

Product Focus: Cancer genome

Exposing the Changes

The latest products and services

Cancer genomes are currently being studied at the genomic, epigenomic and transcriptomic levels. In addition to revealing the range of oncogenic mutations and mutagenic influences, data from these studies help in defining clinically relevant subtypes for prognosis and therapeutic management, as well as enabling the development of new cancer therapies. Some of the latest products, including human cancer models, a novel solution for detecting genetic copy number variations (CNVs), updates to single nucleotide polymorphism (SNP) primer checking tools and microarrays are highlighted. Additionally, details of several services that facilitate cancer research and diagnosis are outlined.

Editing the genome of human cells, to create genetically-defined human disease models, has historically proved far more challenging than in mice. However, **Horizon Discovery** is now providing a solution to this problem using '**GENESIS**'; a gene editing platform that uses the power of recombinant adenovirus-associated virus (rAAV) vectors to perform efficient and error free targeted homologous recombination in mammalian somatic cells. Horizon has assembled over 300 defined and matched pairs of 'X-MAN' (Mutant And Normal) human cancer models, covering key therapeutic targets, such as K-Ras, PI3K, B-Raf and p53, which are now being used by academic and industrial researchers to define how they contribute to disease onset or progression and accelerate the development of more rational targeted therapies, clinical trials and companion diagnostics to support the emerging concept of 'precision' or 'personalised' cancer therapy.

A novel solution for detecting genetic CNVs associated with disease susceptibility, drug response and cancer progression has been launched by **NanoString Technologies, Inc.** The **nCounter® Copy Number Variation CodeSets** enable researchers to interrogate

up to 800 regions of the human genome in a single multiplexed reaction with the least hands-on time of any CNV platform. Researchers previously had no simple, precise, and scalable technology for technically replicating CNV detection or validating CNV correlation with biological processes. The nCounter Copy Number Variation CodeSets, built on the same innovative digital technology underlying NanoString's gene expression and miRNA assays, enable researchers to perform the functional equivalent of 9,600 qPCR reactions (800 regions across 12 samples) with only 25 minutes of hands-on time. Unlike PCR or arrays, the nCounter Analysis System does not rely on analogue signal output or amplification of target molecules. Instead, the system utilises a digital quantification technology that offers superior reproducibility and generates highly accurate data with a linear response to increasing copy numbers. These advantages also make it possible for data generated from multiple sites or studies to be combined for further analysis, thus facilitating multisite studies or comparisons of old and new data sets.

Microarrays are widely used to screen human DNA for abnormalities associated with developmental delay, dysmorphia and other genetic diseases, where the abnormality is carried by every cell in the body. Their use for cancer research is however more challenging as the genetic changes associated with cancer may be different in every cell and vary depending upon the type of cancer, its stage of development, the age and genotype of the patient and exactly which cells are used in the investigation. **BlueGnome's** new cancer platform, **CytoChip Cancer**, combines the cancer gene targeted array design with the company's leading BlueFuse database and analysis software. As CEO Nick Haan explained, "BlueFuse is used by microarray laboratories in over 30 countries because it generates results fully automatically and stores them in a single



Horizon Discovery's X-MAN human cancer cell lines

database, together with the experimental conditions and the latest information on how abnormalities might be linked to the underlying disease. By combining these capabilities with the new cancer design we believe we can give cancer research a major boost by standardising results and interpretation within our customer laboratories and then providing facilities to share data with other laboratories."

A new microarray platform, the **SurePrint G3 Human CGH+SNP**, is available from **Agilent** for studying the genetic basis of cancer and other developmental disorders. This microarray platform is an innovative system for simultaneous analysis of chromosomal copy number changes and copy-neutral aberrations. The system combines CGH and SNP assays on the same microarray, creating a unique cytogenetics tool, allowing researchers to study the genetic basis of developmental disorders

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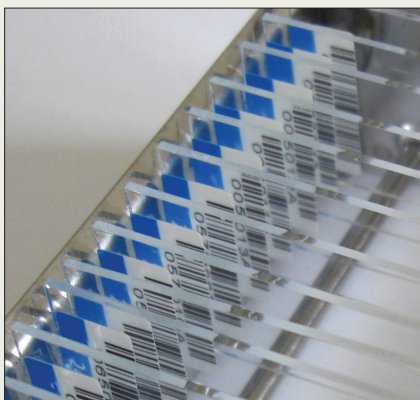
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Nanosting's nCounter Analysis System



BlueGnome's CytoChip microarrays

as well as many cancers. This is also the only two-colour CGH platform that can detect loss of heterozygosity/uniparental disomy (LOH/UPD) with 5- to 10-megabase resolution.

The **National Genetics Reference Laboratory Manchester (NGRL)** has updated its popular primer checking tool to incorporate build 132 of dbSNP. **SNPCheck** is a unique online tool that enables scientists to easily check their primer sequences for common human SNPs that may affect the hybridisation of the primer to the target DNA. A sequence divergence of only one nucleotide between primer and target DNA can produce a detectable effect on hybridisation, leading to allelic dropout and potential genotyping failure. SNPCheck is used by scientists around the world to check primers and array probes for SNPs during the design process. The tool aligns submitted primer pair sequences to the human genome reference sequence and then identifies reported SNPs in that region. This latest release of SNPCheck references dbSNP

build 132, the most up-to-date database of SNPs world-wide.

Affymetrix's unique Molecular Inversion Probe (MIP) technology enables scientists to obtain both high-quality copy number and allele data from recent and archived formalin-fixed, paraffin-embedded (FFPE) samples with low sample input. The company has now launched the **MIP Copy Number Service**, designed for cancer researchers using challenging FFPE samples. The service is available for research use only exclusively through the Affymetrix[®] Research Services Laboratory (ARSL). "Cancer researchers are unlocking the full potential of millions of archived FFPE samples using MIP copy number technology," said Kevin King, President and CEO of Affymetrix. "Leading cancer researchers can now link copy number data with clinical data from these archived resources. This is the tremendous advantage of the MIP assay and will help researchers yield discoveries that could ultimately improve

cancer diagnosis and categorisation of patients, and thus lead to more targeted and individualised strategies for the treatment of cancer."

Source BioScience's healthcare division provides cervical cancer screening and cancer diagnostic services. The company's screening division has a world-leading cloned DNA/RNA library and provides ultra fast sequencing services to academic research groups and to the global pharma industry as part of a molecular diagnostics service to support drug discovery and clinical trials. The company has a national network of laboratories which have CPA, GLP and GCP accreditations in Europe, are licensed by the Human Tissue Authority, and are approved by multiple international regulatory bodies, including the FDA. DNA sequencing and analysis is performed in-house using Source's state-of-the-art high throughput sequencing machines, including an **Illumina HiSeq2000 Genome Analyser™**. The Group has operations in the UK and Germany.

A service for the **CancerTYPE ID[®]** test, developed and performed by bioTheranostics, US is now available from **Lab21**. This test is intended to aid in classification of cancer type in patients with metastatic cancer of uncertain or unknown primary origin (CUP). The CancerTYPE ID[®] test is able to classify 54 different tumour types from formalin-fixed, paraffin embedded metastatic tumour samples. Classification of the primary cancer type will assist pathologists and oncologists in determining the most appropriate treatment for their patients. Lab21 CancerTYPE ID[®] testing has a typical turnaround time of 10-15 working days, and is available exclusively from Lab21 in the UK and Ireland.

Companies mentioned in this Product Focus

Affymetrix

www.affymetrix.com

Agilent

www.agilent.com

Blue Gnome

www.cytochip.com

Horizon Discovery

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Illumina

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Source Bioscience

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