



QIAGEN® Human ID and Forensics

# Massively Parallel Sequencing Solutions for Human Identity



## Sometimes an investigation goes cold

Human identity testing has been a force for good since its inception over three decades ago. HID techniques can be applied to identifying a criminal suspect, confirming someone's innocence, or repatriating a missing person's remains to their grieving family. However, although forensic science has advanced steadily, established methods have their limits, and sometimes an investigation goes cold.

DNA recovered from crime scenes, mass and natural disasters and other human ID scenarios are frequently degraded through exposure and over time. In addition, successful analysis with current methods is frequently hindered by the error-prone PCR assay, which introduces artifacts such as 'stutter' which complicate interpretation. As a result, confidence in results is sometimes low. Furthermore, even when the current methods work correctly, a criminal investigation may stall if there is no suspect to compare to a crime scene sample; there is no way to use the DNA result.

Current methods are often not sufficiently discriminating, particularly in cases involving familial testing, e.g., when addressing complicated paternity questions or identifying victims from mass graves.

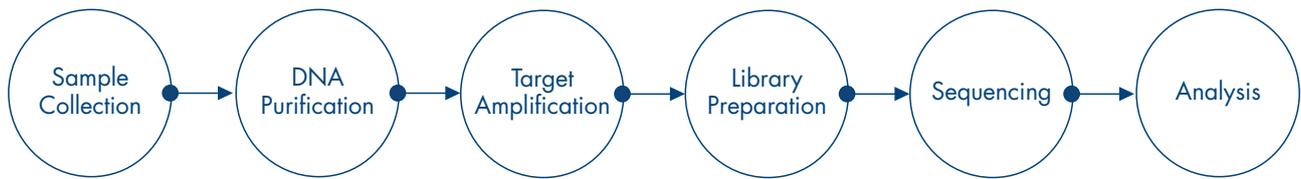
### **Single Nucleotide Polymorphisms: the next paradigm in human identity**

Single nucleotide polymorphisms (SNPs) have the potential to address these challenges in the current human identity workflow. Amplified fragments used to genotype SNPs can be very short, making SNPs potentially more successful than STRs when the DNA is extremely degraded. Such small amplicons are also much easier to multiplex in large numbers offering higher levels of discrimination. SNPs in closely linked sets can be jointly genotyped by MPS (Massively Parallel Sequencing) techniques. These phased microhaplotypes have multiple alleles, thereby increasing their discriminating power. When combined with large SNP multiplexes, these microhaplotypes enable discrimination power unrivaled by STRs. SNPs also offer less opportunity for enzyme slippage during PCR making interpretation easier.

Many forensic genetics experts foresee that an SNP-based approach may best capitalize on the strengths of NGS.

# Next-generation sequencing using QIAGEN's QIAseq<sup>®</sup> – a powerful tool for human identification

Human ID solutions for the MiSeq<sup>®</sup> and S5



## SNPs

|   |   |  |   |  |
|---|---|--|---|--|
| <p>Collectors and Collection Kits</p> <p>Indicating FTA Cards</p> | <p>QIAamp Investigator Kit</p> <p>EZ1 DNA Investigator Kit</p> <p>QIAasymphony Investigator Kit</p> | <p><b>QIAseq Targeted DNA panels:</b><br/>QIAseq targeted DNA custom panels</p> <p><b>Mitochondrial DNA</b></p> <p>QIAseq Human Mitochondria Panel</p> | <p>Compatible with the S5 and MiSeq</p> | <p>Genomics Workbench Client/Server Platform</p> <p>Custom Bioinformatics Services and Solutions</p> |
|---|---|--|---|--|



# QIAseq Targeted DNA Panels

Target the markers you need to inform your investigations

## Accurate

Avoid mistakes in interpretation due to stochastic PCR effects thanks to our Unique Molecular Indices (UMI) technology. UMIs are short sequences or molecular “tags” added to DNA fragments in the library preparation protocol to identify the input DNA molecule. UMIs are added before PCR amplification and every molecule undergoing the first round of PCR is tagged independently. All copies of the same target bear the same tag thereby assuring a significantly higher confidence in the final sequencing data.

