

# Could My Child Have a Rare Genetic Condition?

A page to bring to your pediatrician or family doctor, to help start the conversation about genetic testing.

**How to use this page.** Fill in the checkboxes below that apply to your child, then bring this sheet to your next appointment. You do not need to know the answers or use any medical terms. This page simply helps you ask the questions that matter.

## 1. What I have noticed about my child

- |                                                                              |                                                                                      |
|------------------------------------------------------------------------------|--------------------------------------------------------------------------------------|
| <input type="checkbox"/> Slower development than other children the same age | <input type="checkbox"/> Delayed, unclear, or nasal-sounding speech                  |
| <input type="checkbox"/> Difficulty with learning or schoolwork              | <input type="checkbox"/> An autism diagnosis that does not explain everything        |
| <input type="checkbox"/> Low muscle tone, or was a floppy baby               | <input type="checkbox"/> Trouble with attention or hyperactivity                     |
| <input type="checkbox"/> A tall, slender build with long fingers or limbs    | <input type="checkbox"/> Seizures                                                    |
| <input type="checkbox"/> Curvature of the spine (scoliosis)                  | <input type="checkbox"/> Other relatives, especially boys, with similar difficulties |

## 2. Questions I would like to ask

**Could there be a genetic cause for my child's symptoms?** Many conditions that look like autism or general developmental delay turn out to have a specific genetic cause that can be identified with a test.

**Should my child have genetic testing, and which test?** A chromosomal microarray is often the first test. If that does not find an answer, exome or genome sequencing looks more closely and is what identifies most rare single-gene conditions.

**Can you refer us to a genetics clinic or genetic counselor?** A genetics specialist can review our family history, decide which test is right, and explain what the results mean for our whole family.

**Would knowing the cause change my child's care?** A diagnosis can guide which specialists and therapies to pursue, support school accommodations, and connect our family with others who have the same condition.

**About the cost.** Texas Medicaid and most private insurance plans cover comprehensive genetic testing, such as exome sequencing, for children with unexplained developmental delay or intellectual disability. If cost is a worry, please ask whether my child qualifies for this coverage before we rule testing out.

## 3. Notes from our appointment

Tests ordered or discussed

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Referral made to

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### Why we made this page

The UPF3B Project is a family-founded initiative focused on UPF3B-related disorder, a rare X-linked genetic condition once known as Lujan-Fryns syndrome. It mainly affects boys and can look like autism or unexplained intellectual disability. Families often wait years for an answer. We built this page so that fewer of them have to.

Next appointment

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