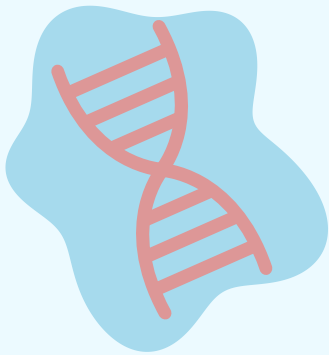


*A Family's Guide to*  
**Neurodevelopmental  
Disorder Genetics**



## What are neurodevelopmental disorders (NDDs)?



- **All children develop differently** according to their own unique bodies, environment, experiences, and more.
- A **neurodevelopmental disorder** (NDD) may be diagnosed when a child shows developmental differences that impact how they function. Children with NDDs may need **extra support** to live their best lives.

1 in 6

children in the US have a developmental difference

- Examples of NDDs include **developmental delay** (delays in meeting developmental milestones), **intellectual disability** (difficulties with mental functions such as learning and communication), and **autism spectrum disorder** (repetitive behaviors and challenges with social communication).



- Neurodevelopmental differences can be caused by a variety of factors. The largest known cause is **genetic differences**, or differences in the body's instruction manuals on how to grow and function. These differences can occur for the first time in a child and may not be found in other family members.

## How can a genetic counselor help?



A **genetic counselor** can talk to you about your child's medical and family history. Based on this information, they may offer **genetic testing** to search for a genetic cause of your child's NDD.

### Potential Benefits of Genetic Testing

Identifying the genetic cause of an NDD can:

- **Explain why** your child has developmental differences.
- Provide a **specific diagnosis** for your child and help us better understand what to expect and how to care for them.
- Increase **access to support** services and communities of children/families with the same diagnosis.
- Inform estimates of the chance that **future children** would experience similar symptoms.

### Potential Drawbacks of Genetic Testing

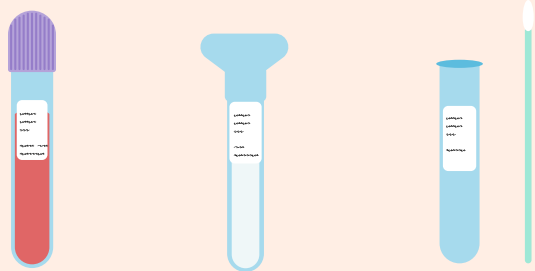
- There is **no guarantee** that genetic testing will identify a genetic cause of your child's NDD or that an identified cause will inform their medical care.
- Genetic testing may find genetic differences that are **unexpected or not well understood**.
- This information may be used by **life, long-term care, and disability insurance** companies when making policy decisions (but NOT by health insurance companies).

## What kind of genetic testing may be recommended?



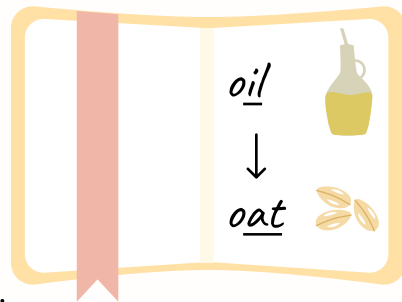
Genetic testing is typically carried out in **steps**, starting with one test and then following with additional tests if results are uninformative. For example: single gene testing for a suspected condition → microarray → whole exome sequencing.

Genetic testing typically requires a blood draw, saliva collection, or cheek swab.



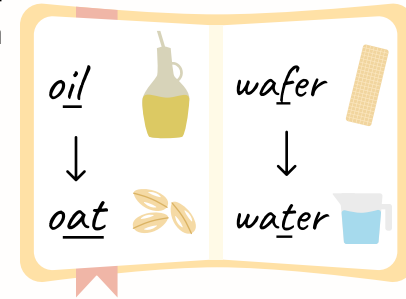
### Single-Gene Testing

- Imagine our genetic information is like a recipe book, with ingredients (*genes*) providing instructions for making baked goods (*proteins*). Single-gene testing is like looking for **misspellings** or other differences in **one** ingredient (*gene*).
- This test may be ordered if a specific genetic condition is strongly suspected. Testing for a condition called “Fragile X syndrome” is often recommended for children with NDDs because this is the most common genetic cause of intellectual differences and autism.



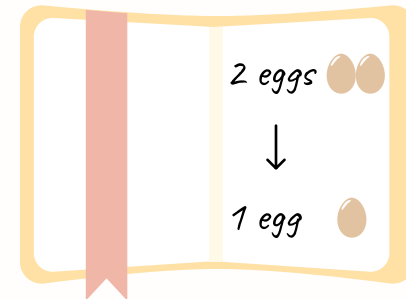
### Multi-Gene Testing

- Multi-gene testing is like looking for **misspellings** or other differences in **multiple** ingredients.
- This test may be ordered when a child has symptoms that fit multiple genetic conditions, or when a condition that is suspected can be caused by differences in multiple genes.



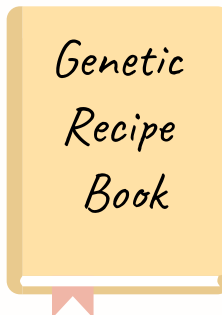
### Microarray

- A microarray is like looking for **extra** or **missing** ingredients.
- This test is often ordered for children with NDDs, as many developmental differences are caused by extra or missing genetic information.



### Whole Exome Sequencing

- Whole exome sequencing is like looking for **misspellings** or other differences in **all** key ingredients (*gene exons*, the parts of genes that are directly used to make proteins).
- This is the most **comprehensive** of these tests, however, insurance companies may not cover this test unless other tests are carried out first.



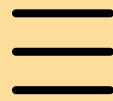
## How can I get connected with a genetic counselor?



- Your child's provider can connect you with a certified genetic counselor by faxing a referral to Genetic Support Foundation at 844-813-3892.
- You can also schedule a secure video appointment with a certified genetic counselor yourself by visiting [geneticsupportfoundation.org/appointments](https://geneticsupportfoundation.org/appointments).



## Summary of main points



- Neurodevelopmental differences occur for a variety of reasons, but the largest known cause is **genetic differences**.
- A **genetic counselor** can talk to you about your child's medical and family history. Based on this information, they may offer **genetic testing**.
- Genetic testing is typically carried out in **steps** and requires a blood draw, saliva collection, or cheek swab.
- Identifying a genetic cause of your child's symptoms can provide a number of potential **benefits**. For example, it can provide a specific diagnosis, inform treatment and management recommendations, and inform estimates of recurrence in future children.

## Support Resources

- You are not alone in navigating a new diagnosis. There are many national and community-specific resources to help support you, your child, and your family.
- For child care support resources, visit [www.childcare.gov](https://www.childcare.gov).
- For tips on talking to your child, family, and friends about diagnoses such as autism, visit [www.childmind.org/article/sharing-an-autism-diagnosis-with-family-and-friends](https://www.childmind.org/article/sharing-an-autism-diagnosis-with-family-and-friends).
- For answers and support from other parents of children with neurodevelopment differences, find a family-led parent center in your state by visiting:
  - Family to Family Health Information Centers: [www.familyvoices.org](https://www.familyvoices.org)
  - Parent Training and Information Centers: [www.parentcenterhub.org](https://www.parentcenterhub.org)

## Connect with Genetic Support Foundation



Call 1-800-364-1641



Visit [www.geneticsupportfoundation.org](https://www.geneticsupportfoundation.org)