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Changes to New York State policies for Chromosomal Microarray Analysis (CMA)

Current New York State (NYS) Cytogenetics Quality Assurance Program policy requires chromosomal microarray testing to be adjunct to routine standard metaphase chromosome analysis for mitotic specimens and requires confirmation of abnormal copy number results by a second, independent method. However, substantial laboratory experience with CMA since this policy was implemented has revealed it to be a robust methodology and improved understanding of its limitations.

In recognition of the current state of CMA testing, Cytogenetics Quality Assurance Program policies have been revised regarding this method.

New Policy: CMA and standard chromosome analysis.

Standard chromosome analysis in conjunction with CMA is not required for pre-implantation genetic diagnosis (PGD) testing or products of conception (POC)/miscarriage samples.

For all other situations, the laboratory may:

1. Establish and use policies and procedures describing the circumstances under which standard chromosome analysis is performed or recommended when CMA is ordered. These policies and procedures must consider criteria including but not limited to the laboratory's historical data, the patient population(s), specimen type (s), reason(s) for referral and potential for mosaicism/clonality. OR
2. Perform adjunct standard metaphase chromosome analysis with each CMA test.

Standard chromosome analysis may be performed by the laboratory performing the CMA or by a reference laboratory, but must be performed by a laboratory holding the appropriate Cytogenetics category on its NYS permit. The referring laboratory is responsible for overall interpretation, as appropriate.

Abbreviated chromosome studies intended to confirm or clarify an aberration detected by CMA are recognized as differing from routine studies. Abbreviated studies are acceptable for this purpose. All other applications, including studies to rule out aberrations not detected by CMA, must adhere to New York State Clinical Laboratory Standard of Practice CG16.

The laboratory's policy regarding standard chromosome analysis and CMA must be available to clients.

New policy: Confirmation of abnormal CMA results

Confirmation of abnormal CMA results is not required for pre-implantation genetic diagnosis (PGD) testing or products of conception (POC)/miscarriage samples.

For all other situations, the laboratory may:

1. Establish and use an individualized quality control plan (IQCP) in accordance with Quality Control Sustaining Standard of Practice 1 (QC Design S1): Design of Individualized Quality Control Plan that discusses confirmation of abnormal CMA results as part of QC/QA for CMA. The IQCP must specify circumstances under which results are confirmed. The IQCP must be based on the laboratory's historical data including aberration type, size and complexity, mosaicism/clonality, and quality control parameters and acceptance criteria for each array and/or batch. The laboratory must have appropriate policies and procedures in place to implement the IQCP. OR

2. Confirm all abnormal chromosomal microarray (CMA) copy number results by an independent method.

The laboratory's policies and procedures regarding CMA and abnormal result confirmation must be submitted as part of method validation for new CMS tests and significant modifications of previously approved CMA tests. These policies must also be available to clients.

Confirmation testing must be performed using approved methods by a NYS permitted laboratory that holds the appropriate category on its permit. Confirmation testing may be performed by the laboratory performing the CMA test or by another NYS permitted laboratory that has appropriate permit and method approvals. Independent methods include but are not limited to standard metaphase chromosome analysis, FISH, qPCR, MLPA, or a second CMA platform. Independent methods do not include additional probes on the same array or repeat testing on the same array platform.

Constitutional aberrations may be confirmed via parental testing.