# Pediatric Genomic Testing Playbook

Implementing Genomic Testing in Pediatric Primary Care

BioLogic Pharma Solutions | Version 2.1 | 2026

GeneDx Cure NDD BioLogic Pharma Solutions

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# Sponsors

This initiative is made possible through the support of organizations committed to advancing equitable access to genetic testing in pediatrics. Their contributions enable education, workflow tools, and implementation resources for clinical teams.

Questions about sponsor programs, services, or patient resources should be directed to the respective sponsor using the links below. Questions about this playbook, clinical workflow tools, or implementation support should be directed to BioLogic Pharma Solutions. This playbook is independently developed; sponsors provide financial support only and do not influence clinical guidance or content.

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## Sponsors

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# Objectives

This playbook helps pediatric practices implement guideline-aligned genomic testing for children with developmental delay, autism, epilepsy, and other suspected genetic disorders. The goal is to shorten the diagnostic journey, improve clinical management, and ensure equitable access to genetic services.

This playbook is intended for pediatric physicians.

The framework integrates genomic testing into routine pediatric visits and existing EHR workflows.

# Practice Implementation Timeline

*A 12-week phased rollout to integrate genomic testing into your pediatric practice.*

**Weeks 1-2**

**Breakfast and Learns**
Materials dissemination, 30-minute sit-in with HCP and ordering nurse

**Week 3-4**

**HCP Orders**
HCPs place orders with support as needed for workflow and prior authorization questions

**Week 5-7**

**Results Review Sessions**
1-hour sit-in with results over 3, 20-minute sessions

**Week 8-9**

**Site Operating Semi-Independently**
Site manages ordering and workflow independently with minimal external support

**Week 10-12**

**Program & Outcomes Assessment**
Assess overall program and results of patients tested

# Why Implement Genomic Testing

**~17%** of children in the U.S. are affected by neurodevelopmental disorders. Many have identifiable genetic causes through chromosomal microarray or exome sequencing.

## Benefits of Genetic Diagnosis

- **Identifies the underlying cause of a child's condition**
  Moves from symptom-based labeling to a confirmed diagnosis by identifying pathogenic variants linked to specific disorders, reducing uncertainty and shortening the diagnostic journey.

- **Guides medical management, surveillance, and prognosis**
  Informs condition-specific care, monitoring, and risk mitigation while providing insight into the expected clinical course to support clinical decisions and family expectations.

- **Supports family planning recurrence risk counseling**
  When a variant is identified as de novo or inherited, families can receive counseling that addresses parental concerns or self-blame and outlines options such as carrier testing, and parental testing, supported by condition-specific guidance in **GeneReviews**®.

- **Enables participation in clinical trials**
  Meets eligibility requirements for several studies, connecting families to research and investigational therapies.

### Guideline References

**American Academy of Pediatrics:** publications.aap.org/pediatrics/article/156/1/e2025072219/202230/

**American College of Medical Genetics:** https://www.acmg.net

**GeneReviews Clinical Reference:** https://www.ncbi.nlm.nih.gov/books/NBK1116/

# Pediatric Genetic Testing Decision Tree

*Desk Reference for Pediatricians — Print for exam rooms or clinician workspaces*

**Concern for GDD**

↓

- **Obtain patient and family history**
- **Additional phenotyping as indicated (including MRI brain, hearing, and vision evaluations)**

↓

**Screen for:**

- **Global Developmental Delay (GDD) or Regression (2+ domains)**
- **Seizures**
- **Dysmorphic Features**
- **Congenital Anomalies**
- **Unexplained Hypotonia**
- **Strong Family History**

**GDD Domains:**

- Motor Skills
- Speech and Language
- Cognitive
- Social and Emotional
- Adaptive Functioning

↓ **If 2+ present**

**<u>Tier 1 Testing</u>**
**Genome Sequencing**
**OR**
**Exome Sequencing + Chromosomal Microarray**
**(sequential or concurrent)**

