AFFYMETRIX®

Data Sheet

GeneChip® Human Mitochondrial Resequencing Array 2.0

Analysis of mitochondrial mutations is informative for a variety of applications from disease genetics to forensic identification. The GeneChip® Human Mitochondrial Resequencing Array 2.0 provides the most efficient and cost-effective method for detecting germ line and heteroplasmic mutations by delivering the complete mitochondrial genome with minimal PCR in only 48 hours.

Features and Benefits

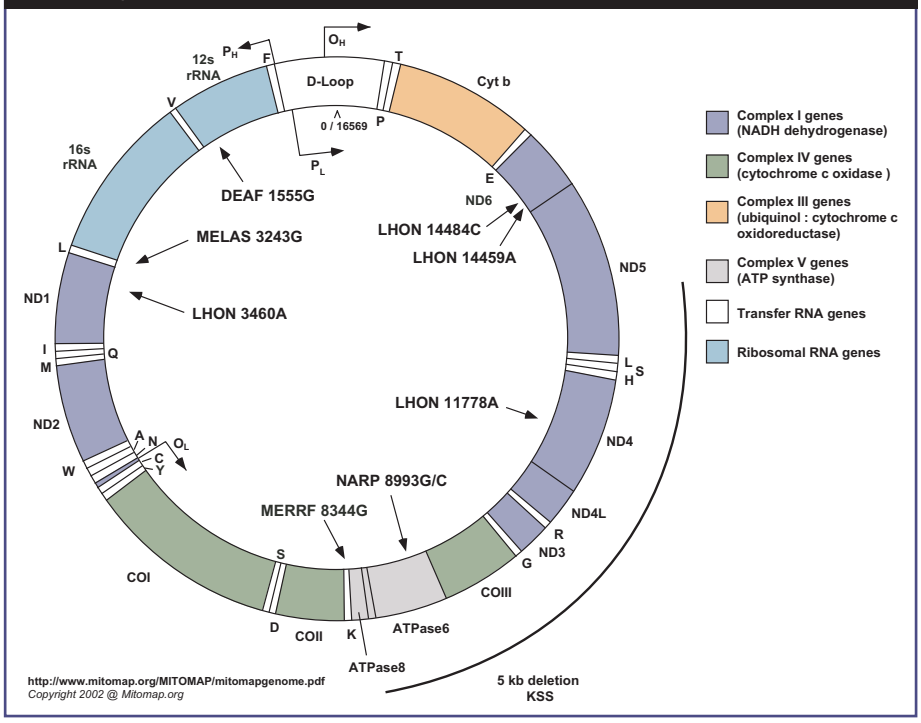
MORE INFORMATION PER EXPERIMENT

The GeneChip® Human Mitochondrial Resequencing Array 2.0 interrogates the entire 16 kb mitochondrial genome on one array. The reference sequence was selected from the public MITOMAP database (www.mitomap.org). Oligonucleotide probes are synthesized *in situ* using the standard resequencing array tiling strategy with eight unique 25-mer probes per base position (4 oligonucleotide probes per strand). Each 25-mer probe is varied at the central position to incorporate each possible nucleotide—A, G, C, or T — allowing for the detection of both known and novel SNPs. This array also contains additional tiling for many of the common variants in the HVI and HVII regions. These variants were selected from the FBI database (www.fbi.gov/hq/lab/fsc/backissu/april2002/miller1.htm).

LESS PCR

The robust Mitochondrial resequencing assay amplifies the entire genome in only three reactions, significantly cutting the time, labor, and cost involved in sample preparation. Traditional primer stategies may also be employed for highly degraded samples.

Figure 1: Mitochondrial Genome
Human Mitochondrial Resequencing Array 2.0 interrogates entire 16kb with additional redundant probes for known variation.



NO ASSEMBLY REQUIRED

The Mitochondrial Resequencing Array 2.0 delivers completed sequence in 48 hours with minimal alignment, curation, or hand editing, providing researchers with a faster, more efficient way to perform large-scale resequencing.

DETECTION OF HETEROPLASMY

The Mitochondrial Resequencing Array 2.0 enables the detection of heteroplasmic mutations.

Applications

DISEASE GENETICS

Mitochondrial instability has been reported in several diseases including degenerative and neurodegenerative disorders, sudden infant death syndrome, and multiple cancer types. The Human Mitochondrial Array provides complete sequence information and enables the detection of both known and novel mutations.

FORENSICS

Due to the availability and inherent stability of mitochondrial DNA, it is widely used in forensic applications when the genomic DNA is too scarce or degraded for standard STR analysis. Traditional methods of mitochondrial resequencing limit analysis to the D-loop regulatory region. By resequencing the entire genome, the Mitochondrial arrays can capture variation in any base of the entire genome and enable better resolution of similar samples.

POPULATION GENETICS

The mitochondrial genome is maternally transmittted as a haploid circular genome and, therefore, ideal for studying human evolution and tracking the migration of populations throughout history.

Critical Specifications

Format	169
Sequence Capacity	16 kb
Feature Size	8 µm
Instrumentation	GeneChip® Scanner 3000
Software	GeneChip® Operating Software 1.4, GeneChip® Sequence Analysis Software 4.0
Heteroplasmy Detection	Yes

Ordering Information

GeneChip® Human Mitochondrial Resequencing Array 2.0
900886 *Contains 5 Arrays*

GeneChip® Resequencing Reagent Kit
900447 *Contains 30 Reaction Kit*

GeneChip® Operating Software (GCOS 1.4)
690051 *1 seat license*

GeneChip® Sequence Analysis Software (GSEQ 4.0)
690052 *1 seat license*

REFERENCES

Cutler D.J., *et al.* Genome High-throughput variation detection and genotyping using microarrays. *Research* **11**(11):1913-25 (2001).

Maitra A., *et al.* Genomic Alterations in Cultured Human Embryonic Stem Cells. *Nature Genetics* EPub (2005 Sep 4).

Maitra A., *et al.* The Human MitoChip: A high-throughput sequencing microarray for mitochondrial mutation detection. *Genomic Research* **14**(5): 812-9 (2004).

Warrington, J.A., *et al.* New developments in high-throughput resequencing and variation detection using high-density microarrays. *Hum Mutat* **19**: 402-9 (2002).

AFFYMETRIX, INC.
3420 Central Expressway
Santa Clara, CA 95051 USA
Tel: 1-888-DNA-CHIP (1-888-362-2447)
Fax: 1-408-731-5441
sales@affymetrix.com
support@affymetrix.com

AFFYMETRIX UK Ltd
Voyager, Mercury Park
Wycombe Lane, Wooburn Green,
High Wycombe HP10 0HH
United Kingdom
UK and Others Tel: +44 (0) 1628 552550
France Tel: 0800919505
Germany Tel: 01803001334
Fax: +44 (0) 1628 552585
saleseurope@affymetrix.com
supporteurope@affymetrix.com

AFFYMETRIX JAPAN K.K.
Mita NN Bldg., 16 F
4-1-23 Shiba, Minato-ku,
Tokyo 108-0014 Japan
Tel: +81-(0)3-5730-8200
Fax: +81-(0)3-5730-8201
salesjapan@affymetrix.com
supportjapan@affymetrix.com

www.affymetrix.com Please visit our web site for international distributor contact information.

For research use only. Not for use in diagnostic procedures.

Part No. 702122 Rev. 2