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## Press Release

### NEW BLUEFUSE OFFERS AUTOMATED ARRAYCGH MAPPING

#### New developments will bring the power of arrayCGH to the clinician

ArrayCGH is a breakthrough technique in human genetics, which is attracting interest from clinicians working in fields as diverse as cancer and IVF. Its widespread adoption as a diagnostic tool has taken a major step forward with the latest release of 'BlueFuse for Microarrays' from BlueGnome, the Cambridge based, specialist bioinformatics company.

ArrayCGH is a specific, sensitive and fast technique that enables the screening of the whole genome in a single test, it will facilitate and accelerate the diagnostic process in human genetics and is expected to have a profound impact on the screening and counselling of patients with genetic disorders.

ArrayCGH is used to identify areas of chromosomes, which exhibit amplifications and deletions in the genomic sequence. It offers a fundamental benefit over conventional CGH - which is well proven in research - improved resolution. It is now possible to identify the exact location on the chromosome where an aberration has occurred and it is possible to map these changes directly onto the genomic sequence.

Despite its many benefits arrayCGH has until now been difficult to scale up for use as a routine diagnostic test and the data analysis has been considered too subjective for patient facing applications.

The new release of the BlueFuse technology has been developed in close collaboration with major arrayCGH labs in both Europe and the US. It is the first commercial product to offer fully integrated and automated analysis of arrayCGH experiments - from microarray image to ideogram - improving both the quality and confidence in the data.

As Nick Haan, CEO BlueGnome, comments: "Full tiling path arrays of 30k spots potentially allow the presence of every human gene to be monitored and qualified simultaneously. Analysing this data presents a series of new mathematical challenges.

Mathematicians working with clinical groups in the major academic centres are already publishing novel copy number detection algorithms however these are generally highly optimised for specific patterns of genetic abnormality. BlueFuse provides a framework by which the most promising of these algorithms may be made available to the broader research community."

The new release fully exploits the capability of BlueGnome's unique statistical approach to overcome the major concern about subjectivity in microarray experiments. Traditionally the operator was required to set thresholds to distinguish between valid biological signal and extraneous experimental noise. BlueFuse technology addresses this fundamental problem by using knowledge of how the signal and noise are generated to distinguish between the two without the need for operator intervention. On this basis BlueFuse is able to automatically analyse 20 or more arrays, returning to the operator a list of regions of copy number change for each experiment in the batch.

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## Case-study: New clinical tool for genetic screening

The microarray core facility at the VU Medical Centre in Amsterdam has pioneered the use of arrayCGH using synthetically engineering oligo libraries. Most array CGH experiments spot BACs (bacterial artificial chromosomes) spaced at 1Mb intervals along the genome. While relatively straight forward to produce in small numbers amplifying sufficient quantities of the 30,000 BACs required for widespread adoption of a full tiling path array remains a significant challenge. Ready availability of oligos appears to provide one possible solution. However, the reduced signal and higher noise inherent in oligo based experiments moves the bottleneck from production to analysis. In order to address this analysis bottleneck the VU Medical Centre has licensed BlueFuse for Microarrays.

The head of the microarray facility, Dr Bauke Ylstra, explains: "Our facility is funded to provide diagnostic tools that improve patient outcome. In order for clinicians to act on our results it is critical that they be of the highest quality and full repeatable. By implementing BlueFuse as an integral part of our high throughput microarray process we have removed operator subjectivity and therefore deliver the highest levels of experimental repeatability. As part of our extensive validation of BlueFuse we confirmed that the technology is able to increase the amount of usable data recovered from each experiment from 83% to nearly 100%. Finally by combining the availability of oligos with automated analysis we are able to plan a scale of experimentation that would not previously have been considered. In a research environment these improvements would have been very exciting; in a clinical context they are fundamental."

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