PRENATAL BoBs™
IMPROVED DETECTION IN PRENATAL DIAGNOSIS

Prenatal BoBs™
Part of the BACs-on-Beads™ product family
Prenatal BoBs™ is a CE marked IVD test/assay based on PerkinElmer’s BACs-on-Beads™ technology. In addition to detecting copy number changes of chromosomes 21, 18, 13, X and Y, the product enables detection of 9 additional chromosomal regions in which a clear correlation between a loss and an adverse outcome has been demonstrated.¹

Copy number changes in these targeted microdeletion regions are not easily found with other commonly used methods. Also the microdeletion syndromes detected by Prenatal BoBs™ often are not inherited and do not display ultrasound abnormalities, so they may otherwise be missed in a prenatal setting.²

How BACs-on-Beads™ technology works?

1. The assay involves colored beads wherein each bead is uniquely colored; the colors are obtained using different proportions of two dyes, red and infra-red.

2. Each bead is coated with a probe derived from the region of interest i.e. a region where detection of a gain or a loss of genomic material is required.

3. The information carried is like that on a microarray, but the medium is readily suspendable in liquid this enables more efficient hybridization, high throughput and cost efficient molecular karyotyping.

4. The sample DNA is labelled with Biotin. The BACs-on-Beads Bead Mix is added to the sample upon which target specific hybridization occurs. Following hybridization a fluorescent phyco-erythrin reporter tag is added.

5. The bead then passes in single file by two lasers - a red laser excites the dye within the bead, classifies each bead by color and thus identifies the target. The green laser excites the phyco-erythrin tag to quantify the amount of sample DNA hybridized to the beads-probes.

6. The amount of target bound to each bead type is then compared to the amount of target bound in reference samples. The results are showed as ratio plots indicating where a gain or a loss of genomic material has occurred.
Advantages of Prenatal BoBs™

- Low specimen is required. Analysis using Prenatal BoBs™ can be performed with 50-250 ng of genomic DNA, an amount which can be obtained from 5 mL of amniotic fluid or from 10 mg villus.

- Most common aneuploidies and common 9 microdeletion syndromes can be detected by Prenatal BoBs™. There are no unknown clinical significance results as there is a clear genotype-phenotype correlation for the microdeletion syndromes detected by Prenatal BoBs™.6

- Prenatal BoBs™ has high sensitivity and specificity (upto 99% detection rate with false positive <1%; false negative rate of <2%).4

- Prenatal BoBs™ has not only a turnaround time similar to FISH but also provides additional information regarding the microdeletion syndrome.4

- Less expensive than microarray and does not identifies deletions / duplications / other abnormalities of unknown pathogenic significance.

Where is Prenatal BoBs™ recommended?

- Pregnancies with positive screening via conventional screening (FTS, Triple, QD etc).

- Ultrasound findings of increased Nuchal Translucency (NT).

- Previous family history of aneuploidies and microdeletions which BoBs™ can detect.
Limitations of Prenatal BoBs™

- Prenatal BoBs™ cannot detect polyploidies.
- Only gross changes in the chromosome may be detected whereas other alteration that may give rise to the syndrome may be missed.
- Cannot detect gains and losses in the regions which are not covered by the assay.
- Point mutation, balanced rearrangement (inversion and translocations) cannot be detected.
- The technique cannot detect all mosaic cases (mosaicism detection capability is region dependent).
- Maternal cell contamination may give erroneous result.

GLOSSARY OF TERMS
• RATs- Rapid Aneuploidy Test
• QF-PCR- Quantitative Fluorescence Polymerase Chain Reaction
• FISH- Fluorescence in Situ Hybridization
• MLPA- Multiplex Ligation-Dependent Probe Amplification
• CMA- Chromosomal Microarray- based Analysis
• Array (CGH)- Array Comparative Genomic Hybridization
• VOUS- Variants of Uncertain Clinical Significance

REFERENCES:
1 OMIM: http://www.ncbi.nlm.nih.gov/omim/ #105830, #123450, #176270, #182290, #188400, #194050, #194190, and #247200
2 PKI Brochure: 1244-9963-05, December 2012
5 Shaffer LG et al., The development of a rapid assay for prenatal testing of common aneuploidies and microdeletion syndromes, Prenat Diagn. 2011 Aug;31(8):778-87. Signature development of extended panel using BoBs technology
6 PerkinElmer Brochure 1244-1306-11, June 2013

SEE ALSO:
• Vialard F et al., Prenatal BACs-on-Beads: the prospective experience of five prenatal diagnosis laboratories, Prenat Diagn. 2012 Apr;32(4):329-335. Five laboratories with 1653 samples‘ Prenatal BoBs assay combines a short turn-around-time with valuable detection of the most frequent microdeletion syndromes that cannot be detected in cytogenetic analyses’

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