

UW Cytogenetic Services

Wisconsin State Laboratory of Hygiene

Fall 2015

Policy Updates

UW Cytogenetics and Molecular Genetics within the Wisconsin State Laboratory of Hygiene (WSLH) have recently updated its policy regarding parental and private variant carrier testing. The updated policies are listed below.

Policy on Parental Testing: WSLH Cytogenetics and Molecular Genetics will request parental specimens in the event analysis of the parents will aid in the interpretation of a variant of uncertain significance or when the assessment of phase is critical to a determination of pathogenicity. Studies requested for these purposes will be done as an additional test free of charge and may use an alternative method at the director's discretion using proband specimen as an assay positive control. Requests will be made as a recommendation in the proband's final report. Parental results, if successful, will be reported as an amendment to the proband's original record and will not be issued independently.

Policy on Carrier Testing of a Private Variant: WSLH Cytogenetics and Molecular Genetics will offer carrier testing of a private variant as a fee for service test when a pathogenic or likely pathogenic variant has been detected in a proband using a clinically validated test and confirmed with a variant specific assay. The fee for the assay will depend on the method selected as the confirmatory test. Clients interested in carrier testing of a private variant should contact the laboratory for test method details and fees.

Definitions

Phase assessment: Evaluation of variants for cis or trans allele positioning

Parents: Proband's biological parents

Proband: An affected person identified independently of family members

Private Variant: A variant that is unique to a family or specific population

Pathogenic Variant: A variant consistent with the classification of pathogenic according to ACMG guidelines and recommendations or reported as such in a clinical genetic report

Likely Pathogenic Variant: A variant consistent with the classification of likely pathogenic according to the ACMG guidelines and recommendations or reported as such in a clinical genetic report

Variant of uncertain significance: A variant where criteria for pathogenic or benign are not met or are contradictory

Please call our laboratories at 608-262-0402 with any questions.

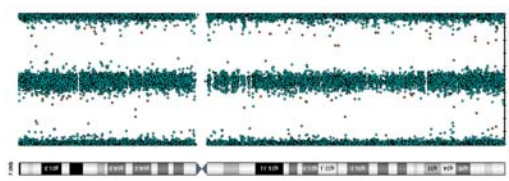
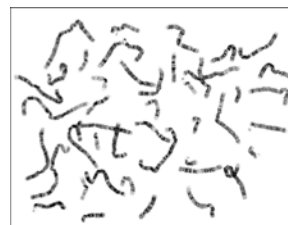
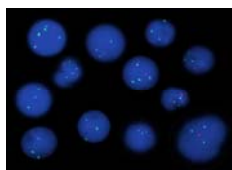
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FAQs about follow-up testing for an abnormal microarray

How is this testing performed?

Confirmation of array results can be accomplished by various methodologies, including fluorescence in situ hybridization (FISH) analysis and quantitative PCR (qPCR). Parental studies may be performed using either of the above methods or a targeted analysis microarray. Method of confirmation will be chosen by the UW Cytogenetics Laboratory based on size and location of the aberration.

What is the turn-around time?

Confirmatory, parental, or private variant testing will be completed in approximately 7-21 days.

How can I order this follow-up testing?

Testing can be initiated by either calling our laboratory at 608-262-0402 or by writing in your testing request on our 'Genetic Diagnosis' requisition form and faxing it to our laboratory at 608-265-7818

How long do I have to add on this testing?

Confirmatory or parental testing can be ordered up to one year after the test is ordered and is subject to specimen availability.

How can I tell if testing will be done at no charge?

We will have language in our interpretation stating whether further testing is warranted and at what cost.

Examples:

“Parental studies are recommended to determine if this copy number variant is *de novo* or inherited and to help determine the clinical significance of this finding. This testing will be provided free of charge.”

Or

“Parental studies are recommended to determine if this copy number variant is *de novo* or inherited and to help determine recurrence risk estimates. This testing is available through our laboratory for an additional service fee.”

What should I include on the test requisition form when ordering this testing?

For parental follow-up testing, we require either the name and date of birth of the proband or the case number of the proband's initial report.

For familial (private variant) testing we request the same information, or at a minimum, a complete ISCN to complete any follow-up testing.

Please call our laboratory at 608-262-0402 with any questions.