↓ **Continue if no diagnosis obtained**

**Refer to genetics**

## Key References Databases

**ClinGen Actionability Database:** https://actionability.clinicalgenome.org

**GeneReviews:** https://www.ncbi.nlm.nih.gov/books/NBK1116/

# Seven-Minute Pre-Test Counseling Script Card

*Quick Reference Tool — Print for use in exam rooms*

### MIN 1 Reason for Testing

*"We are recommending genetic testing because it may help identify the cause of your child's symptoms."*

### MIN 2 What the Test Looks For

*"The test analyzes genes that may be related to developmental or neurologic conditions."*

### MIN 3 Possible Results

*"Three possible outcomes: a diagnosis explaining the condition, no findings, or uncertain findings."*

### MIN 4 Variants of Uncertain Significance

*"Sometimes we find changes in genes whose meaning we do not yet understand."*

### MIN 5 Secondary Findings

*"Occasionally testing may reveal unrelated health risks that could affect future medical care."*

### MIN 6 Family Implications

*"Results may provide useful information for other family members."*

### MIN 7 Insurance and Consent

*"Most laboratories help with insurance authorization. With your permission we will proceed."*

## Family Resources

**MedlinePlus Genetics:** https://medlineplus.gov/genetics/

**National Society of Genetic Counselors:** https://www.nsgc.org

# Ordering Genetic Testing

Many pediatric practices partner with clinical genetics laboratories that support ordering and insurance navigation.

## Genetic Laboratories

### GeneDx

https://www.genedx.com/providers/

## Requisition Forms

**ExomeDx™ and Reanalysis Test Requisition Form:** https://cdn.prod.website-files.com/69179b7c79bdf6bd80578e72/6984bd7b3b67f5cbd44e97ec_67897-Exome-TRF-v260205.pdf

**GenomeDx™ and Reanalysis Test Requisition Form:** https://cdn.prod.website-files.com/69179b7c79bdf6bd80578e72/698f87145c12bfb179ce7e85_67827-Genome-TRF-v260202.pdf

# ICD-10 Coding and Documentation

Use the following codes to support documentation and prior authorization for genetic testing orders.

| Category | ICD-10 Code Description | ICD-10 Code(s) |
|---|---|---|
| Autism | Autism Spectrum Disorder | F84.0 |
| Developmental Delay | Global Developmental Delay | F88 |
| Lack of expected normal physiological development in childhood | Developmental Delay, Unspecified | R62.50 |
| Epilepsy | Epilepsy | G40 |
| Congenital malformations | Multiple Congenital Anomalies | Q89.7 |
| Intellectual disabilities | Profound Intellectual Disabilities | F73 |
| | Severe Intellectual Disabilities | F72 |
| | Moderate Intellectual Disabilities | F71 |
| Family history | Family history of intellectual disabilities | Z81.0 |
| | Family history of other mental and behavioral disorders | Z81.8 |
| | Family history of epilepsy and other diseases of the nervous system | Z82.0 |
| | Family history of carrier of genetic disease | Z84.81 |
| | Family history of other congenital malformations, deformations and chromosomal abnormalities | Z82.79 |

## Coding Guidance

**CDC ICD-10-CM Resource Tool:** icd10cmtool.cdc.gov

# Prior Authorization Strategy

## Key Elements for a Successful PA Request

**1** **Clear Clinical Indication**
State the suspected diagnosis and why genomic testing is medically necessary for this patient, linking the request directly to presenting signs, symptoms, and clinical findings.

**2** **Guideline Citation**
Reference recognized clinical guidelines or authoritative sources that support genomic testing for the patient's presentation to demonstrate that the request aligns with established standards of care.

**3** **Documentation of Symptoms**
Provide concise but thorough clinical notes that capture the patient's phenotype, prior evaluations, and relevant history to substantiate the need for testing.