## Discriminating

QIAseq panels confer unparalleled levels of discrimination with highly multiplexed SNP-based DNA markers that are more informative than previous methods (STR). Complex family relationships can be confirmed where previously such questions remained unanswered.

## Sensitive

QIAseq panels allow unrivaled confidence in your results, even with highly degraded, trace samples, meaning more casework samples are viable for testing than ever before.

## Intelligent

QIAseq panels also offer valuable information to further human ID forensic investigations when there are no suspects, such as trait prediction and ancestry information.

The power of massively parallel sequencing for human identity testing is finally realized with QIAGEN’s QIAseq.

## Robust

Optimized buffers, conditions and primer design deliver balanced results from your sequencing reactions, even for GC-rich regions, enabling easier downstream analysis and interpretation.

## Platform agnostic

Each panel is a one-box, NGS platform-agnostic solution that contains all the necessary components to amplify your required targets and construct libraries ready for sequencing.

## Flexible

In addition to offering separate QIAseq Investigator® panels for a wide range of HID applications, custom panels can be designed easily and quickly to target your regions of interest. This enables you to design your own panel to meet your own specific research or casework needs. Panels can be built and delivered within a few weeks. Using the robust QIAseq chemistry gives you high confidence that your panel will work straight out of the box without further optimization.



Visit GeneGlobe to discover the opportunities available to you to customize and optimize your workflow. <https://geneglobe.qiagen.com/product-groups/qiaseq-targeted-dna-panels>

## QIAseq Investigator Missing Persons SNP panel

Developed in partnership with the International Commission on Missing Persons (ICMP), the QIAseq Investigator Missing Persons SNP panel includes more than 1200 tri-allelic kinship SNPs in addition to 34 X-chromosome SNPs and 55 ancestry informative SNPs. Furthermore, the panel includes a set of 46 microhaplotypes, and these combine with the kinship SNPs to provide an unparalleled level of discrimination.



Catalogue number:  
CDHS-15861Z-2897, 333525

Number of primers:

**2897**

## QIAseq Investigator ID SNP panel

Developed with leading paternity scientists, the QIAseq Investigator ID SNP panel is designed to provide additional discrimination where STRs fall short, either because of DNA quality or the complexity of the analyzed (e.g., complex pedigree/paternity/immigration cases).



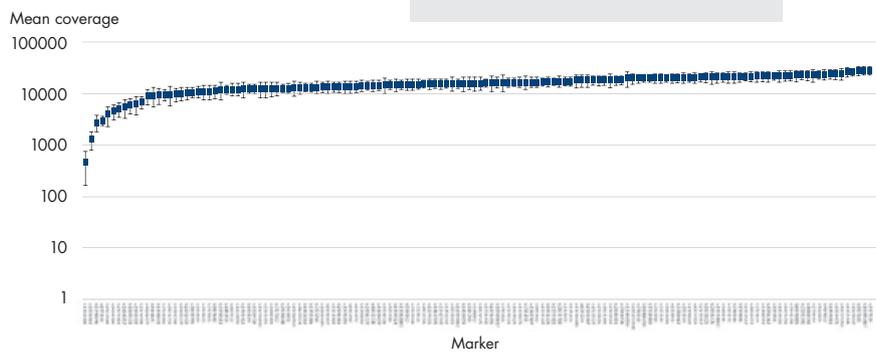
Catalogue number:  
CDHS-14055Z-277, 333525

Number of primers:

**277**

**Figure 1.**

Uniformity of coverage, for the 49 blood samples, for the 139 SNPs in the target enrichment panel sequenced on Illumina MiSeq. All 139 SNPs are shown along the x-axis and a logarithmic scale of coverage on the y-axis. Only 1 SNP was below 1000X coverage (the coverage of this SNP was enough to make accurate calls).





