counselor or mental health counselor.

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USING PEER SUPPORT

Connecting with others who share your experience can be helpful for people with rare conditions. This can be done in person, over the phone, or online through social media channels and email. Advocacy groups and forums like NORD (rarediseases.org), Global Genes (globalgenes.org), and Our Odyssey (ourodyssey.org) connect people with all kinds of conditions and offer many resources and opportunities to meet others.

At the same time, the day-to-day life of someone with XLH can be unique. In a study of one rare disease community, participants valued connecting with others who shared their diagnosis.[6] There are disease-specific advocacy groups like The XLH Network (xlhnetwork.org), which hosts several events, as well as a Facebook page to connect and build community.

DEALING WITH CRISIS

If you are thinking about suicide, or are worried about a friend or relative, call the suicide hotline for support. People are available to talk and help 24/7.

1-800-273-8255
suicidepreventionlifeline.org

RESOURCES

Global Genes
Navigating Emotions: A Guide for Teenagers with Rare Disease

National Institute of Mental Health
nimh.nih.gov

Anxiety and Depression Association of America
adaa.org

National Alliance on Mental Illness
nami.org

Ditch the Label
(International Anti-Bullying Organization)
us.ditchthelabel.org

Stop Bullying
stopbullying.gov

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HEADING TO COLLEGE

Download the full **XLH Transitions Toolkit** at **XLHlink.com**

Everyone's path is different, and the life changes (transitions) you need to prepare for are based on the specific path you're on. For those that are interested and able to pursue secondary education through college, it's important to be aware of the support that's available to you, and what you need to do to access it.

All public and private colleges that receive federal funding **must provide equal access** to education for students with disabilities and cannot discriminate against people with disabilities.[1] Common challenges with XLH, such as hearing problems or difficulty walking far distances, would be covered under these laws. Some key things to remember are:

- If you had an Individualized Education Plan (IEPs) in high school, it does not apply in college; however, your **504 plan** does if the college receives federal money.
- Students are responsible for locating the disabilities office, identifying themselves, requesting accommodations, and providing any required documentation (like a 504). The school is not required to help **unless you ask**.
- Typically, the student is responsible for **requesting accommodations *each year***.[2] An accommodation for XLH might be having more time to get across campus for class or permission to make up assignments missed during an absence due to a hospitalization or required medical appointment.

If you haven't communicated about your condition and documented or requested accommodations ahead of time, your professors aren't required to make exceptions later. Determine exactly what you may need, and when you make the request try to be as specific as possible.

UNDERSTANDING FERPA (FAMILY EDUCATIONAL RIGHTS AND PRIVACY ACT)

This law gives parents certain rights regarding disclosure of their children's educational records. It's important to note that these rights transfer from the parent to the student after the student turns 18. Therefore, colleges must have a student's permission to release any of their information, including health information, to their parents.
https://www2.ed.gov/policy/gen/guid/fpco/ferpa/index.html

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Also, keep in mind that you'll be juggling class, personal, and medication schedules. Use some of the resources in this toolkit to help you with self-care and to make sure you stay on track with your disease management plan.

"When I went to college, no one was telling me to take my medication anymore so I wasn't as adherent as I should have been."

ENTERING THE WORKFORCE

Many people choose to enter the workforce after high school rather than go to college. Resources like state-based Vocational Rehabilitation Agencies can help individuals with disabilities find and secure employment through a variety of support services. Once you enter a job or career, reference the **Navigating the Workplace** section of this toolkit for important information about communication with employers and co-workers about XLH.

RESOURCE

Find more information about secondary education and the workplace in the Global Genes toolkit:
Optimizing Your Potential as a Young Adult with a Rare Disease
globalgenes.org/wp-content/uploads/2015/06/GG_toolkit_Taking-Control-2_web.pdf

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2. Student disability accommodation policy. Maryland Institute College of Art; 2019. https://www.mica.edu/mica-dna/policies/disability-accommodation-policies/student-disability-accommodation-policy/. Accessed December 8, 2019.

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Navigating the Workplace

Download the full **XLH Transitions Toolkit** at **XLHlink.com**

As someone living with a chronic and potentially disabling condition, it's important to know that there are laws in place to protect you and guarantee certain rights in the workplace. Familiarizing yourself with your rights as you choose or navigate your career can help you succeed.