## Sample Justification Language

*Genetic testing is recommended for individuals with unexplained developmental delay and suspected genetic etiology. The American Academy of Pediatrics (AAP) and the American College of Medical Genetics (ACMG) guidelines support exome and genome sequencing as first-tier genetic tests due to their higher diagnostic yield compared with chromosomal microarray alone. A confirmed genetic diagnosis can inform medical management, guide family planning, provide prognosis insights, and enable access to clinical trials and emerging targeted therapies.*

**Payer Policy Reference:** Concert Genetics — tracks coverage policies across U.S. payers

# Managing Test Results

## Positive Result

**What it means:** A pathogenic or likely pathogenic variant has been identified that explains the child's condition.

**Recommended action:** Discuss diagnosis with family, refer to genetics specialist, initiate condition-specific management and surveillance.

## Negative Result

**What it means:** No clinically significant findings identified in the genes evaluated.

**Recommended action:** Review clinical diagnosis, consider whether broader testing (exome) is indicated, offer reanalysis in 18–24 months.

## Variant of Uncertain Significance (VUS)

**What it means:** A variant has been identified but its clinical significance is currently unclear.

**Recommended action:** Do not use for clinical decision-making. Inform family. Variant may be reclassified over time; plan for recontact.

## Family Resources

**ClinVar:** https://www.ncbi.nlm.nih.gov/clinvar/

**ClinGen:** https://clinicalgenome.org

# Follow-Up and Reanalysis

## Recommended Workflow

**1**

### Review Results with Family

Explain the findings in clear terms, including what was identified, what remains uncertain, and how results affect care and next steps.

**2**

### Refer to Genetics Specialist

Coordinate follow-up with a genetics professional for detailed interpretation, counseling, and condition-specific management guidance.

**3**

### Offer Family Testing

Discuss targeted testing for parents or relatives when appropriate to clarify inheritance, variant significance, and recurrence risk.

**4**

### Schedule Reanalysis

Plan periodic reanalysis of genomic data as knowledge evolves, which may yield new insights or updated variant interpretations over time.

**GeneDx**
https://www.genedx.com/providers/

### Rare Disease Resources

**NORD:** https://rarediseases.org

**Global Genes:** https://globalgenes.org

# Practice Performance Metrics

Track these metrics to monitor program effectiveness, operational efficiency, and health equity.

## Clinical Metrics

**Patient Management Change (%)**
Proportion of patients showing clinical improvement or change in management attributable to test results.

**Diagnostic Yield (%)**
Proportion of tests that return a pathogenic or likely pathogenic finding including variant data.

**Age of Testing (#)**
Patient age at time of testing.

**Indications for Testing**
Count of patients presenting with a documented clinical phenotype.

## Operational Metrics

**Eligible Patients Tested (%)**
Proportion of eligible patients who receive genomic testing, indicating program uptake.

**Test Selection**
Which test was ordered (genome, exome, trio)

**Tele-genetics utilization (%)**
Adoption of remote genetic counseling or specialist services within the testing workflow.

**Program Insights**
Comments on overall program performance, including observed bottlenecks, operational gaps, opportunities to improve.

## Equity Metrics

**Testing Access for Underserved Populations (%)**
Distribution of genomic testing across defined underserved groups.

**Insurance Approval Rate (%)**
Payer success rate for genomic testing requests.

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# GeneDx billing

## What you and your patients can expect.

GeneDx accepts all commercial insurance, Medicaid, Medicare, and Tricare. We also offer direct billing for institutions and self-pay options.

Eligible patients may also benefit from interest-free payment plans and our Patient Access Solutions, including our Financial Assistance Program, which aim to increase the affordability of testing.

## Importance of submitting complete supporting documentation

For all insurance types, the following information should be included at the time of order placement to increase the chances of insurance approval and reduce back and forth for you and your patients.

