



# Newsletter

## Al Borg Medical Laboratories 6<sup>th</sup> Newsletter

# CHROMOSOME ANALYSIS

Chromosomes contain the thousands of genes in the human genome that direct cellular processes controlling growth, development, and functioning of the human body. Gains or losses in chromosome content lead to alterations in gene function and disrupt these vital cellular processes. This can result in mental retardation or other developmental abnormalities, birth defects, and many well-characterized genetic syndromes.

This test will detect abnormalities in chromosome number (aneuploidy) e.g. Down Syndrome with an extra chromosome 21 , Edward Syndrome with an extra chromosome 18 , Patau Syndrome with an extra chromosome 13 , Klinefelters Syndrome in males with an extra chromosome X or Turner Syndrome in females with a single X. Not only will this test detect numerical aberrations but structural aberrations as well can be detected in the form of translocations (Reciprocal or Robertsonian) , deletions , duplications , or isochromosomes.

### **When should a Karyotype be ordered ?**

*Problems of early growth & development* : failure to thrive , developmental delay , dystrophic facies , multiple malformations , short stature , ambiguous genitalia , & mental retardation.

*Stillbirth & Neonatal death* : the incidence of chromosome abnormalities is much higher among stillbirths & early neonatal deaths.

*Fertility problems* : women with amenorrhea or recurrent miscarriages & men with infertility may have a chromosomal aberration leading to infertility & hence may be offered a different mode of Assisted Reproductive Techniques combined with Pre Implantation Genetic Diagnosis.

*Family History* : a known or suspected chromosome abnormality in a first degree relative is an indication for chromosome analysis under some circumstances.



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*Neoplasia* : Virtually all cancers are associated with one or more chromosome abnormalities. Chromosome analysis in the appropriate tissue , the tumor itself or the bone marrow , can provide useful diagnostic & prognostic information.

Pregnancy in woman of advanced age : There is an increased risk of chromosome abnormality in fetuses conceived by women older than about 35 years. Fetal chromosome analysis should be offered as a routine part of prenatal care in such pregnancies.