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# MICROBIOLOGICAL LABORATORY



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# Prenatal Aneuploidy

Rapid Screening for Chromosomal Aneuploidies

## Test Details

Test Name : Aneuploidy

Test Code : 16002

Sample : POC/ Amniotic Fluid/  
CVS

Methodolgy : MLPA

Report On : 4 Days

Chromosomal abnormality is found in about half of first-trimester abortions. Karyotype is the gold standard to detect Chromosomal Abnormalities. **Multiplex Ligation-dependent Probe Amplification (MLPA)** offers advantage over karyotype in terms of lower failure rate, faster turnaround time, and much higher resolution than conventional karyotyping and found to be 98% concordant with conventional karyotype.

Detectable Aneuploidies with Frequency

<b>Patau Syndrome</b> (47, XY, +13)	<b>1 in 5000</b> live-born infants	<b>Down Syndrome</b>	<b>1 in 2500</b> newborn GIRLS	<b>Triple X Syndrome</b>	<b>1 in 500</b> newborn BOYS	<b>Triploidy Syndrome</b>	<b>1 in 1000</b> MALE Births
<b>1 in 10000</b> live-born infants	<b>Edward Syndrome</b>	<b>1 in 800</b> new borns	<b>Turner Syndrome</b>	<b>1 in 1000</b> newborn GIRLS	<b>Klinefelter Syndrome</b>	<b>1 in 1000</b> newborn BOYS	<b>XYY Syndrome</b>

## Detailed Detection

Detection of aberrant copy number of 50 genomic DNA/RNA sequences in a single, PCR-based reaction.

## Accurate Region

MLPA determine the methylation status of imprinted and promotor regions and also detect known point mutations and SNP's

## Also Used on Degraded DNA

MLPA can also used on partially degraded DNA, such as DNA extracted from paraffin imbedded, formalin treated tissues.