Complementary Technologies for Precision Genetic Analysis

Agilent Technologies

Discover More with Complementary NGS, CGH and FISH Solutions

Complementary Technologies

Next Generation Sequencing Target Enrichment (NGS TE), Comparative Genomic Hybridization (CGH), and Fluorescence *In Situ* Hybridization (FISH) enable clinical researchers to accurately detect, characterize and confirm a broad spectrum of genetic aberrations. These range from point mutations to gene rearrangements and whole-genome aneuploidy.

Comprehensive Workflow Solutions

Identify genetic variants with confidence using Agilent SureSelect and HaloPlex NGS TE systems, SurePrint and GenetiSure CGH microarrays, and FISH probes. Each platform utilizes long RNA or DNA probes manufactured using Agilent's industry-leading Oligonucleotide Library Synthesis (OLS) manufacturing process to ensure high detection sensitivity, specificity and accuracy of results.

CytoGenomics and SureCall data analysis software provide guided workflows for CGH and NGS to analyze, visualize and contextualize data without the need for complex bioinformatics infrastructure. Combined with unparalleled sample QC systems and automation solutions, Agilent provides a comprehensive portfolio for confident variant identification.

One Trusted Partner

Agilent provides a comprehensive genetic analysis portfolio with premium performance you can trust, in addition to service and support resources to assure that these leading-edge solutions will meet your needs.

The Range of Genetic Aberrations Best Detected by Targeted NGS, CGH Microarrays and FISH

	Point mutations	Indels	Gene copy number changes	Balanced translocations & inversions	Other rearrangements	LOH/UPD	Whole-genome aneuploidy
NGS	✓	✓	✓	✓		✓	
CGH			✓		✓	✓	✓
FISH			✓	✓	✓		

Next Generation Sequencing Solutions

High sensitivity, targeted mutation and CNV analysis with single-base resolution

NGS target enrichment enables highly sensitive and parallel analysis of samples for efficient identification of disease-associated variants. SureSelect and HaloPlex play major roles in the identification of the involved genes by providing flexible solutions that couple expert-optimized designs with quick and easy workflows. Agilent NGS systematically addresses the need for deep target coverage, comprehensive and accurate variant calling and faster sample-to-data workflows.

Complement your NGS target enrichment with CGH for the detection or confirmation of CNVs.

Complement your NGS target enrichment with FISH for further characterization of structural rearrangements.

Comprehensive Mutation Analysis

Solutions for both comprehensive coverage of the genome (SureSelect Clinical Research Exome) and focused coverage of disease-associated regions (SureSelect Focused Exome) are compatible with desktop or high-output sequencers.

Expert-Optimized Targeted Panels

ClearSeq

ClearSeq NGS Disease Research Panels provide accurate and focused variant identification for faster time to answers.

Comprehensive Constitutional Research

OneSeq

The OneSeq Constitutional Research Panel, powered by SureSelect, identifies genome-wide copy number variants, copy-neutral LOH, point mutations and indels in one assay.

Agilent SureCall Software

- Easy to use, 3-step workflow from raw data to mutation categorization and annotation
- Richly annotated variants with information from many public sources
- Reduce time-to-results from days to minutes with existing infrastructure and hardware

Comparative Genomic Hybridization Solutions

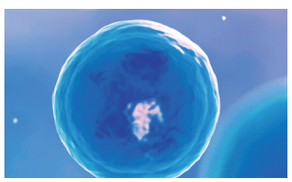
High resolution, genome-wide analysis of copy number variation and loss of heterozygosity

For cytogenetics research labs, we offer a CGH microarray solution comprised of reagents, instruments and software that enables accurate detection of genomic aberrations. Agilent custom and catalog CGH microarrays provide exceptional sensitivity and flexibility and are optimized for the most challenging sample types, including single cells, amniotic fluid and buccal swabs.

FISH — Complement your microarray with FISH for deeper characterization of an aberration.

NGS — Complement your microarray with NGS target enrichment for the detection of single base pair mutations and indels.

Single Cell



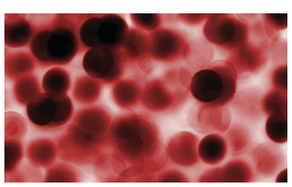
CGH microarrays designed for single-cell analysis deliver rapid detection of aneuploidy in embryos using the GenetiSure Pre-Screen Kit.

GenetiSure

Amniotic Fluid & CVS

Several CGH array content options facilitate research using amniotic fluid and Chorionic Villus Sampling (CVS) samples, including International Standard Cytogenetic Array (ISCA)-designed and Baylor College of Medicine microarrays.

Buccal Swabs & Blood



A wide selection of arrays for buccal swab and blood samples provides content specific for research involving developmental delay and intellectual disability.

Agilent CytoGenomics Software

- Designed specifically to put data into biological context
- Accurate detection of copy-number changes and copy-neutral variations, including LOH and UPD
- Streamlined, automation-enabled workflow for data upload and analysis

Fluorescence *In Situ* Hybridization Solutions

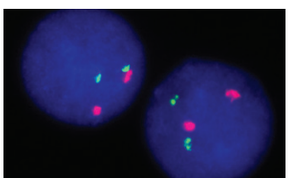
Accurate identification of structural rearrangements and copy number changes at critical loci

While synthetic DNA is widely used for CGH and NGS platforms, FISH still employs fragmented human DNA from Bacterial Artificial Chromosome (BAC) clones as probes to a large extent. Agilent is the only comprehensive DNA FISH supplier that provides synthetic, oligo-based FISH probes. These probes enable customers to spend less time at the microscope, due to high probe specificity and quality.

