CytoSure

A Sysmex Group Company

Medical Research Exome Array

Features

Highly targeted optimised probes

- Detect single or multiple exonic CNVs

Medical research relevant content

- Over 4600 hand-curated, research-validated genes

Bespoke custom array design service

- Select from our proprietary Medical Research Exome database

Optimised labelling kits and integrated sample tracking probes

- For confident analysis and reporting

Complete coverage of all medically relevant genes at exon-level resolution

The CytoSure® Medical Research Exome Array is a highly targeted exon-focussed array capable of detecting medically relevant microdeletions and microduplications. This array has been developed in collaboration with leading molecular genetics experts at Emory University and makes an ideal complement to an exome sequencing approach, providing a comprehensive mutation spectrum analysis in rare disease.

Highly targeted optimised probes

Using a proprietary probe design algorithm, we are able to design highly targeted, optimised probes throughout the majority of the genome. A range of probe performance metrics are evaluated to ensure optimal array performance. A poorly performing probe can result in inaccurate data and even false calls. All of our probes are first tested in silico and scored on quality. Probes with the highest score are printed on an array and tested in the laboratory. These probes are ranked on performance, and only the most accurate, best-performing probes are used in the final design. Probes have been selected to target the exonic regions of 4,645 genes. For the majority of genes there are a minimum of 4 probes per exon. For very large exons, probes are distributed evenly along the exon with 1 probe every 125bp. For any array design, it is also important to provide good backbone coverage to ensure accurate normalisation. In the untargeted backbone, the CytoSure Medical Research Exome array has one probe every 42kb. Using optimised probes enables the detection of small amplifications and deletions (Figure 1).

Medical research relevant content

Working with the leading clinical research experts at Emory University has enabled us to design an array covering the most medically relevant regions of the genome gathered from their research into molecular disorders. It has been hand-curated and grouped into disease- and syndromespecific panels. The content of the CytoSure Medical Research Exome Array is grouped into over sixty different panels. Each panel contains genes relevant to a specific syndrome or genetically linked disease. Working with these panels has enabled Emory Genetics to create a set of ten 4x180k custom arrays containing probes validated on the CytoSure Medical Research Exome Array. These custom arrays provide a more cost-effective, routine option for detailed research into small, exon-specific aberrations.

