مختبر فرايبورج الطبي الشرق الأوسط (ذ.م.م)
FREIBURG MEDICAL LABORATORY MIDDLE EAST (L.L.C.)

Comparative Genomic Hybridization (CGH) at FML
Detection of genomic aberrations

Genomic imbalance and DNA copy number variation are major causes of developmental disorders. Freiburg Medical Laboratory now offers the improved array Comparative Genomic Hybridization (array-CGH). In this test the patient's entire genome is compared to the genomes of healthy control subjects. This array is very sensitive in detecting DNA copy number variations and other alterations on the entire genome. These alterations can consist of hundreds to several millions of basepairs by addition or deletion of certain regions on the chromosome or of entire chromosomes. Detecting these changes opens new possibilities for finding the underlying causes of many developmental and medical problems, which is very helpful in tumor cytogenetics and postnatal analyses. Sub-microscopic micro-deletions and micro-duplications are often the cause for syndromes associated with malformations and mental retardation. The diagnostic value in prenatal disorders as well as in premature ovarian failure is currently under evaluation.

This new technology will not only help in clarifying unexplained cases of malformations and mental retardation but it can also provide more detailed information about already detected structural chromosomal disruptions, enabling a more precise genotype-phenotype correlation. A previous cytogenetic result can be defined more precisely, especially when it comes to structural chromosomal aberrations. Suspected chromosomal aberrations can be confirmed or excluded. The diagnostic yield of this method is 10 – 20% based on published reports and > 99% of all so far known pathogenic abnormalities regarding deletions and duplications in cases of developmental delay and mental retardation.

This high resolution array is oligonucleotide-based, enabling a higher precision for detecting chromosomal aberrations and elucidating chromosomal imbalances. This resolution is approximately 100-200 times better than the resolution achieved in conventional chromosomal analysis. Validation of detected imbalances is done using fluorescence-in-situ-hybridization (FISH) or multiplex ligation-dependent probe amplification (MLPA).

What can you expect from the array:

- Higher resolution than chromosome analysis: detailed definition of breaks in DNA, determination of additions and deletions of DNA.
- Clarification rate of about 10% in mental retardation syndromes

In case of any questions please contact FREIBURG MEDICAL LABORATORY www.fml-dubai.com

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Order: array-CGH

Indication: mental retardation or developmental delay, autistic traits, unclear ultrasound during pregnancy, pre- and post-natal growth retardation, specific growth anomalies (such as microcephaly or macrocephaly), two or more facial dysmorphisms (such as hypertelorism, nose and/or ear anomalies), congenital anomalies (such as heart defects, hand anomalies, hypospadia), cerebral seizures, behavioral disorders.

Material: 3-5 ml fresh EDTA-blood and 3 ml Heparin-blood (for potential FISH-validation). Also potentially 3-5 ml EDTA-blood and 3 ml Heparin-blood from the parents for comparison in case an aberration is detected.

TAT: 6 - 8 weeks

Literature:

Edelmann L, Hirschhorn K. Clinical utility of array CGH for the detection of chromosomal imbalances associated with mental retardation and multiple congenital anomalies. Ann N Y Acad Sci. 2009 Jan; 1151: 157-66. Review.

Fan YS et al. Detection of pathogenic gene copy number variations in patients with mental retardation by genomewide oligonucleotide array comparative genomic hybridization. Hum Mutat. 2007 Nov; 28(11): 1124-32.

Shaikh TH. Oligonucleotide Arrays for High-resolution Analysis of Copy Number Alteration in Mental Retardation/Multiple Congenital Anomalies. Genet Med, 2007, 9(9): 617-25.

Hochstenbach R et al. Array analysis and karyotyping: workflow consequences based on a retrospective study of 36,325 patients with idiopathic developmental delay in the Netherlands. Eur J Med Genet. 2009 Jul-Aug; 52(4): 161-9.