



Patient First Name _____ MI _____ Last Name _____
 Gender: Male Female Unknown
 Date of Birth: (mm/dd/yy) _____
 Patient Address: Street _____ Apt # _____
 City/State/Zip Code _____
 Patient Phone: Home _____ Work Phone _____
 Medical Record Number _____
 Other Submitter's Patient Id(s) _____

Reporting Address

Physician/CGC _____
 Address _____
 Phone _____ Fax (*Important) _____
 Email _____ Beeper _____

Duplicate Report Address

Physician/CGC _____
 Address _____
 Phone _____ Fax (*Important) _____
 Email _____ Beeper _____

Informed Consent

I have read the consent document and give GeneDx permission to perform oligo array testing and gene sequencing as described.
 (Sign here): _____

Ordering Checklist

- Sample Submission form(s) (page 1-4)
- Completed Payment Option form (page 3)
- Specimen Tube(s), appropriately labeled

For Testing Services for Rare Mendelian Disorders, use Separate GeneDx Sample Submission Form
For ONLY GenomeDx Oligo Microarray Analysis, use separate GenomeDx Sample Submission

Specimen Requirements

1-3 mL of whole blood in lavender top (EDTA) tube per person. Samples from parents are highly recommended for the most accurate and rapid interpretation of the child's result.

Specimen Submitted: Patient Mother Father

Blood in EDTA
 Date Obtained _____
 Mother's First Name _____ MI _____ Last _____
 Father's First Name _____ MI _____ Last _____

Clinical Information (Check all that apply):

This information is crucial for accurate interpretation of the test results.

General:

- Developmental delay
- FTT
- IUGR
- Short stature

Cardiac:

- Congenital heart disease

Skeletal:

- Rib anomalies
- Scoliosis
- Skeletal abnormalities

Neurological:

- Agenesis of the corpus callosum
- Autism/ASD
- Asperger Syndrome
- Chiari malformation
- Holoprosencephaly
- Hypertonia
- Hypotonia
- Lissencephaly
- Pervasive Developmental Disorder (PDD)
- RETT Syndrome
- Seizures
- Speech delay

Hand/feet:

- Clinodactyly
- Club foot
- Missing digit(s)
- Polydactyly/Syndactyly
- Rocker bottom feet

Uro-Genital:

- Ambiguous genitalia
- Horseshoe kidney
- Hydronephrosis
- Hypospadias
- Renal agenesis

Head:

- Cleft lip/palate
- Macrocephaly (>2.5 SD)
- Microcephaly

Other:

- Biliary atresia
- Choanal atresia
- Cong. Diaphragm. Hernia
- Malrotation of gut
- Tracheoesophageal fistula

Eyes:

- Aniridia
- Cataract
- Coloboma

Dysmorphic features: _____
 Other medically significant problems: _____
 Other neurological features: _____
 Prev. cyto. result (attach if avail.): _____
 Suspected specific syndrome(s): _____

Family History (check all that apply)

Positive for any clinical feature: No Yes
 Which feature: _____
 Relative: _____
 Which feature: _____
 Relative: _____
 Does the mother have any significant medical problems? _____
 Does the father have any significant medical problems? _____

Payment: Please Complete Page 3

AutismDx: Autism Diagnostic Testing

**For GenomeDx Oligo Microarray CGH Analysis, Use Separate GenomeDx Sample Submission Form
For Genetic Testing Of Other Mendelian Disorders, Use Separate GeneDx Sample Submission Form**
Please check appropriate boxes and send or fax all sheets

Test Code Test Name

ASD and Macrocephaly

307 Autism Panel 1: FEMALE

- Tier 1 GenomeDx 105k Oligo Array CGH and PTEN sequencing
- Tier 2 MECP2 sequencing
- Tier 3 CDKL5 sequencing

ASD and Macrocephaly

308 Autism Panel 1: MALE

- Tier 1 GenomeDx 105k Oligo Array CGH and PTEN sequencing
- Tier 3 CDKL5 sequencing

Test Code Test Name

ASD without Macrocephaly

309 Autism Panel 2: FEMALE

- Tier 1 GenomeDx 105k Oligo Array CGH and MECP2 sequencing
- Tier 2 CDKL5 sequencing

ASD without Macrocephaly

310 Autism Panel 2: MALE

- Tier 1 GenomeDx 105k Oligo Array CGH and Tier 2 CDKL5 sequencing

Test Code Test Name

311 Autism Panel 3

Female or Male with syndromic ASD

(multiple congenital anomalies present or specific genetic syndrome suspected)

GenomeDx 105k Oligo Array CGH

Reflex to specific gene testing according to clinical presentation and suspected diagnosis (*à la carte* testing)

Cowden Syndrome / Bannayan-Riley-Ruvalcaba Syndrome

- 1951 PTEN Sequencing
- 1952 PTEN MLPA, deletion/duplication testing, if appropriate

Rett Syndrome / Atypical Rett Syndrome

- 3041 MECP2 Sequencing
- 3042 MECP2 MLPA, deletion/duplication testing, if appropriate