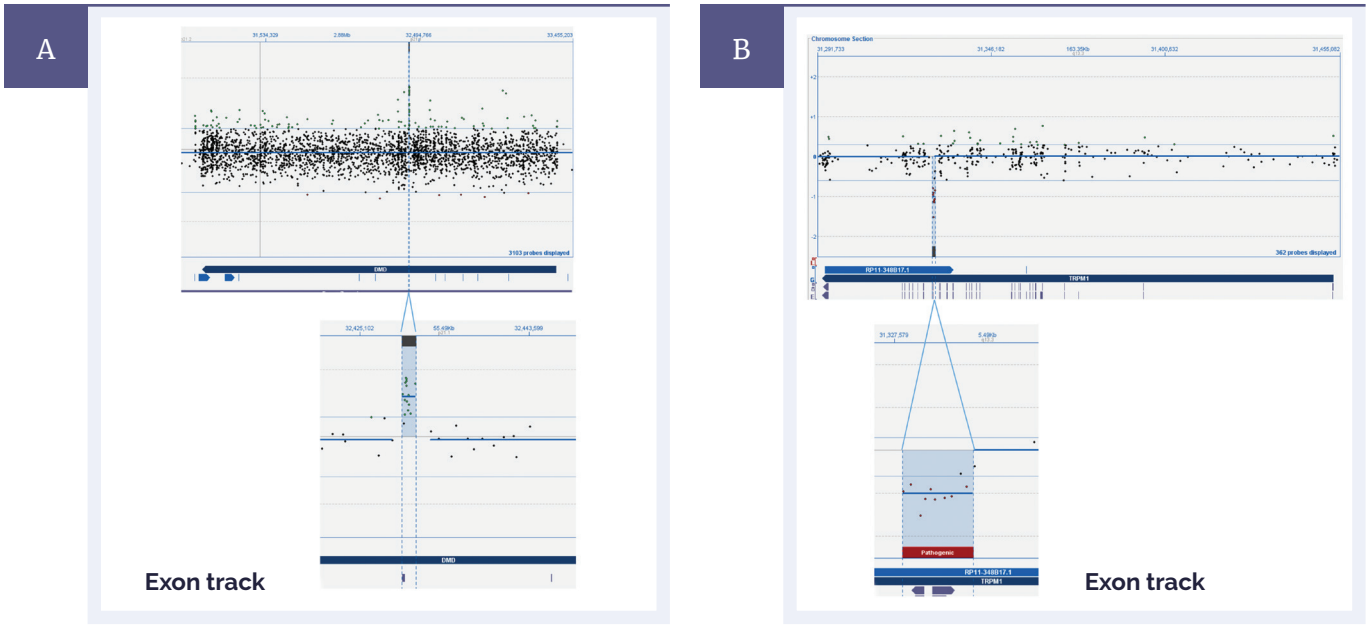

Figure 1: CytoSure Interpret Software clearly displays small aberrations and enables easy identification of genes and exons. A Duchenne muscular dystrophy (DMD) is caused by a mutation in the DMD gene which is one of the largest genes in the genome with over 79 exons. Shown here in the top panel is the complete view of the whole gene. The bottom panel is a close-up view showing a small duplication of 1.4kb in the DMD gene which spans a single exon. B Mutations in TRPM1 are often associated with congenital stationary night blindness. Shown here in the top panel is an overview of the whole gene. In the bottom panel, the close-up view shows a very small 684bp deletion which contains 10 probes, and also spans a single exon. Data kindly provided by Madhuri Hegde, Ph.D., FACMG, Emory University.

Bespoke custom array design service

Genetic research into molecular disorder samples is typically done in batches to ensure cost-effective processing. However, collecting sufficient samples, especially for rare disorders can significantly delay analysis. Combining content for rare disorders with more commonly occurring conditions on to a single array allows immediate processing of a diverse range of samples, thereby reducing both time to analysis and cost. Select from our proprietary database of over 26 million *in silico* validated probes and build a bespoke custom array based on your laboratory's requirements. Four array formats are available, 8x60k, 4x180k, 2x400k and 1x1M, allowing you to choose the one most suited to your content and throughput needs. Like all of our arrays, custom arrays are optimised to work with our range of high-quality labelling kits and are supplied with CytoSure Interpret Software, our class-leading data analysis package. It is easy to get started as full on-site wet lab and data analysis training is provided as standard. OGT validation data and customer feedback confirms that using CytoSure Genomic DNA Labelling Kits improves DLRS values and signal-to-noise ratios, ensuring accurate detection of even the smallest aberration. Poor quality metrics from using non-optimised kits can indicate an increased level of technical noise on the array which may have an impact on the ability to detect small aberrations.

Easy data interpretation

CytoSure Interpret Software is a powerful, easy-to-use package for the analysis of aCGH data. Innovative features such as the Accelerate Workflow enable standardised and automated data analysis, including automatic aberration detection and classification. It includes extensive annotation tracks covering syndromes, genes, exons, CNVs and recombination hotspots — each of which link to publicly available databases such as ISCA, Decipher and the Database of Genomic Variants and the Cancer Gene Consensus Genes (Figure 2) providing results in context. It is possible to select which tracks are displayed allowing only tracks of specific interest to be viewed (e.g. syndrome-specific tracks) ensuring easy data interpretation. Each track can reference hg18, hg19 or GRCh38 (hg20) information. Annotations within a track can be coloured allowing easy visualisation. It is also possible to customise what information from the tracks is saved in the report (e.g. how many common variants overlap with an aberration).