- Prior authorization (PA) approval documentation, if obtained in advance
  - In most cases GeneDx can submit the authorization request to insurance on behalf of the ordering provider through our third-party vendor, careviso.
- Insurance-required prior authorization form, if applicable
- Supporting documentation that demonstrates why the test is medically necessary, including:
  - Documentation of why the test ordered (e.g., exome or genome) is the most appropriate test for your patient based on their personal and family history (e.g., clinical notes and previous test results)
  - Documentation of how test results could potentially impact management, including explicit details and examples (e.g., clinical notes)
  - If applicable, documentation that genetic counseling was performed (e.g., separate genetic counseling consult note or documentation of counseling by the ordering provider)
- All relevant diagnosis codes (ICD-10)
- A copy of the patient's primary insurance card (front and back)

To access downloadable templated letters to help establish medical necessity for exome and genome, please visit **GeneDx.com/LMN.**

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## Requirements for non-commercial insurance providers

Additional documentation is required by some insurance providers, including Medicaid, Tricare, and Medicare.

### Medicaid

GeneDx accepts all Medicaid plans. In certain states without explicit Medicaid coverage for exome sequencing, patients may be able to obtain coverage for testing through the Early and Periodic Screening, Diagnostic and Treatment (EPSDT) program. Please talk to your GeneDx Regional Account Executive to understand if this is an option in your state and to access any required forms.

### Tricare

GeneDx accepts all Tricare plans. If a prior authorization is not obtained in advance of placing the order, the following should be included at the time of the test order:

- For Tricare members with Prime and U.S. Family Health plan: Tricare PCM referral form
- If complete documentation supporting medical necessity is not included, then the appropriate Tricare Laboratory Developed Test (LDT) attestation form must be provided. To access these forms, please visit **GeneDx.com/tricare-forms**.

### Medicare

For exome orders, patients with traditional Medicare will be required to sign an Advanced Beneficiary Notice of Non-Coverage (ABN), as Medicare does not have an explicit exome coverage policy. If an ABN is required, GeneDx will reach out to the patient after receiving the order.

## Submitting complete diagnosis codes

For test orders billed to a patient's insurance, submission of all relevant ICD-10 codes is critical to support medical necessity.

- The primary ICD-10 code should reflect the primary reason for ordering the test.
- Additional relevant ICD-10 codes should be submitted.
- For exome and genome testing, more than one diagnosis code is often needed to obtain insurance approval.

To access a list of frequently used ICD-10 codes in pediatric neurology, please visit **GeneDx.com/ICD10**.

* GeneDx reserves the right to modify which health plans are held for prior authorization determination at any time.
† If a test is canceled for billing reasons, it may be reactivated by contacting us at Support@GeneDx.com.

The information described here applies to orders placed within the United States.

## Prior authorizations

A prior authorization (PA) is almost always required by insurance for genetic testing.

Certain insurance providers, such as Aetna, require specific PA forms. Ask your Regional Account Executive or Client Success Manager for these forms.

! *Testing for outpatient (non-rapid) genome orders will be held until we receive an approved PA from the insurance company. The ordering provider may select an alternative option at the time of ordering in the event the PA is denied.*

! *For patients with an insurance requirement for pre-test genetic counseling, we can connect your patients with a third-party genetic counseling service. To request this service, please submit the genetic counseling referral form at* ***GeneDx.com/gc-referral****.*

## PA support with careviso

GeneDx encourages ordering providers to obtain a PA in advance of placing an order.

In most cases, GeneDx can also submit the authorization request to insurance on behalf of the ordering provider through our third-party vendor, careviso. Careviso works directly with insurance providers to obtain PAs and submits the approval and/or denial information to GeneDx.

However, some payors, such as UHC and some Blues plans, require ordering providers to complete enrollment with careviso in order to utilize their services. In these instances, if an ordering provider is not registered with careviso, PA approval documentation must be submitted with the order or testing may remain on hold until documentation is received.

