

July
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Newsletter for the Genetic Screening
and Clinical Diagnostics Laboratory



Well & Healthy



FOR THE BETTER

HOW PERKINELMER IS WORKING TO PROTECT THE HEALTH OF EXPECTANT MOTHERS, BABIES AND FAMILIES

PerkinElmer touches over 27 million pregnancies and/or births every year. As well as being the number one supplier of neonatal screening systems for the detection of inherited metabolic disorders, it is also the global leader in first trimester prenatal screening. In addition PerkinElmer is the second biggest provider of umbilical cord blood collection and storage services. The stored blood provides the potential to treat 40 life-threatening diseases. Page 2

ALSO IN THIS ISSUE: • Signature Genomic Laboratories, LLC – now part of PerkinElmer • Laboratories in Italy and France have been evaluating the new Prenatal BoBs™ product • How BACS-on-Beads™ works • Highly relevant microdeletion syndromes found by Prenatal BoBs • Dried blood spot samples benefits 1st trimester prenatal screening • Pilot UK NEQAS for first trimester Down Syndrome screening with DBS samples • Early detection plays a key role in the fight against pre-eclampsia • Aspirin therapy promising – but must be started early • DELFIA® Xpress / Viewpoint link streamlining data transfer at Kings College Hospital

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Strong growth in maternal and fetal health

The company has a strong growth record. In prenatal screening, for example, PerkinElmer developed its first products in the early 90's, but now has more than 400 customers in over 50 countries. The customers range from state (State of California, 400,000 screens) and national (The Netherlands) screening programs to OSCAR clinics. Through its acquisition of NTD Laboratories in 2006, PerkinElmer was able to improve its service portfolio. 75% of women that receive first trimester screening in the USA are screened by NTD.

PerkinElmer has developed its knowledge base by engaging in research collaborations with leaders in their fields, such as Professor Howard Cuckle, Professor Kypros Nicolaides and Professor Justin Konje. Such collaborations provide a background for PerkinElmer's work to introduce pre-eclampsia screening and develop markers for pre-term birth.

New products coming

New products and developments are constantly in the pipeline. For example, the new generation dried blood spot (DBS) DELFIA assays offer a solution for implementing prenatal screening in regions of the world for which serum-based screening is unsuitable. PerkinElmer has recently opened a service laboratory in Chennai, India. One of the services that will be provided over the whole of India will be prenatal screening using DBS samples.

A particularly fast growing area for PerkinElmer has been cytogenetics. Organic growth has been driven by a fruitful collaboration with Luminex, and the consequent emergence of the BACs-on-Beads product line. The recent acquisition of Signature Genomics demonstrates significant commitment to cytogenetics and presents an exciting opportunity to be at the forefront in developing the next generation of innovative products.



**SIGNATURE
GENOMIC
LABORATORIES, LLC
NOW PART OF
PERKINELMER**

Dr Lisa Shaffer, Co-founder and President of Signature Genomic Laboratories.

This spring, PerkinElmer acquired Signature Genomic Laboratories, a front-runner in the field of molecular cytogenetics.

Signature Genomic Laboratories was founded in 2003, by Lisa G. Shaffer, PhD and Bassem A. Bejjani, MD. It was the first laboratory to provide microarray-based cytogenetic diagnostics with its proprietary SignatureChip® and is the leader in providing microarray-based chromosome analysis for individuals with unexplained physical and developmental disabilities. Signature's microarrays are offered for both pre-natal and post-natal identification of DNA alterations associated with genetic disease. More recently, Signature launched a suite of services for the diagnosis of patients with leukemia. Signature currently employs approximately 120 staff at its headquarters in Spokane, WA, including seven American Board of Medical Genetics-certified laboratory directors and six American Board of Genetic Counseling-certified genetic counselors.

Signature's global client base includes clinical geneticists, neurologists, pediatricians, neonatologists, obstetricians, and the research community. Signature is CAP accredited, CLIA certified, and certified to ISO 9001:2008 requirements. Signature has clinical licenses from New York, California, Rhode Island and Florida.

