Reg No 198904227G

KK Women's and Children's Hospital
SingHealth

CLINICAL GENETICS REQUEST
CHROMOSOME MICROARRAY ANALYSIS (CMA)

Patient's name label

(For downtime use)
Name:
MRN:
Account number:
Date of birth:
Sex: M / F

Patient Type ☐ Gynae ☐ Obst ☐ Neonate ☐ Paed Med ☐ Paed Surg
Ward/Bed: _____ Clinic: _____ Class: _____

Type of specimen:
☐ Peripheral blood in EDTA (3ml unless otherwise specified)
☐ Others: _____

Specimen taken Date: _____ Time: _____

Accompanying sample (s):

Please specify relationship
(spouse / sibling / parent / child etc)

Name & signature of requesting doctor
Contact no. (if urgent)

Name of consultant i/c

Date

CONSENT REQUIRED FOR ALL TESTS (SEE NEXT PAGE)

DNA120 / DNA120E ☐ CMA (180K High Resolution Copy Number)

Clinical indications: Please list all the clinical features the patient is known to have

Family History
[ ] Parents consanguineous
[ ] Parents with ≥ 2 miscarriages
[ ] Other relatives with similar clinical history
Explain:_____
_____
_____

Perinatal History
[ ] IUGR
[ ] Non-immune hydrops fetalis
[ ] Oligohydramnios
[ ] Polyhydramnios
[ ] Prematurity
[ ] Other: _____

Developmental
[ ] Fine motor delay
[ ] Gross motor delay
[ ] Speech delay
[ ] Other: _____

Cognitive
[ ] Intellectual disability/MR
List IQ, if known: _____
[ ] Learning disability
[ ] Other: _____

Behavioural/Psychiatric
[ ] Attention deficit hyperactivity disorder
[ ] Autism spectrum disorder
[ ] Other behavioural/ psychiatric abnormality
Specify: _____

Karyotype:
[ ] Not done
[ ] Normal: 46,XX / 46,XY
[ ] Others:_____
_____

Hearing/Vision
[ ] Abnormality of Vision
Specify: _____
[ ] Abnormality of Eye Movement
Specify: _____
[ ] Hearing loss
Specify: _____
[ ] Other: _____
_____

Growth
[ ] Failure to thrive
[ ] Overgrowth
[ ] Short stature
[ ] Other: _____

Cardiac
[ ] Atrial septal defect
[ ] Coarctation of the aorta
[ ] Tetralogy of Fallot
[ ] Ventricular septal defect
[ ] Other: _____

Craniofacial
[ ] Cleft lip
[ ] Cleft palate
[ ] Coloboma of eye
[ ] Craniosynotosis
[ ] Dysmorphic facial features
Specify: _____
[ ] Ear malformation
Specify: _____
[ ] Macrocephaly
[ ] Microcephaly
[ ] Other: _____

Cutaneous
[ ] Hyperpigmentation
[ ] Hypopigmentation
[ ] Other: _____

Gastroinstestinal
[ ] Anal atresia/imperforate anus
[ ] Gastroschisis
[ ] Omphalocele
[ ] Pyloric stenosis
[ ] Tracheoesophageal fistula
[ ] Other: _____

Genitourinary
[ ] Ambiguous genitalia
[ ] Cryptorchidism
[ ] Hydronephrosis
[ ] Hypospadias
[ ] Kidney malformation
Specify: _____
[ ] Other: _____

Musculoskeletal
[ ] Club foot
[ ] Contractures
[ ] Diaphragmatic hernia
[ ] Limb anomaly
Specify: _____
[ ] Polydactyly
Specify: _____
[ ] Syndactyly
Specify: _____
[ ] Vertebral anomaly
Specify: _____
[ ] Other: _____

Neurological
[ ] Cerebral palsy
[ ] Encephalopathy
[ ] Hypertonia/ spasticity
[ ] Hypotonia
[ ] Seizures
[ ] Structural brain anomaly
Specify: _____
[ ] Other: _____

All DNA tests are developed "in-house", pending registration with HSA.
For after office hour queries on DNA tests, please contact Dr H Y Law or Dr Y M Tan through operator.

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KK Women's and Children's Hospital
SingHealth

DNA Diagnostic & Research Laboratory
Genetics Service

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CONSENT FOR CHROMOSOME MICROARRAY ANALYSIS (CMA)

Regarding the test

I have discussed the Microarray test with my / my child's doctor.

I have read the Patient Information Sheet.

I understand that this is a specialised genetic test that may help to explain the medical condition I / my child have / has.

Regarding the results

I understand that the findings from the test may be inconclusive in some cases and blood sample from parents may be requested to help clarify the results.

I understand that the test may reveal non-paternity, meaning that the stated father is not the biological father.

I understand that results are interpreted using current medical knowledge. Future findings may change how the results are interpreted.

I understand that the test may provide unexpected findings that could be clinically significant.

I understand that the results are strictly confidential and will not be released to anyone other than my / my child's doctor without my consent.

I understand that some individuals who are found to have genetic changes leading to disease may experience discrimination (insurance, employment and / or social).

I give consent for my and /or my child's samples to be used for medical education or test validation after personal identification is removed.

Signature _____
Date _____

If person signing consent is the parent/guardian:
Name _____
NRIC No. _____
Relationship to patient _____

Doctor/nurse taking consent

Signature _____
Name _____
Designation _____

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PATIENT INFORMATION SHEET: CHROMOSOME MICROARRAY ANALYSIS (CMA)

What is Chromosome Microarray Analysis (CMA)?

Human bodies are made up of millions of cells. Each cell contains our genetic material, or DNA. The complete set of all our genetic material is called our genome. Chromosome microarray analysis (CMA) is a genetic test that looks for extra or missing DNA materials in a person's genome.

There are usually two copies of each gene in our cells. If a person has more than two, or less than two, copies of a particular gene, this may result in problems such as the functioning and development of that individual.

What are the benefits of CMA?

CMA has greater sensitivity than the conventional chromosomal analysis. It can detect abnormalities that would not have been previously identified from chromosomal analysis. If an abnormality is identified, the results may provide important information that can be used to guide medical decisions and/or future reproductive decisions.

W**hen is CMA ordered?**

CMA is commonly ordered for individuals with any clinical features that may be caused by chromosome imbalances, e.g. developmental delay/intellectual disability, unusual physical features, autism spectrum disorder, multiple congenital anomalies, etc.

What is needed for CMA?

A small sample of blood will be obtained and submitted for analysis. Blood from both parents may also be requested.

Does CMA test for everything?

Although microarray is a very sensitive test, it only detects changes in the number of DNA copies. It cannot detect many other conditions in which there is a change, or a mutation, in a gene, but the number of DNA copies remains the same.

What are some of the issues that may be associated with having CMA?

Because CMA looks at almost the whole genome, it can generate a lot of data from one test. Although most of this information is useful, the data can be difficult to interpret, and can lead to results that are of uncertain or unknown significance. Some families find it difficult to deal with these results.

Sometimes, CMA testing may also give rise to unexpected results, particularly for conditions that appear later in life, which a person may not want to know about.

If you would like genetic counselling before agreeing to CMA, please let your doctor know.