



Patient information

First name _____ Last name _____
 Gender: Female Male Unknown
 Date of birth (mm/dd/yy) _____
 Mailing address _____
 City _____ State _____ Zip code _____
 Home phone _____ Work Phone _____

Reporting Address

Physician / Genetic counselor _____
 Mailing address _____
 City _____ State _____ Zip code _____
 Phone _____ Fax (Important) _____
 Email _____ Beeper _____

Duplicate Report Address

Physician / Genetic counselor _____
 Mailing address _____
 Phone _____ Fax (Important) _____
 Email _____

Sample Information

Medical record # _____ Specimen ID # _____
 Date sample obtained (mm/dd/yy) _____
Sample Type:
 blood in EDTA (for array CGH; purple top - single tube of 2-5mL)
 blood in heparin (for FISH; green top - single tube of 2-5mL)
 DNA: source _____ (tissue?) / concentration _____ (µg/mL)
 Fetal sample _____ (tissue?)

If other samples submitted

Relationship to patient _____ Name _____ Sample type _____
 Relationship to patient _____ Name _____ Sample type _____

Test Requested

Please select choice and provide clinical information below

Whole-genome high-resolution array CGH 105K (GenomeDx)
 Targeted array CGH 15K (FISHonChipDx)
 FISH (locus-specific). Please specify genomic region or disorder below.
 1. _____ 2. _____
 write chromosomal region or disorder
 Follow-up parental testing / Other family member testing

Clinical Information (Check all that apply):

This information is crucial for interpretation of array CGH results

General:

- Developmental delay
- Failure to thrive
- Intrauterine growth retardation
- Short stature

Neurological:

- Agensis of the corpus callosum
- Autism
- Holoprosencephaly
- Hypertonia
- Hypotonia
- Lissencephaly
- Seizures

Head:

- Cleft lip/palate
- Macrocephaly
- Microcephaly

Eyes:

- Aniridia
- Cataract
- Coloboma

Skeletal:

- Rib anomalies
- Scoliosis
- Skeletal abnormalities
- Clinodactyly
- Club foot
- Missing digit(s)
- Polydactyly/Syndactyly
- Rocker bottom feet

Uro-Genital:

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

Other:

- Biliary atresia
- Choanal atresia
- Cong. diaphr. hernia
- Malrotation
- Tracheoesophageal fistula

Dysmorphic features: _____
 Congenital heart disease: _____
 Other medical problems: _____
 Suspected syndrome(s): _____
 Previous cytogenetics: _____ (attach copy)
Family history: _____

Expedited Testing. I would like expedited testing for the reason:

Pregnancy (gestational age: _____ weeks)
 Transplantation
 Other _____

Ordering Checklist

Sample submission form Completed payment option form (page 2)
 Informed consent (page 3) Specimen tube, appropriately labeled

For office use only:

Payment Options

Institutional billing information:

GeneDx Account # _____
Hospital/Lab Name _____
Contact Name _____
Address _____
City _____ State _____ Zip Code _____
Phone _____ Fax _____

BILLING STAMP

Test

Insurance policy

Patient responsibility

Whole-genome array CGH (GenomeDx)

GeneDx will bill and appeal on patient's behalf

Co-pay and unmet deductibles only

Targeted array CGH (FISHonChipDx) and FISH for proband's relatives

GeneDx will courtesy bill insurance and then bill unpaid balance to patient's credit card

Any unpaid balance after insurance reimbursement is complete

Insurance billing

(Note: FISHonChipDx and FISH tests require credit card information)

I have prior authorization # _____

Name of insured _____ Date of birth (mm/dd/yy) _____

Relationship to insured: Self Spouse Child Other _____

Insurance carrier _____

ID # _____

Group # _____ Policy name _____

Social security # _____

Please include a copy of the front and back of the patient's insurance card.

GeneDx will bill the insurance company and appeal for payment on the patient's behalf.

