PROJECT ALIVE

Planning Ahead: Options & Considerations for Your Family

This document is meant to provide clear, balanced information for families expecting a baby boy when the mother is a known carrier of Hunter syndrome (MPS II). Every family's situation, values, and comfort level are different. There is no right or wrong choice — only what feels best for you.

Understanding MPS II Variability

- MPS II can present very differently from one individual to another.
- Many boys have no signs at birth or in infancy.
- Symptoms may not appear until toddler or preschool years, as was the case for some individuals.
- A normal pregnancy and delivery are very common, even for children who later receive a diagnosis.

Option 1: Amniocentesis During Pregnancy

What it is:
Amniocentesis is a prenatal diagnostic test usually performed between 15–20 weeks of pregnancy. It involves sampling amniotic fluid to test fetal DNA.

Potential Pros

- Can provide a definitive diagnosis before birth
- Allows families time to:
    - Emotionally prepare
    - Learn about MPS II and current treatments
    - Identify specialty care teams early
    - Plan delivery at or near a specialty center if desired
- Can reduce uncertainty for some families

Potential Cons

- Small risk of miscarriage (often quoted around 0.1–0.3%, depending on provider)
- Can increase anxiety during pregnancy
- Results may create emotional stress without changing immediate medical care
- Some families feel that knowing during pregnancy does not change how they will love or parent their child

Option 2: Choosing Not to Do Amniocentesis

Many families choose not to pursue prenatal diagnostic testing, and this is a completely valid option.

Potential Pros

- Avoids the medical risks associated with invasive testing
- Allows pregnancy to progress without added procedural stress
- Gives families time to focus on pregnancy and bonding
- Some families prefer to wait until after birth for clarity

Potential Considerations

- Diagnosis, if present, would occur after birth
- Families may still want a plan for:
    - Early monitoring
    - Pediatric follow-up
    - Genetic and metabolic evaluations

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Post-Birth Options (Regardless of Prenatal Testing Choice)

Whether or not prenatal testing is done, families can:

- Connect with a genetic counselor
- Establish care with a metabolic or genetics specialist
- Discuss early testing after birth, if desired
- Monitor development closely with a pediatrician
- Learn about current research, treatments, and clinical trials

Early diagnosis after birth can still allow for timely care and intervention.

Emotional Considerations

- It is normal to feel overwhelmed, scared, hopeful, or uncertain — sometimes all at once.
- Decisions may change as pregnancy progresses, and that's okay.
- Support from professionals, other families, and advocacy organizations can make a meaningful difference.

Support Available Through Project Alive

Project Alive can help connect families with:

- Genetic counselors and specialists
- Educational resources and research updates
- Other families who have navigated similar decisions
- Ongoing emotional and community support

You do not have to make these decisions alone.

This document is intended for educational purposes and does not replace medical advice. Families are encouraged to discuss all options with their healthcare providers.

Clinical Trials:

MPS II Active Clinical Trials | Project Alive

Mucopolysaccharidosis type 2 | Prenatal Enzyme Replacement for Lysosomal Diseases | PEARL Trial

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Questions to Ask Your Provider

Families may find it helpful to bring some of the following questions to appointments with their OB, genetic counselor, or specialist:

- What testing options are available to us during pregnancy and after birth?
- What are the risks and benefits of amniocentesis in our specific situation?
- If we choose not to do prenatal testing, what monitoring or testing can be done after birth?
- How early can MPS II be diagnosed after delivery?
- Should we meet with a genetic counselor or metabolic specialist now?
- If a diagnosis is confirmed, what early care or treatments are available?
- Are there specialty centers or providers you recommend for MPS II care?

Writing down questions ahead of time and bringing a support person to appointments can be helpful during these conversations.

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Decision-Making Checklist

Families may find it helpful to reflect on the following questions as they consider their options:

☐ Do we feel more comfortable having as much information as possible before birth, or waiting until after delivery?

☐ How do we personally feel about the medical risks associated with invasive testing?

☐ Would a prenatal diagnosis change how we prepare emotionally or practically?

☐ Would knowing before birth help reduce uncertainty for us, or increase anxiety?

☐ Do we have access to a genetic counselor or specialist we trust?

☐ What kind of support system do we have during pregnancy and after birth?

There is no requirement to check every box or answer every question right away. Many families revisit these questions over time.

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Timeline: What Planning May Look Like Over Time

This timeline is meant to show possible touchpoints, not a required path. Families may move through these stages differently.

Pregnancy

- Carrier status confirmed
- Prenatal counseling with an OB and/or genetic counselor
- Decision-making around prenatal testing (such as amniocentesis)
- Learning about MPS II and available resources
- Emotional support and planning at your own pace

Birth & Newborn Period

- Normal delivery in most cases
- Discussion of newborn or early diagnostic testing, if desired
- Establishing care with a pediatrician familiar with your family history
- Connecting with a genetics or metabolic specialist if indicated

Infancy & Early Childhood

- Monitoring growth and developmental milestones
- Ongoing pediatric care and follow-up
- Diagnostic testing if concerns arise or if previously planned
- Early intervention services if needed
- Continued education, support, and community connection

This timeline is flexible and can be adapted to fit your family's needs and comfort level.