**Global ancestry informative marker set:**

Catalogue number:  
CDHS-12534Z-204, 333525

Number of primers:

**204**

**Middle East supplementary marker set:**

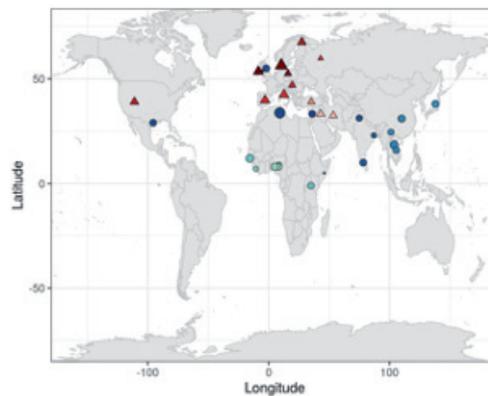
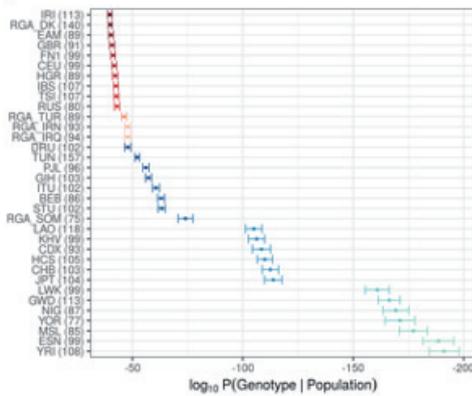
Catalogue number:  
CDHS-12533Z-169, 333525

Number of primers:

**169**

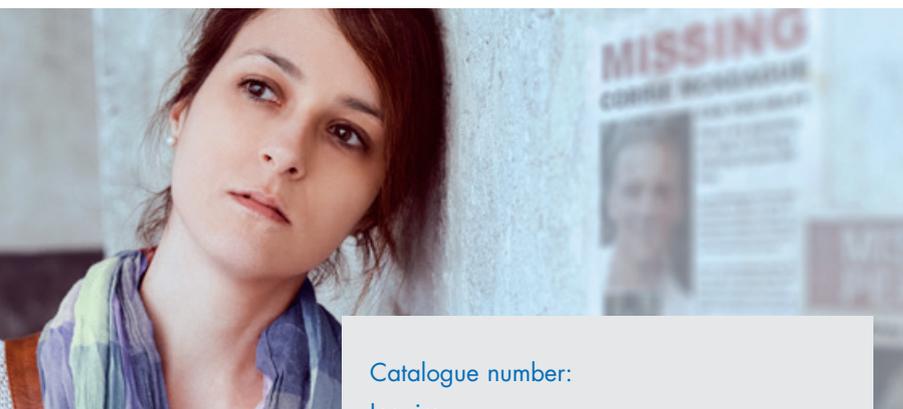
## QIAseq Investigator Ancestry SNP panel

Based on the SNPforID consortium’s exhaustive characterization of ancestry-informative markers, the QIAseq Investigator Ancestry SNP panel comprises subsets of markers aimed at accurately defining the key global population groups (Oceanian, American, African, European, East Asian).



**Figure 2.**

Sequence data from the Investigator QIAseq Ancestry SNP panel can be further analyzed using online tools such as GenoGeographer [1]. Image reproduced with kind permission of the authors.



Catalogue number:  
[Inquire](#)

## QIAseq Investigator Microhaplotype panel

Ideally suited for applications in casework and human identity such as mixture interpretation and kinship testing, the QIAseq Investigator Microhaplotype panel provides a comparable level of discrimination to typical STR kits. These markers can be further applied to missing person identification and paternity testing.

