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**
  - 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.

- **Diagnostic of known, expected condition**
  - 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).

- **Variant of uncertain significance (VUS)**
  - Further testing such as parental studies may be useful
  - For how to order test, please refer to www.genetics.edu.au/FS16
  - Consider referral to genetics clinic for interpretation of report and diagnostic review.

- **Variant with unexpected implications (incidental finding)**
  - 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 more detailed fact sheet about CMA testing and contacts for local genetic services see Centre for Genetics Education www.genetics.edu.au