

Questions to ask before your child's genetic test

Not all tests and laboratories are the same

1. Is rare disease the laboratory's primary area of expertise?



Some laboratories focus specifically on rare disease, while others test for many different types of conditions.

2. How long has the laboratory performed rare disease genetic testing, and how many rare disease families has it tested?



Rare disease experience can help labs recognize similar conditions, genetic findings, and ancestral backgrounds.

3. If my child's results are complex, does the laboratory have the right experts to interpret them?



Specialized expertise can help labs understand difficult genetic findings and what they may mean for your family.

4. Will we have access to the laboratory's experts if we have questions before or after testing?



Genetics experts can help explain results and appropriate next steps. Access to support varies by laboratory.



The lab behind your child's genetic test matters.

Laboratory-specific quality factors influence genetic test results

	Laboratory A Limited rare disease experience, expertise, and support	Laboratory B Extensive rare disease experience, expertise, and support
Rare disease experience	Limited experience testing families impacted by rare disease.	Extensive experience evaluating complex rare disease cases and genetic changes to help find answers for families.
Diverse, representative data	Less representative population data can lead to greater uncertainty when interpreting results.	Broader representation supports more accurate, confident, and equitable interpretation across patient populations.
Expertise & support	Less access to rare disease experts may make complex results harder to interpret and act on.	In-house experts help interpret complex findings and support families and clinicians in understanding what results may mean for care.

What could this mean for your child?

Differences in laboratory experience, data, and expertise may affect the likelihood of finding an answer and understanding next steps for care.