→ To get started with careviso, please visit **careviso.com/enrollment.**

## Patient Access Solutions

GeneDx's Patient Access Solutions offer a suite of tools and programs to help address patients' final cost of testing. Below is more information on two of these solutions. To learn more, please visit **GeneDx.com/patient-access-solutions.**

### Financial Assistance Program

Financial assistance can reduce the amount owed by the patient for testing billed through insurance.

## Patient Access Solutions *(continued)*

Patients can fill out an application for the Financial Assistance Program and submit it with their sample or apply after receiving their bill. The application form is included in the patient sample collection kit or available to download at **GeneDx.com/financial-assistance-program**.
To check patient eligibility for this program, visit the Financial Application Tool at **GeneDx.com/financial-assistance-program**.

### Cost estimate for patients

For patients with contracted commercial insurance and Medicare Advantage plans, we offer the option to contact the patients health insurer to obtain a cost estimate.

The cost estimate is provided by your patient's insurance provider and is based on their response at the time of request. The calculation is based on the patient's current benefits, including their deductible and, if applicable, coinsurance. This is only an estimate and the final amount will not be determined until the claim is processed. This estimate does not consider whether your patient meets coverage criteria or the final status of a prior authorization, which is required for most genetic testing.

For out-of-network plans, contact the payor directly to determine patients out-of-network coverage and request a cost estimate.

**How it works:**

- If the estimated out-of-pocket cost is less than $250, GeneDx will run the test as ordered.
- If the estimated out-of-pocket cost is greater than $250, GeneDx will attempt to contact the patient to discuss their options. If we are unable to reach the patient, we will run the test as ordered 10 days after the last outreach attempt.

If you would like to hold the order until an estimate is provided by the patient's insurance provider, please use one of the methods below at the time of order placement:

1. Check "hold test for cost estimate" checkbox in the portal (if present) and document it in the billing notes
2. Add "hold test for cost estimate" in the billing notes within the portal
3. Check or write "hold test for cost estimate" on the paper test requisition

! *GeneDx does not conduct a cost estimate for:*

- *Medicaid insurance (as we do not balance bill those patients)*
- *Out-of-network insurance providers (as we cannot access enough information to enable accurate estimates)*

**Do you or your patients have billing questions?**

Contact your GeneDx representative or get in touch with our dedicated billing team at **Billing@GeneDx.com**. Patients can also call us at **888-729-1206**.

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# Exome and genome ordering checklist

Take the guesswork our of placing a GeneDx order. When placing an order with GeneDx, please be sure to follow each of these steps:

- ○ **Obtain informed consent** from the patient and/or their legal guardian
- ○ **Provide the patient or legal guardian's phone number** in case GeneDx needs to contact them directly regarding insurance or billing
- ○ **Clearly mark whether the patient is currently a hospital inpatient** within the payment section
- ○ **Upload or attach at the time of order:**
  - Prior authorization (PA) approval documentation, if obtained in advance
  - Insurance-required prior authorization form, if applicable
  - A clinical note that helps demonstrate why the test is medically necessary, including why it's the most appropriate test and how test results could potentially impact medical management
  - All relevant diagnosis codes (ICD-10)
  - A copy of the patient's primary insurance card (front and back)

Please note: Missing information could result in delayed or canceled testing. If you need to send information after submitting your order, please upload it in the Provider Portal or email the documents to Support@GeneDx.com, including the accession or portal order number. If using secure email, please also include patient name and date of birth.

- ○ **Sign every test order,** either on the paper test requisition form (TRF) or electronically via the Provider Portal
- ○ If requesting genetic counseling services, please **complete the referral form** and email it to **Referral@GeneDx.com**
- ○ If relative samples are included for duo or trio testing, we will need their names and dates of birth.
- ○ If samples are not collected in clinic and kit should be mailed, we will need relevant addresses.

The information described here applies to orders placed within the United States.

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