Pediatric Genomic Testing Playbook

Implementing Genomic Testing in Pediatric Primary Care

BioLogic Pharma Solutions | Version 1.0 | 2026

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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.

Why Pediatric Practices Should 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. Interpretation supported by **GeneReviews**®, reducing uncertainty and shortening the diagnostic journey.
- **Guides medical management and surveillance**
  Informs condition-specific care, monitoring, and risk mitigation based on known disease patterns.
- **Informs prognosis**
  Provides insight into expected disease course to support clinical decisions and set 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 many studies, connecting families to research and investigational therapies.

Guideline References

American Academy of Pediatrics: https://publications.aap.org/pediatrics/article/143/4/e20183995

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

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

Which Patients to Consider for Genetic Testing

Consider genomic testing when children present with unexplained developmental or neurologic conditions.

Clinical Triggers

- Global developmental delay
- Intellectual disability
- Autism spectrum disorder
- Epilepsy with developmental delay
- Multiple congenital anomalies
- Dysmorphic features
- Family history of rare genetic disorders

Screening Checklist

If 2 or more are present, consider testing

☐ Developmental delay
☐ Seizures
☐ Dysmorphic features
☐ Congenital anomalies
☐ Unexplained hypotonia
☐ Developmental regression
☐ Strong family history

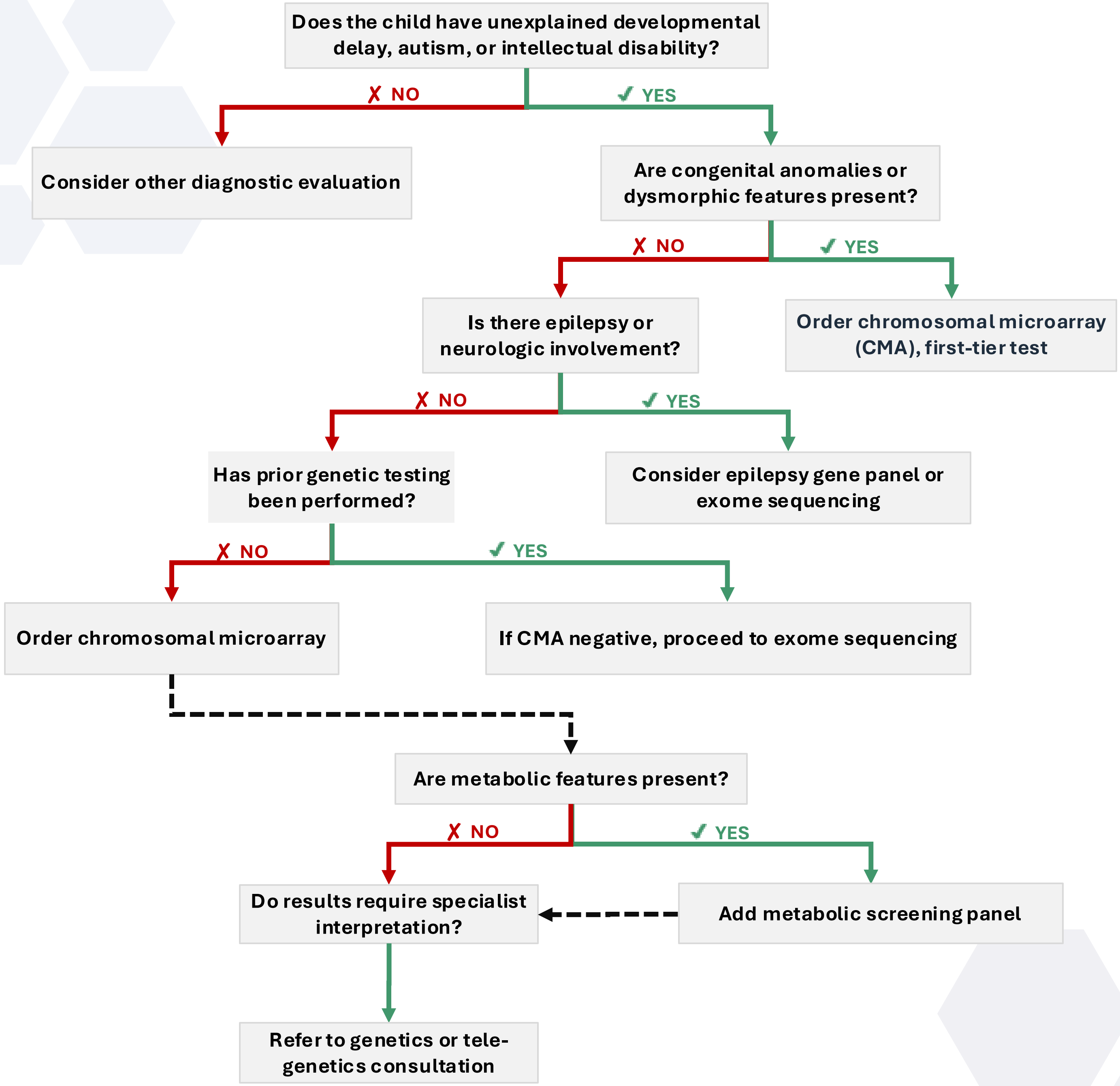
Reference Tools

ClinGen: https://clinicalgenome.org

OMIM: https://www.omim.org

Pediatric Genetic Testing Decision Tree

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



Does the child have unexplained developmental delay, autism, or intellectual disability?

