Foundation for Research in Genetics & Endocrinology (FRIGE: E – 13237) ISO 9001:2008

INSTITUTE OF HUMAN GENETICS
GENETICS CENTRE Reg. No. : 952

FRIGE HOUSE, Jodhpur Gam Road, Satellite, Ahmedabad-380015, Gujarat, INDIA

Array CGH in Prenatal cases

Sample requirement: Chorionic Villus (CV), Amniotic Fluid (AF), Abortus
Structural fetal abnormalities identified by ultrasound scan (including NT >3mm)

Contact Details

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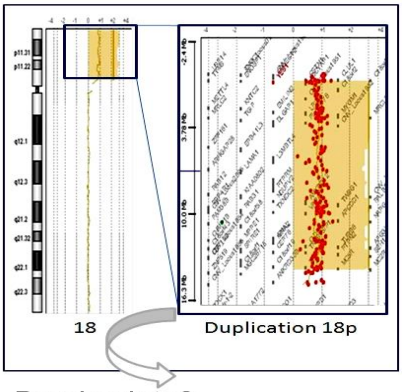
Sample required

- 15-20 ml amniotic fluid or 12 mg chorionic villus biopsy in transport medium
- A completed request card should accompany all samples.
- Ultrasound findings including NT measurement and gestation must be provided in order to allow clinical interpretation of the microarray

Patient details

To facilitate accurate testing and reporting please provide patient demographic details (full name, date of birth, address and ethnic origin), details of any relevant family history and full contact details for the referring clinician.



Breakpoints?
Mechanism of formation?
Phenotypic consequences?

Introduction

Genome-wide chromosomal microarray (CMA) detects pathogenic copy number variants (pCNV) in 15-20% of patients with developmental delay, intellectual disability or congenital abnormalities.

CMA may also detect copy number variants of uncertain clinical significance which may require parental follow up testing to aid interpretation.

CMA may also detect pCNVs which are not associated with the presenting phenotype (incidental findings)

Referrals

- Samples from pregnancies that have structural abnormalities identified by ultrasound scan where rapid trisomy testing is negative. Note that absent nasal bone and isolated IUGR are not classified as structural fetal abnormalities
- NT>3mm (before 14 weeks' gestation) where rapid trisomy testing is negative

Prenatal Reporting

pCNV identified by the microarray are reported as pathogenic if they are associated with fetal scan abnormalities or with fully prenatal intellectual disability

In line with RCPath guidelines, CNV which are associated with susceptibility to neurodevelopmental disorders (variable penetrance) are reported only if there is a published risk of fetal structural abnormality.

CNV of uncertain clinical significance are reviewed by a panel of Clinical Geneticist and Clinical Scientists and will only be reported if they require segregation analysis (parental carrier status) to aid interpretation of the clinical significance

pCNV identified by microarray are confirmed using karyotype, qPCR or Fluorescence In Situ Hybridization (FISH). These targeted tests are then applied to family members for carrier testing and for prenatal analysis in future pregnancies. Please contact the laboratory for further details.

Service offered

Whole genome microarray analysis at a practical resolution of 200kb for copy number variants (chromosomal deletions and duplications) and additional SNP based identification of uniparental isodisomy and ploidy level changes.

Technical

Whole genome chromosomal microarray analysis using the Affymetrix 750k microarray is performed on DNA extracted from EDTA venous blood. The microarray design uses both single nucleotide polymorphic (SNP) probes and non-polymorphic probes to enable consistent genomic coverage. Copy Number variations (CNV) and regions with absence of heterozygosity (AOH) are identified using infoQuant Fusion software. The estimated practical resolution is 200 kb; CNVs below this threshold may not be identified. The microarray will not detect balanced structural chromosome abnormalities and may not detect mosaicism. AOH may indicate uniparental isodisomy or regions identical by descent. The CNV identified by the CMA are compared to databases of known genetic variation and to reports of known pathogenic changes. Variants which are not known to have a pathogenic effect or do not have a high risk of pathogenicity may not be reported. AOH of non-imprinted chromosomal regions will not be reported.

pCNVs identified by microarray are confirmed using karyotype, qPCR or Fluorescence In Situ Hybridization (FISH). These targeted tests are then applied to family members for carrier testing and for prenatal analysis in future pregnancies. Please contact the laboratory for further details.

If pCNV or variants of unknown significance are reported, further samples may be requested (EDTA venous blood and/or lithium heparin venous blood from parents) to aid clinical interpretation and provide a recurrence risk for the family.

Target reporting time

Routine analysis – 3 to 4 wks

Cost: Rs 15,000/

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Recognized as Research Organization (SIRO)
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