West Syndrome / Infantile Spasms, Developmental delay / Early onset epilepsy

- 3051 CDKL5 (STK9) Sequencing

Test Code Test Name

Smith-Lemli-Opitz Syndrome

- 2501 DHCR7 Sequencing

Smith-Magenis Syndrome

- 2511 RAI1 Sequencing
- 2512 RAI1 CopyDx, deletion/duplication testing, if appropriate

Cohen Syndrome

- 227 COH1 Sequencing

Payment Options

I. Institutional Billing Information:

GeneDx Account # _____

Hospital/Lab Name _____

Contact Name _____

Address _____

City _____ State _____ Zip Code _____

Phone _____ Fax _____

BILLING STAMP

2. Payment by credit card

The full amount of the test fee is charged at the time of sample submission.

Name as it appears on card _____

Billing address _____

City _____ State _____ Zip Code _____

Phone _____

Mastercard Visa Discover American Express

Account Number _____

Expiration date _____ 3/4 Digit Security Code _____

Please bill my credit card in the amount of \$ _____ for diagnostic laboratory tests performed by GeneDx, Inc.

Signature (Required) _____ Date _____

3. Payment by check or money order:

Minimum of 75% of the cost of the test is required at the time of sample submission, with the remainder of the fee billed at the time of test completion.*

Check or money order enclosed in the amount of \$ _____.

*** For patients from outside the United States, 100% of the fee is due at the time of sample submission**

4. Insurance Billing:

GeneDx does not bill insurance companies directly unless all of the following is submitted:

- Credit card information (complete part 2) to which any outstanding balance may be billed;
- An authorization number or letter of agreement from the insurance company.
 - The letter of agreement should be directed to GeneDx
 - detail the reimbursement rate
 - the name of the department or individual to whom the bill will be sent (including address, phone and fax numbers)
 - the patient's name and policy number.
- Copy of both sides of the insurance card.
- ICD9 codes (to be provided by physician) _____

I UNDERSTAND THAT I AM RESPONSIBLE IN ALL CASES FOR ALL FEES NOT COVERED BY INSURANCE.

Signature (Required) _____

Note

IF YOU plan to apply on your own to your insurance carrier for reimbursement of *your* expenses for this test, the following information may be helpful in the case that GeneDx is requested by the carrier to prepare supporting documentation for you to use in your insurance claim:

Insurance Carrier _____
Is this a Blue Cross/Blue Shield Plan? YES NO

Subscriber Name _____
Is this a Medicaid plan? YES NO

Subscriber DOB _____

ID# _____

Informed Consent for Oligo Array CGH and Gene Sequencing

My signature below or on page 1 of the Sample Submission Form for AutismDx indicates that I have been informed of the following facts about the AutismDx test and I have had the opportunity to have any questions answered.

Why is this test done?

1. In the AutismDx test (my/my child's) DNA will be studied to look for genes or spaces between genes where the number of copies is lower or higher than usual.
2. Many genetic diseases and syndromes are caused by a deletion or duplication of one or more genes.
3. Many other diseases and syndromes are caused by small DNA changes in one or more specific genes rather than in gene copy number. In the AutismDx test (my/my child's) DNA will also be studied by DNA sequencing to look for such a small change (mutation) in a subset of genes that have been associated with my/my child's diagnosis.
4. This test is not the only way to look for genetic changes, and my physician may recommend this test before or after doing other genetic tests.

What might I find out from this test?

5. I might learn that no gene duplications or deletions or gene mutations were found. This outcome does not mean that (I/my child) does not have a genetic disease.
6. I might learn that there is a DNA mutation in a specific gene or that a gene is duplicated or deleted, explaining the cause of a disorder that I already know (I have/my child has).
7. I might learn that a specific mutation or gene duplication deletion was found that can predict possible long term medical problems that I do not already know about. My physician will be informed of any long term risks that become apparent through this test, according to current medical understanding.
8. This test does not have the ability to detect all the long term medical risks (I/my child) might experience.

What is learned by comparing parent and child DNA?

9. Some genes or spaces between genes tend to have duplications or deletions or DNA mutations that do not cause medical problems. They may be normal genetic variations between individuals.
10. When a duplication or deletion or DNA mutation is found in the patient being tested, it is important to find out if a parent also has it. If it is a spontaneous change in the child's DNA, then it is more likely that the duplication or deletion or DNA mutation is responsible for the child's medical problem.
11. By sending the parents' and child's DNA at the same time, just in case the parents' DNA might be needed, the test interpretation can be sped up and the reporting of ambiguous results can be reduced.
12. If a parent is not the actual biological parent of the child, the laboratory may or may not recognize the situation when parental DNA is tested. If the laboratory was not informed in advance about any non-biological relationship and does not deduce it, false conclusions may be drawn about the significance of duplications, deletions, or DNA mutation in the child.
13. If the laboratory does deduce that there is a non-biological relationship between a parent and child, it may be necessary to disclose the fact to the physician and/or to call the child's result inconclusive.

Sign here or on the Sample Submission (Order) Form.

Signature: _____ **Date:** _____