✗ NO

Consider other diagnostic evaluation

✓ YES

Are congenital anomalies or dysmorphic features present?

✗ NO

Is there epilepsy or neurologic involvement?

✓ YES

Order chromosomal microarray (CMA), first-tier test

✗ NO

Has prior genetic testing been performed?

✓ YES

Consider epilepsy gene panel or exome sequencing

✗ NO

Order chromosomal microarray

✓ YES

If CMA negative, proceed to exome sequencing

Are metabolic features present?

✗ NO

Do results require specialist interpretation?

✓ YES

Add metabolic screening panel

Refer to genetics or tele-genetics consultation

Key Reference Databases

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

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

Selecting the Appropriate Genetic Test

Pathway A — Developmental Delay or Autism

Step 1

Chromosomal Microarray (CMA)

First-tier test for developmental delay, ASD, or intellectual disability

→

Step 2

Exome Sequencing

If CMA is negative and clinical suspicion remains

Pathway B — Epilepsy with Developmental Delay

Step 1

Epilepsy Gene Panel or Exome Sequencing

Initial evaluation for epilepsy with developmental features

→

Step 2

Metabolic Evaluation

If clinically indicated based on presentation

Key Resources

ClinGen: https://clinicalgenome.org

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

Project Find Out: https://projectfindout.org/

PLUGS: https://www.schplugs.org/plugs-patient-toolkit/

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.

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

Sponsored Lab 2
sponsoredlabwebsite.com

Sponsored Lab 3
sponsoredlab.com

GeneDx Genetic Tests

Whole Genome Sequencing
Analyzes the entire genome, both coding and noncoding regions, to help identify genetic variants that may contribute to disease.

Prenatal Genetic Testing
Offers CMA, ExomeDx™ Prenatal, and GenomeDx™ Prenatal

Whole Exome Sequencing
Analyzes the protein-coding regions of ~20,000 genes to help identify the molecular basis of suspected genetic disease.

Targeted Variant Testing
Analyzes a specific, previously identified genetic variant. Commonly for cascade testing, carrier testing, confirmatory testing, and mosaic variant analysis.

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.

ICD-10 Code	Description
F84.0	Autism Spectrum Disorder
F88	Global Developmental Delay
R62.50	Developmental Delay, Unspecified
G40	Epilepsy
Q89.7	Multiple Congenital Anomalies

Coding Guidance

AAP Practice Management Coding Resources: https://www.aap.org/en/practice-management/coding/

https://cdn.prod.website-files.com/69179b7c79bdf6bd80578e72/6984bd7b3b67f5cbd44e97ec_67897-Exome-TRF-v260205.pdf

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 due to unexplained developmental delay and suspected genetic etiology. First-tier chromosomal microarray testing is recommended by both the American Academy of Pediatrics and the American College of Medical Genetics as part of standard evaluation for this presentation.*

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

Integrating Tele-Genetic Services

Tele-genetics expands access to genetic counseling and interpretation, particularly for underserved populations.

Tele-Genetics Providers

Genome Medical

https://www.genomemedical.com

InformedDNA

https://informeddna.com

TeleRare Health

https://telerarehealth.com

Use Cases

Pre-Test Counseling

Provide remote education on testing purpose, potential outcomes, limitations, and implications to support informed consent before ordering genomic testing.

Complex Case Interpretation

Enable specialist review of challenging or ambiguous results to support accurate variant interpretation and appropriate clinical decision-making.

Family Cascade Testing

Facilitate remote coordination of testing for at-risk relatives to identify carriers or affected individuals and clarify inheritance patterns.

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.
(ex of real result)

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. Guidance on result interpretation and family communication can be found from **GeneDx**.

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/

TeleRare Health
https://telerarehealth.com

Ambry Genetics
https://www.ambrygen.com/providers

Rare Disease Resources

NORD: https://rarediseases.org

Global Genes: https://globalgenes.org

Practice Implementation Timeline

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

Weeks 1-2

Breakfast and Learns

Materials dump, 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

Site Operating Semi-Independently

Sites manages most ordering and workflow independently with minimal external support

Week 8-9

Results Review Sessions

1-hour sit-in with results over 3, 20-minute sessions

Week 10-12

Program & Outcomes Assessment

Assess overall program and results of patients tested

Practice Performance Metrics

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

Clinical Metrics

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

Overall improvement rate (%)
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.

Time from clinical suspicion to test order (days)
Diagnostic workflow efficiency from initial suspicion to test initiation.

Operational Metrics

Prior authorization approval rate (%)
Payer success rate for genomic testing requests.

Time from order to specimen submission (days)
Operational speed from test ordering to sample collection and shipment.

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.

Telehealth counseling utilization (%)
Uptake of tele-genetic services among patients receiving genetic counseling.