

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

1. Patient to undergo CMA testing, a genetic test which checks for DNA copy number variations, not including fragile X
2. Discussion of test process, limitations and counselling about variety of possible outcomes listed below
3. Blood sample collected (5-10ml in EDTA - Confirm sample requirements with local laboratory)
4. Possible laboratory findings include the following:

| No abnormality found | Diagnostic of known, expected condition | Variant of unknown significance found | Variant with unexpected implications found |
|---|---|--|--|
| <ul style="list-style-type: none"> • Normal result or known, benign change detected • Consider referral to a genetics clinic if concerns remain about a genetic diagnosis or recurrence in another pregnancy • No further testing required at this stage | <ul style="list-style-type: none"> • Known copy number variant (CNV) identified • Consider referral to genetics clinic for genetic counselling • No further testing required at this stage | <ul style="list-style-type: none"> • Copy number variant of unknown significance (VOUS) identified • Consider referral to genetics clinic for interpretation of report and diagnostic review • Further testing such as parental studies may be useful | <ul style="list-style-type: none"> • Copy number variant of unexpected significance identified • Consider referral to genetics clinic for interpretation of report and genetic counselling • Further testing such as parental studies may be useful |



For a more detailed fact sheet about CMA testing or information about local genetic services see www.genetics.edu.au

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1. CMA testing is a genetic test which 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. The test is carried out by collecting a small sample of blood
3. It is important that you discuss the possible test results with your doctor when you are considering a CMA test
4. Possible test results include the following:

| No abnormality found | Results show a DNA variant which explains the concerns being tested for | Results show a DNA variant of unknown significance has been found | A DNA variant with unexpected implications has been found |
|--|---|---|---|
| <p>A normal balanced amount of DNA was found at this time.</p> <p>or</p> <p>A known imbalance of DNA (variant) was found which is not usually the cause of a health or developmental problem.</p> <p>You or your doctor may consider referral to a genetics clinic if you are still concerned.</p> | <p>An extra or missing piece of DNA has been detected</p> <p>The DNA imbalance (variant) is known to be the cause of the type of condition being tested for.</p> <p>An appointment at the genetics clinic may be recommended.</p> | <p>An extra or missing piece of DNA has been detected.</p> <p>The DNA imbalance (variant) is not a known cause of a condition</p> <p>Your doctor may need to check if the imbalance is normal in your family by doing the same test on other family members such as parents.</p> <p>An appointment at the genetics clinic may be recommended.</p> | <p>An extra or missing piece of DNA has been detected.</p> <p>The DNA imbalance (variant) is known to be the cause of a condition which you may <u>not</u> have expected (e.g. CMA test done for learning problems but result detects a DNA variant related to heart disease).</p> <p>An appointment at the genetics clinic may be recommended by your doctor as well as further testing.</p> |



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