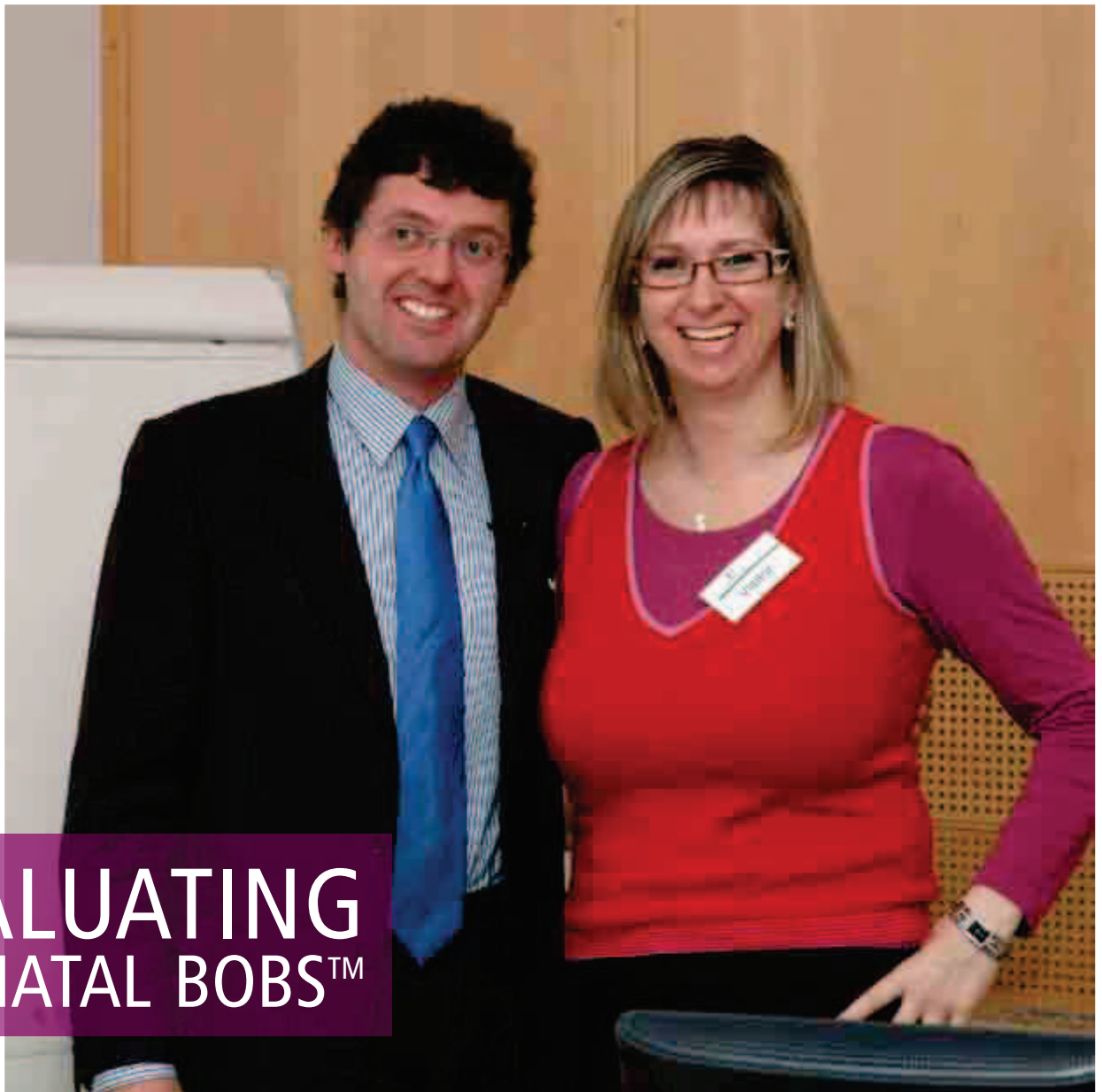
Molecular cytogenetics think-tank

A truly leading edge company, Signature will form a "think tank" for PerkinElmer. It will be responsible for generating new ideas and new products. At the same time, it will raise the knowledge and expertise in molecular cytogenetics throughout the company.