Quickly confirm your microarray findings with a copy number FISH probe that targets the exact region of interest.

Complement your NGS target enrichment with FISH for further characterization of structural rearrangements.

High Signal-to-Noise Ratio



Agilent oligo probes are free of repetitive DNA sequences, reducing hybridization background. No Cot-1 blocking agent is required, minimizing overall signal suppression that can plague BAC FISH probes.

High Probe Specificity

In silico design enables probe coverage with base pair level precision. This translates into high probe specificity, of particular importance for the detection of small deletions.

High Lot-to-Lot Consistency

Synthetic oligo probe production using industry-leading OLS technology completely eliminates the biological bacterial clone production process, which can cause significant lot-to-lot variation.

IQFISH Workflow: Fastest Time to Result!

- Reduce hybridization time from overnight to 90 minutes
- Go from sample to result in less than 4 hours

CUSTOM

Your Vision. Your Design. Endless Possibilities.

The ability to answer significant genomics questions requires rapid iteration of NGS, CGH or FISH designs to progress from general to more focused approaches, using automation and multiple technologies for validation. Agilent's custom program provides the flexibility and experience to manufacture products to meet specific needs on a proven platform that will maintain high-quality and consistent data.

NGS

Leverage optimized probes, upload your own, or design *de novo* probes to create a custom panel. Use as a standalone or library blend with the OneSeq CNV backbone.

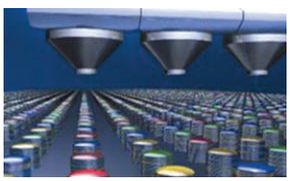
CGH

Select from a database of optimized probes or design your own to create a custom microarray.

FISH

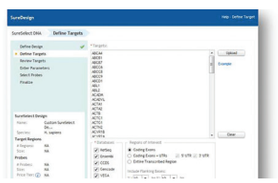
Harness the power of *in silico* oligo design and QC to create high-quality custom FISH probes targeting precise loci of interest.

Quality



Agilent custom genomic products are manufactured using the same patented Oligonucleotide Library Synthesis (OLS) platform as our catalog products to assure the highest quality in the industry.

Flexibility



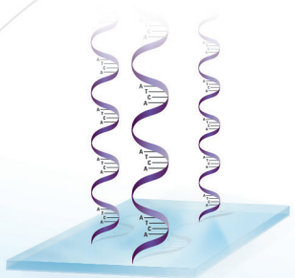
The free SureDesign online application enables rapid creation of arrays, NGS target enrichment libraries and FISH probes to meet experimental requirements.

Experience

Agilent custom probe synthesis is backed by over a decade of customer support experience, thousands of publications and highly knowledgeable Agilent genomics experts.

Agilent SureDesign Software

- Choose from pre-designed content or your own
- Review Agilent catalog and custom designs for purchase
- Collaborate within your workgroup or with other researchers



Agilent OLS Technology

Parallel synthesis of long oligonucleotides

Agilent produces some of the longest oligo libraries in the industry, including 120-mer NGS libraries, 60-mer arrays, and 150+mer FISH probes. Longer oligos produce brighter array signals, better unbiased target enrichment for SNP/indels, and more specific FISH hybridizations.

Complementary NGS, CGH and FISH Workflow

Featured Publication

Zhu, J. *et al*. Duplication of C7orf58, WNT16 and FAM3C in an obese female with a t(7;22)(q32.1;q11.2) chromosomal translocation and clinical features resembling Coffin-Siris Syndrome (2012) PLoS ONE 8(9): 10.1371.

Study Objective

Molecular characterization of the genetic variation underlying a phenotype resembling Coffin-Siris Syndrome.

Approaches Used in Publication

Workflow

Genetic Analysis	Screening for Deleterious Mutations	Genome-wide analysis of structural variants	Mapping of translocation breakpoint and duplicated region	Identification of potentially causative mutations
Technology	Exome sequencing using SureSelect Human All Exon	244K CGH microarray	FISH	Sanger sequencing of exons and splice junctional regions from C7orf58, WNT16 and FAM3C
Result	None identified	Chromosome 7 duplication that included the tail end of an uncharacterized gene termed C7orf58 and spanned the entire WNT16 and FAM3C genes	The duplicated region and all three genes in it were located on both derivative chromosomes 7 and 22	SNPs identified in the three genes that could augment the detrimental effect of the duplication

Other Publications Using These Complementary Technologies

1. Askree SH, *et al*. Detection limit of intragenic deletions with targeted array comparative genomic hybridization. BMC Genetics (2013) 14:116.

2. Nectoux J, *et al*. Detection of TRIM32 deletions in LGMD patients analyzed by a combined strategy of CGH array and massively parallel sequencing. European Journal of Human Genetics (2014) doi:10.1038/ejhg.2014.223.

3. Schluth-Bolard C, *et al*. Breakpoint mapping by next generation sequencing reveals causative gene disruption in patients carrying apparently balanced chromosome rearrangements with intellectual deficiency and/or congenital malformations. J Med Genet. (2013) 3:144-50. doi: 10.1136/jmedgenet-2012-101351.

4. Carvalho CM, *et al*. Inverted genomic segments and complex triplication rearrangements are mediated by inverted repeats in the human genome. Nature Genetics (2011) 43: 1074–1081 doi:10.1038/ng.944.

5. Kang SH, *et al*. Insertional translocation detected using FISH confirmation of array-comparative genomic hybridization (aCGH) results. Am J Med Genet A. (2010) 152A:1111-26. doi: 10.1002/ajmg.a.33278.

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