Knowing the Laws

The Americans with Disabilities Act (ADA) is a law that prohibits discrimination solely on the basis of disability in employment, public services, and accommodations.[1] This law also protects employees from retaliation when they invoke their rights under the ADA.

In order to be protected by the ADA, you must have a disability *and* be qualified to perform the functions or duties of a job, with or without reasonable accommodation.[2] In other words, you need to satisfy the employer's requirements regarding things like education, work experience, and skills. Here are some important things to know:

- The ADA defines a disability as a physical or mental impairment that substantially limits a major life activity, such as hearing, seeing, speaking, walking, breathing, or performing manual tasks.
- The ADA was updated in 2008 to make important changes to the definition of the term "disability." A disability is still defined as an impairment that substantially limits one or more life activities, but the list of life activities now includes reading, bending, major bodily functions, and more.[3]
- The 2008 amendment makes it easier to establish that you have a covered disability.
- You are also protected by the ADA if you have a history of a disability.

If XLH makes it difficult to perform some tasks at work, you may benefit from **requesting accommodations** to do your job successfully. Remember, your employer has to provide reasonable accommodation, but employers are only required to provide this for disabilities **of which they are aware**.[2] This means it is your responsibility to tell your employer if you need an accommodation.

Facing Challenges

Unfortunately, not all employers are knowledgeable, experienced, or fully compliant with all laws, which can make it difficult for employees with disabilities to get the accommodations they need. In more resistant workplaces, you may need to attend a legal proceeding (a hearing) with your employer and show documentation of your condition and need. It's important that you play an active role in advocating for yourself and helping your employer understand your rights and protections under the law. **Don't give up.**

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TALKING TO EMPLOYERS

It's hard to know when – and if – to tell an employer about your XLH. **You don't need to disclose your condition to your employer.** But choosing to may provide some benefits. The conversation will be more successful if you come prepared.

Think about what you want to say and how to say it. You may want to develop an "elevator pitch" — a simple description that people can quickly understand — so that you have a clear, concise way to explain your XLH.

Find an example in the Dating and Relationships section of the toolkit.

Before talking to your employer, write down any accommodations you might need. **Be as specific as possible.** Think about the following questions:

- Do you need to sit down at regular intervals?
- Do you need to stand at regular intervals, or, how long can you sit (for instance, for a long meeting or training)?
- Is there a maximum amount of weight you can lift?
- Would adaptive equipment, such as a lifting aid, hearing assistive technology, adjustable/ergonomic seating, or a footrest, be helpful?
- Do you need a step stool in the community kitchen?
- Do you need hearing caption phones?
- Are meeting rooms located within a comfortable walking distance?
- Do you need an alternate schedule to be able to receive treatments?
- Would you benefit from a closer parking spot?

It might help to talk to an expert. Consider contacting the Job Accommodation Network online at askjan.org or by phone at 800.526.7234.

Remember that **it's okay to ask** – not only is it your right, but most employers have provisions in their budget to cover the cost to make accommodations.

Jennifer, living with XLH

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UNDERSTANDING HEALTH INSURANCE

Getting your own health insurance as a young adult with XLH may seem overwhelming, but it's not impossible – provided you do a little research to understand your options. Many employers offer health insurance, and for those who don't, you have other options, including:

- **Marketplace Plans:** for people who can't get insurance from their employers or don't qualify for Medicare/Medicaid.
- **Medicare:** a government (federal) health insurance program for people age 65 or older and people under age 65 with certain disabilities and serious health conditions.
- **Medicaid:** a government (federal and state) health insurance program for people with a low income.

Once you turn 26, you may no longer be eligible for insurance coverage under your parents' plan(s).

If you're insured through a parent's **employer plan**, coverage will usually end right when you turn 26. If you're insured through a parent's **Marketplace** plan, coverage will last until December 31 of the year in which you turn 26.

In either case, be sure to plan ahead to avoid a lapse in coverage.

See Understanding Health Insurance for more information on these types of insurance.

If you are buying insurance as an individual in the marketplace or through an employer, always read the **"Summary of Benefits and Coverage (SBC)"** document. The SBC offers an easy-to-read explanation of the health plan's benefits and coverage in simple language.

During the hiring process (and once you've received a job offer), make sure to ask your prospective employer about the health insurance, dental insurance, prescription drug coverage, and any other benefits they may offer. Everyone should be aware of what's covered by their health insurance, but it's especially important for people with disabilities or complex health conditions, since you may have more specific healthcare needs.