I would like GeneDx to bill my credit card for my portion of the test cost

Name as it appears on the card _____

Mastercard Visa Discover American Express

Account number _____ Expiration date (mm/yy) _____ CVC _____

Signature _____ Date _____

Patient pre-payment

I understand that my credit card will be charged the full amount for the testing

Please bill my credit card (all major cards accepted)

Name as it appears on the card _____

Mastercard Visa Discover

American Express

Account number _____

Expiration date (mm/yy) _____ CVC _____

Amount (USD) _____

Signature _____ Date _____

Note: GeneDx provides a financial assistance program or hardship discount for patients who do not carry a commercial insurance plan and cannot afford to pay out-of-pocket. Please call 301-519-2100 x6106

Informed Consent for Cytogenetic Oligo Array CGH

My signature below indicates that I have been informed of the following facts about array CGH testing and I have had the opportunity to have any questions answered.

Description of the test

1. Array CGH is indicated for clinical disorders in which a chromosomal abnormality is suspected.
2. This test examines the human genome for loss or gain of DNA material on chromosomes. Many genetic disorders are caused by deletion or duplication of one or more genes. This array can detect deletions or duplications that extend 100 kilobases or higher across the non-repetitive regions of the whole human genome and 5-20 kilobases in >150 specifically targeted clinically significant regions.
3. A positive result indicates that a clinically significant genomic imbalance is detected in the patient. A negative report indicates that genomic imbalances of known or expected clinical significance were not identified. In a small number of cases, the array detects genomic imbalances of unknown significance and parental testing is often useful to determine the significance in these cases.
4. An array CGH test is not the only method used to detect genetic changes, and my physician may recommend this test before or after doing other genetic tests. Array CGH cannot detect certain chromosomal imbalances in which the amount of DNA material remains unaltered. Very small changes that are beyond the resolution of the array can also go undetected.

What can I find out from this test?

1. I may learn that no loss or gain of genetic material was detected. This outcome does not mean that (I/my child) do not have a genetic disease.
2. I may learn that a specific gene or genes is duplicated or deleted, explaining the cause of a disorder that I already know I have or that my child has.
3. I may learn that gene duplications or deletions were identified and may have possible long-term medical concerns that I do not already know about. My physician will be informed of any such long-term risks, according to current medical understanding. This test does not detect all long-term medical risks that (I/my child) may be subject to.

Parental testing

1. Some areas of the human genome can exist in less or more than the normal quantity (copy number) and not cause medical problems. These occurrences represent normal genetic variation between individuals.
2. When a duplication or deletion is found, it is important to find out if a parent also carries it. If the duplication or deletion proves to be a spontaneous change in the child's DNA it is more likely (but not absolutely certainly) that it is responsible for the child's medical problem.
3. If an individual is not the actual biological parent of a child who is tested, the laboratory may or may not recognize this relationship during testing. If the laboratory is not provided with accurate information on biological relationships in the family tested, false conclusions may be drawn about the significance of duplications and deletions detected in the child. If the laboratory finds a discrepancy between submitted information and the test results (showing a non-biological relationship between a child and a parent), it may be necessary to disclose this to the physician and/or to call the results inconclusive.

Parental Confidentiality and counseling

1. Only the referring physician, genetic counselor, or ordering laboratory will receive copies of test results to maintain confidentiality.
2. It is recommended that the patient/family receive genetic counseling regarding array CGH before and after the test. Further testing or additional consultations with physicians may be necessary.

Specimen retention

1. Submitted specimen are not banked at GeneDx and DNA samples are not returned to individuals or referring physicians. In some cases, if further diagnostic tests are needed, a referring physician may request in writing that additional tests be performed on an existing DNA sample (additional costs apply).

Sign here to provide consent for array CGH testing at GeneDx:

Signature: _____ **Date:** _____

GeneDx participates in the International Standard Cytogenetic Array (ISCA) consortium. Anonymized array CGH data and clinical information may be submitted to a HIPAA-compliant, de-identified public database as part of the National Institute of Health's effort to improve diagnostic testing and enhance our understanding of the relationships between genetic changes and clinical symptoms (visit the consortium website at <https://isca.genetics.emory.edu>). Confidentiality of each sample is maintained. If you wish not have genomic information derived from your blood/DNA sample submitted to a database, although it is anonymous, please check the box below.

Refusal for inclusion in these efforts may be indicated by checking this box. (If a box is not marked, consent is implied.)