Robert Friel, chairman and chief executive officer of PerkinElmer comments, "We are delighted to have Signature become part of the PerkinElmer team. Signature brings very strong complementary assets for diagnosing disorders that are often undetected by traditional genetic tests."

"Patients, families, researchers and clinicians will benefit from our combined expertise, as accurate diagnosis is crucial to providing answers that lead to better care, treatment and understanding of patients with birth defects, behavioral issues, delay in development or learning problems."

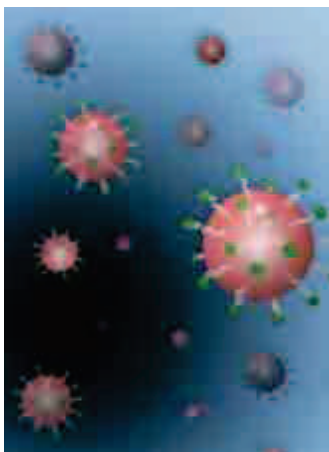
Additional information about Signature Genomic Laboratories is available at www.signaturegenomics.com.



Dr François Vialard and Dr Francesca Romana Grati recently visited PerkinElmer's Turku, Finland facility to present results for the evaluations that were performed at their laboratories in France and Italy respectively.

EVALUATING PRENATAL BOBS™

Laboratories in France and Italy report on their work



Prenatal BoBs™ is a new PerkinElmer product that detects aneuploidies of chromosomes 13, 18, 21, X and Y, as well as gains and losses in 9 common microdeletion syndrome regions. The syndromes show high genotype-phenotype correlation thus avoiding answers of unknown clinical significance. The product belongs to the BACs-on-Beads™ product family utilizing a multiplexing technology enabling molecular karyotyping in a well (see side article on page 4).

Prenatal BoBs belongs to the category of rapid prenatal tests. Similar to other rapid prenatal testing methods such as QF-PCR and FISH, the Prenatal BoBs assay detects common aneuploidies. As the product also focuses on microdeletions causative of severe constitutional disorders it provides more actionable information. Prenatal BoBs can therefore be used for the same indications as QF-PCR and FISH, however it is also suitable for settings in which a more thorough genomic view is requested.

Laboratories in Italy and France have recently reported on their evaluations of the product.

The Italian experience

Dr Francesca Romana Grati is head of the Research & Development Unit at TOMA Advanced Biomedical Assays, S.p.A., Busto Arsizio, Varese, Italy. At the TOMA laboratory, all commonly applied prenatal diagnostics methods are used. In the opinion of Dr Grati, each of them has limitations that set the type of analysis for which the method can be used.

In evaluating Prenatal BoBs the laboratory worked with 255 samples. These represented a wide range of different specimen types

including both fresh and cultured amniotic fluid (AF) and chorionic villi sampling (CVS) specimens. Furthermore, the samples showed different concentrations and quality measures, and had been processed differently with regard to extraction and storage. 68 of the samples had known abnormal karyotypes.

Results

- Samples with a normal male or female karyotype were classified correctly
- Aneuploidies and partial unbalances for regions included in the BoBs kit were all detected
- Microdeletions were identified correctly
- There were no false positive results
- False negative calls were related to 69,XXX polyploidy. Also, one case showing 48,XXXY was not identified.

Observations

The analyses could be performed successfully on samples differing widely in terms of DNA amount, concentration and quality. Results could be obtained on DNA showing relatively low DNA quality or concentrations.

It takes less than 24 hours to perform the Prenatal BoBs assay. Results are therefore obtained after 30 hours from receiving the sample.

The results are robust due to the comparison to both male and female references