

CONSENT FOR ARRAY COMPARATIVE GENOMIC HYBRIDIZATION (aCGH) ANALYSIS

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Patient Name: _____

DOB: _____

Parent/Guardian Name: _____

Date: _____

Test Description and Limitations

- 1) Some individuals have developmental delays, intellectual disability, autism spectrum disorders, and/or birth defects as the result of loss(es) or gain(s) of chromosomal information, which are referred to as *copy number changes*. aCGH analysis evaluates an individual's chromosomal information for such changes by comparing it to a known reference sample.
- 2) CombiMatrix Diagnostics uses an oligonucleotide array with high density coverage (probe spacing of ~3kb) of regions of known clinical significance and extensive backbone coverage (probe spacing of ~22kb) of all other genomic regions. This array covers 510 genes associated with congenital/developmental disorders and also 2126 annotated genes not currently associated with syndromes.
- 3) aCGH analysis may be ordered prior to, in tandem with, or following a routine karyotype. aCGH analysis cannot detect chromosomal rearrangements that do not result in a net gain or loss of chromosomal information. aCGH analysis also cannot detect very small changes that are beyond the resolution of the test. Such changes may only be detectable through other forms of molecular testing, such as sequence analysis.

Test Results

- 1) **Abnormal/Positive result:** a copy number change associated with a known phenotype was identified.
- 2) **Normal/Negative result:** no clinically significant copy number changes were identified. (Please note that an individual can have a normal aCGH result and still have a genetic or chromosomal disorder.)
 - Some regions of the genome are known to show copy number variability without phenotypic consequences. This is considered to be part of normal human genetic variation.
 - Copy number changes that are seen in at least 1% of the phenotypically normal population are considered *benign copy number changes*, and will be listed as such, when present.
- 3) **Non-diagnostic result:** a copy number change that has not been associated with a known phenotype, but is also not considered a benign copy number change, was identified. Parental studies are often helpful in determining the nature of the copy number change.

Parental Testing

- 1) If an abnormal or non-diagnostic result is encountered, testing of the individual's biological parents is recommended to help determine whether the alteration is new in the individual or inherited from a parent.
- 2) If the parent tested is not the actual biological parent (e.g. misattributed paternity), this can lead to inaccurate interpretation of the patient's results.

Confidentiality and Genetic Counseling

- 1) Test results will be released only to the referring physician, genetic counselor, and/or reference laboratory in order to protect patient confidentiality.
- 2) All samples are destroyed after 60 days; however, extracted DNA is retained on all specimens indefinitely. Testing for validation or educational purposes may be performed only after all protected health information is removed and the sample de-identified.

If you do not wish for your/your child's de-identified sample to be used for these purposes, please initial here: _____

- 3) Genetic counseling to discuss the benefits and limitations of aCGH analysis is recommended prior to testing.
- 4) Once the aCGH results are complete, genetic counseling is recommended for results discussion. Depending upon the results of the aCGH analysis, further testing and/or diagnostic evaluations may be indicated.

Authorization

I request and authorize CombiMatrix Diagnostics to perform aCGH analysis on my/my child's sample. I understand the information above, and have had an opportunity to ask questions, which have been answered to my satisfaction.

X

Parent/Guardian Signature

Date

I have explained aCGH analysis and its limitations to this patient and/or legal guardian, and answered all questions.

X

Physician/Genetic Counselor Signature

Date

As a participant in the International Standards for Cytogenomic Arrays (ISCA) Consortium, CombiMatrix Diagnostics contributes de-identified clinical information and aCGH results to a HIPAA-compliant public database, which is part of the NIH's effort to improve our understand of the relationships between genetic changes and clinical symptoms. The confidentiality of each sample is maintained. If the patient/guardian does not wish to have their de-identified genomic information submitted to this database, please check the box below.

The patient/ guardian does not wish to provide this information to the ISCA database. (If the box is not marked, consent is implied.)