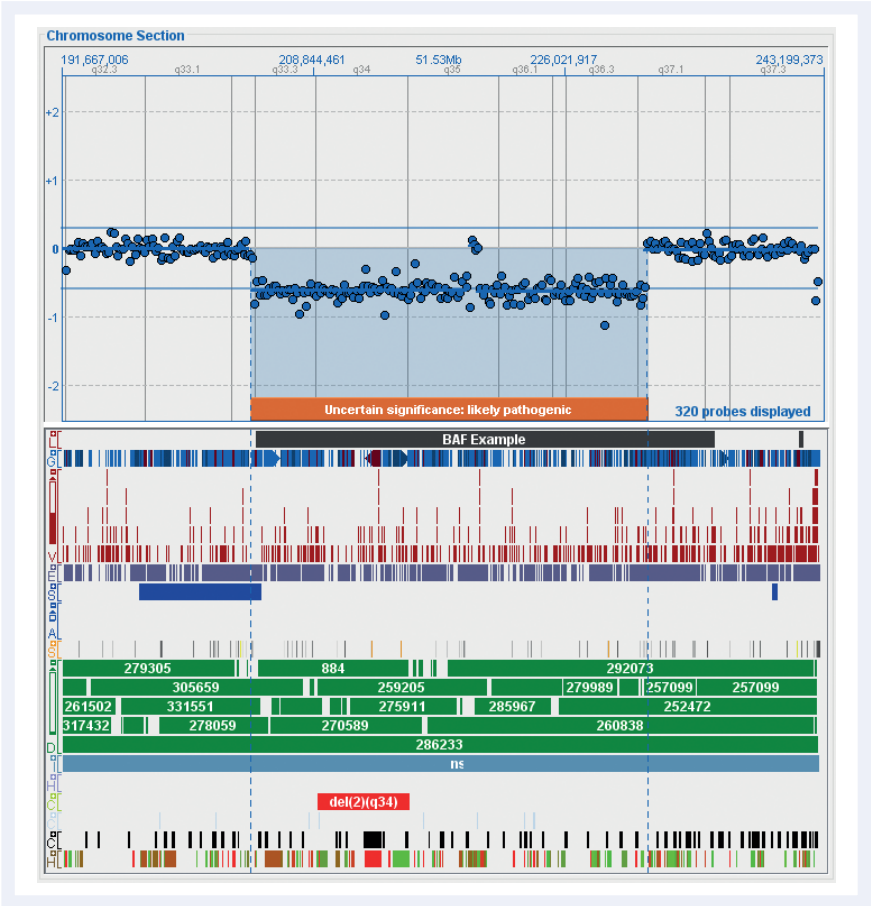

Figure 2: Chromosome overviews are clearly displayed in CytoSure Interpret Software. Shown here is an overview of the distribution of probes on chromosome 2 in the CytoSure Medical Research Exome array and the currently available standard tracks. These fully customisable tracks simplify the interpretation of aberrations.

Ordering information

Product	Contents	Cat. No.
CytoSure Medical Research Exome Array (1x1M)	Microarray with one array of 1,000,000 spots; CytoSure Interpret Software	020100
CytoSure Custom Molecular Array (1x1M, 2x400k, 4x180k or 8x60k)	Microarray with one array of 1,000,000 spots; two arrays of 400,000 spots; four arrays of 180,000 spots; or eight arrays of 60,000 spots; CytoSure Interpret Software	700001
CytoSure Genomic DNA Genomic Labelling Kit	24 reactions: clean-up columns, dyes, nucleotide mix, random primers, enzyme, collection tubes	020020
CytoSure HT Genomic DNA Labelling Kit	96 reactions: 2 purification plates, nucleotide mix, random primers, enzyme	500040
CytoSure Sample Tracking Spike-ins A – H	Sample Tracking Probes sufficient for 12 reactions supplied in three aliquots	various
CytoSure Interpret Software	Class-leading data analysis software provided with CytoSure arrays. Complimentary with all array purchases	020022

For more information on CytoSure products, visit ogt.com/medical.

Ordering information

UK +44 (0) 1865 856800

US +1 914 467 5285

contact@ogt.com

ogt.com

A Sysmex Group Company

What binds us, makes us.

Oxford Gene Technology Ltd., Begbroke Science Park, Woodstock Road, Begbroke, Oxfordshire, OX5 1PF, UK