INSURANCE FOR DIFFICULT HEALTH SITUATIONS

Disability insurance provides income in the event you're unable to work due to a disability. Short-term disability insurance policies provide a portion of your salary if you're unable to work for 3-to-6 months; long-term disability insurance applies after 6 or more months. However, both short- and long-term policies require you to be disabled for a certain length of time before you can qualify for the benefits. There are many types of plans with different rules, so it's best to do your homework and/or ask your human resources department.

The Family Medical Leave Act (FMLA) allows an employee to take up to 12 weeks off per year for medical emergencies, but this is unpaid time. The benefit is that your job is held and you keep the same insurance plan, and at the same cost. This law only applies to companies and employees that meet certain criteria.[4] For more information, read the U.S. Department of Labor's fact sheet on FMLA at: dol.gov/whd/regs/compliance/whdfs28.pdf. A few states also have Paid Family Leave policies for varying amounts of time. Research your state so you know what's available to you.

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RESOURCES

Accessible Parking and the Workplace
adata.org/publication/review-issues-relating-accessible-parking-and-workplace-under-americans-disabilities-act

ADA National Network
adata.org

Administration for Community Living
ACL.gov

"Gaining Independence" Toolkit
globalgenes.org/wp-content/uploads/2014/07/GG_toolkit_nine_20140725_web.pdf

Global Genes
globalgenes.org

Job Accommodation Network
askjan.org

National Disability Navigator Resource Collaborative
nationaldisabilitynavigator.org

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3. Notice Concerning The Americans With Disabilities Act (ADA) Amendments Act of 2008. U.S. Equal Employment Opportunity Commission; 2019. https://www.eeoc.gov/laws/statutes/adaaa_notice.cfm. Accessed September 26, 2019.
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Dating and Relationships

Download the full **XLH Transitions Toolkit** at **XLHlink.com**

If you are starting to date, are dating, or are in a relationship, you may be wondering when – and how – to talk to your partner about XLH. As someone living with a rare disease, you've likely told people close to you – family, friends, and possibly colleagues – about your condition. But it may feel different to talk to a romantic interest or partner. It's common to wonder how they'll react or whether they'll change their perception of you.

There's no right answer; it all depends on what makes you feel most comfortable. Take some time to think about your hopes and expectations for the relationship. Everyone is different, but these tips might help you decide what's best for you:

Talk to other people with XLH about their dating experiences or ask for their advice.

Decide in advance how much you want to share and how you want to tell your story, so you feel in control of the conversation.

Develop an "elevator pitch" — a clear and concise way to explain XLH that's easily understood and lasts about as long as an elevator ride.

SAMPLE ELEVATOR PITCH:

"I have a genetic, chronic condition called XLH. My body doesn't retain enough phosphorus, which affects my bones and makes them weak. I manage it through treatment, but it still affects me daily, and some days are worse than others. I can have trouble walking and getting around. I have a lot of pain, and I can be exhausted very easily so I have to be careful how I plan my days."

- Practice in front of the mirror until you feel comfortable.
- Like most serious conversations, it's best to have this one in person.
- Give people time to process the information and let them ask questions.
- Have information or resources ready, in case they would like to read or research more on their own.
- Recognize that they may be uncomfortable talking about it at first. Be patient.

