Chromosome Microarray (CMA) Testing Guide – Children and Adults

Adapted from: Palmer et al. Chromosome microarray in Australia: A guide for paediatricians. Journal of Paediatrics and Child Health 48 (2012) E59–E67

DOCTOR'S GUIDE

1. Patient to undergo CMA testing, which checks for DNA copy number variations, **not** including fragile X syndrome
2. Discussion of test process, limitations and a variety of possible outcomes listed below
3. Blood, saliva or other tissue sample collected (2-10ml in EDTA – confirm sample requirements with local laboratory)
4. Possible laboratory findings include the following:

No variants reported	Diagnostic of known, expected condition	Variant of uncertain significance (VUS)	Variant with unexpected implications (incidental finding)
• Also called negative result or uninformative • Consider referral to a genetics clinic if concerns or questions remain about a genetic diagnosis or recurrence in another pregnancy • Further testing such as a gene panel, whole exome or whole genome sequencing may be considered.	• Known copy number variant (CNV) identified • Consider referral to genetics clinic for genetic counselling as required • No further testing required unless other family members may benefit from a test (e.g. a familial condition or if planning a family).	• Further testing such as parental studies may be useful • For how to order test, please refer to CMA testing • Consider referral to genetics clinic for interpretation of report and diagnostic review.	• Copy number variant with unexpected significance identified (incidental finding) • Consider referral to specialist or genetics clinic for genetic counselling • Further testing such as testing of parents may be considered.

For a more detailed fact sheet about CMA testing and contacts for local genetic services see Centre for Genetics Education www.genetics.edu.au

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INFORMATION FOR INDIVIDUALS AND FAMILIES

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1. CMA testing checks for extra or missing segments of a person's DNA. DNA is the building block of our genes and it is important that there is the correct amount of DNA in each cell of the body for normal health and development. If there is extra or missing DNA, the test results show a "variant" is present.
2. This is not a comprehensive genetic test for all health problems and further testing may be considered.
3. The test is carried out by collecting a small sample of blood or other sample such as saliva.
4. It is important that you discuss the possible test results with your doctor when you are considering a CMA test.
5. Possible test results may include the following:

No abnormality found	DNA variant found which explains the condition being tested for	DNA variant of uncertain significance (VUS) has been found	DNA variant with unexpected implications has been found
• Also called negative result or uninformative • A balanced amount of DNA was found at this time. • You or your doctor may consider more extensive testing such as genomic testing referral to a genetics clinic.	• An extra (duplication) or missing piece (deletion) of DNA has been found • This DNA imbalance (variant) is known to be the cause of the type of condition being tested for. • An appointment at the genetics clinic or another specialist may be recommended.	• An extra or missing piece of DNA has been found • This DNA imbalance (variant) is not a known cause of a condition or the presenting health condition • Your doctor may need to check if the imbalance is causing a health problem or not by doing the same test on other family members such as parents. • An appointment at the genetics clinic may be recommended.	• Also called an incidental finding • An extra or missing piece of DNA has been found • The DNA imbalance (variant) is known to be the cause of a condition which you may not have expected (e.g. CMA test done for learning disability but result shows a DNA variant related to heart disease). • An appointment at the genetics clinic or with a specialist may be recommended by your doctor as well as further testing.

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UPDATED JUNE 2022 JOB ID 55974
SHPN: (HETI): 240950