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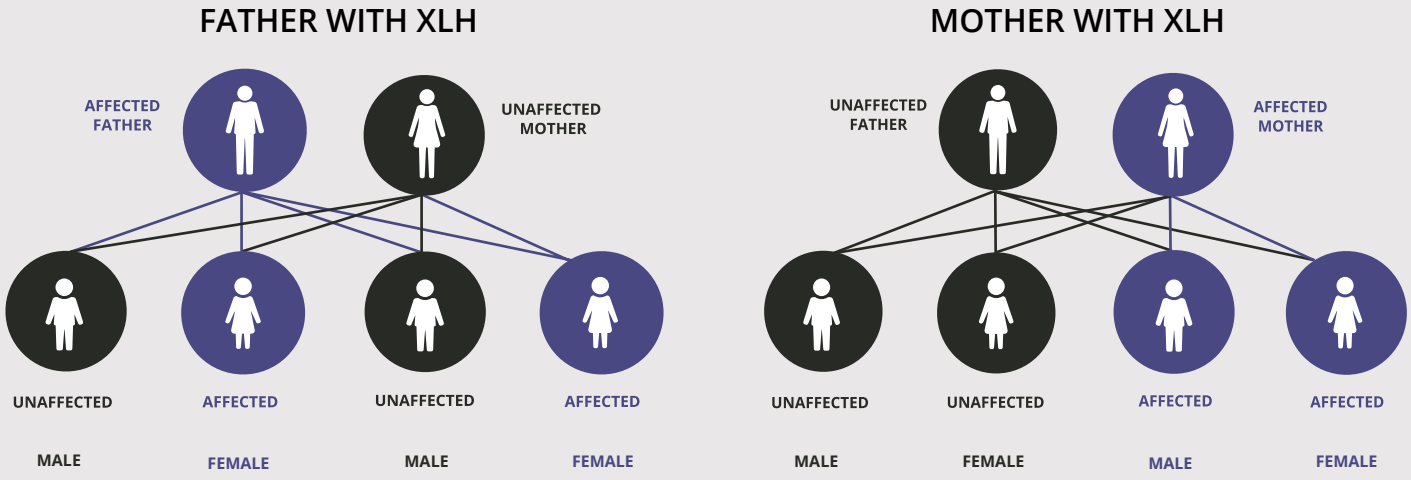
THINKING ABOUT STARTING A FAMILY

At some point, you may consider starting a family. This is an exciting milestone, but one that can present unique challenges for someone with XLH. **If you decide to have a child, it is a deeply personal decision,** and you may consider options like in vitro fertilization (IVF), sperm or egg donation, surrogacy, or adoption.

If you are a woman with XLH considering pregnancy, consult an obstetrician to discuss any potential health risks to you. Also, it's important to understand how XLH can be passed down in families. **XLH is an inherited disease**, which means that an affected parent may pass it down to his or her children. The "X" in XLH stands for "X-linked" because the disease results from a mutation on the X chromosome. Females have two X chromosomes and males have both an X and a Y chromosome.[1]

A male with XLH passes down his X chromosome to his female children and his Y chromosome to his male children, which means all of his female children will have XLH. If a female has XLH, each of her children, regardless of sex, has a 50% chance of having XLH. The chance of having an affected child is the same for each pregnancy regardless of what has happened in a previous pregnancy.

XLH INHERITANCE PATTERN



In some cases, people with XLH will have unaffected children. For example, a woman with XLH (whose partner does not have XLH) has a 50 percent chance of having a child (male or female) without XLH. A man with XLH (whose partner does not have XLH) could have male children without XLH.

Sometimes children can have XLH, even if neither parent is affected. This is called a **spontaneous case,** and it happens in about one-third (30%) of XLH cases.[1] Once a person has XLH, they can pass it down to their children, following the X-linked inheritance pattern.

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If you decide to have a child, consulting a **genetic counselor** can help you:

- Understand the XLH inheritance pattern.
- Learn about your reproductive options.
- Decide what's best for you.

Hospitals and health systems often have genetic counselors on staff. If you go to a hospital for your XLH care, ask your specialist to refer you. You may also find it helpful to talk to people with XLH who've had experience with pregnancy or have made a decision about family planning.

Elizabeth and her son, Simon, living with XLH

To learn more about genetic counseling, or to find a counselor near you, visit the National Society of Genetic Counselors at aboutgeneticcounselors.com.

XLH AND PREGNANCY

When you become pregnant, you'll likely see an obstetrician, midwife, or doula in addition to your primary care physician or XLH specialist. It's important to **talk to all your healthcare providers about how XLH affects your body** and to keep careful records that include your medical history, test results, all medications, and any symptoms you experience. It's also important for your healthcare providers to talk to each other to ensure coordination of care.

Be diligent about prenatal care, proper nutrition, and rest. Pregnancy can be more challenging for women with XLH because they may already have joint stiffness, pain, and fatigue. You will also need to discuss whether you can continue your XLH treatment and any other medications you take while pregnant, and possibly while breastfeeding.

To learn more about managing pregnancy with a rare condition see the Global Genes toolkit "Women with Rare Disease: The Reproductive Years."

Remember, people may have opinions, but whether or how you start a family is your choice, and you should choose what is best for you.

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NOTES

Bringing people and ideas together.

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