**Footnote**

1. Tvedebrink, T., Eriksen, P.S., Mogensen, H.S. and Morling, N. (2017) GenoGeographer – A tool for genogeographic inference. *Forensic Sci. Int.: Genet.* **6**, e463. <https://doi.org/10.1016/j.fsigs.2017.09.196>

# Mitochondrial markers

Mitochondrial DNA (mtDNA) presents several characteristics useful for forensic studies, especially related to the lack of recombination, a high copy number and matrilineal inheritance. Typing of mtDNA based on sequences of the control region or full genomic sequences is used to analyze a variety of forensic samples such as old bones, teeth and hair, as well as other biological samples where the DNA content is low. These panels give laboratories performing mtDNA casework the flexibility to detect variation within noncoding control region sequences or exploit the genetic diversity by analyzing full mtGenome sequence.

## QIAseq Human Mitochondria panel

Ideally suited for challenging or degraded remains, the QIAseq Human Mitochondria panel enables sequencing of the whole mitochondrial genome for casework, missing persons and all applications in human identification.

| Date: Thu, 18 May 2019 14:54:11 |  |                       |  |               |
|---------------------------------|--|-----------------------|--|---------------|
| 2 Haplogroups                   |  |                       |  |               |
| Name                            | Score  | # Found SNP           | # All used SNP                                       | # Private SNP |
| L2a1+143+16189+16192+16399      | 413  | 40                    | 3  | 2             |
| 3 Haplogroups (variants)        |  |                       |  |               |
| Name                            | Found SNP  | Missing SNP           | Private SNP  |               |
| L3a1+143+16189+16192+16399      | A393C, G148A, T146C, T152C, T193G, A203G, A295G, C209A, C1018A, A243G, A270G, C278G, C302T, A318G, A429G, C202T, T173C, C229G, C224G, G205A, A277C, G205A, A293C, A283G, A253C, T244C, T301C, A1029G, G1171A, C1194A, T1194G, A1309G, C1250T, C1168A, C1365T, A1383G, A1054G, C1456T, C1338A, A1338G, T1578A, C1427T, C1427T, C1679T, C1679T, C1638A, C1638T | T249G, T1687C, T1689C | C1337, T274C, C1058T, C1338T, A1339G, T6899C, C1629T |               |

Figure 3.

QIAGEN's QIAseq mitochondrial DNA panels can be analyzed with ease using the AQME mitochondrial DNA plug-in tool for the CLC Genomics Workbench. Analysis is straightforward and summary reports can be customized to meet your needs.

## QIAseq Investigator Human Mitochondria Control Region panel

In cases where only the control region sequence is required, the QIAseq Investigator Human Mitochondria Control Region panel enables a simpler and easier alternative to whole mitochondrial DNA sequencing.



**Whole mitochondrial genome marker set:**

Catalogue numbers:  
DHS-105Z, 333502 or 333505

Number of primers:

222

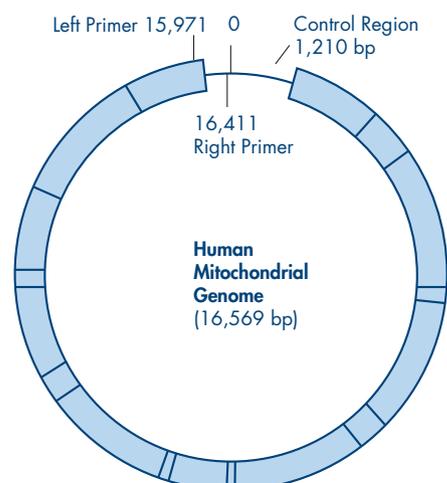
**Mitochondrial control region marker set:**

Catalogue number:  
CDHS-13743Z-27, 333525

Number of primers:

27

### Mitochondrial Control Region





## CLC Genomics Workbench

For analyzing, comparing and visualizing SNPs, mitochondrial DNA and other NGS data for human identity

CLC Genomics Workbench is a highly accurate and comprehensive desktop NGS data analysis platform. It provides user-friendly and customizable bioinformatics pipelines for SNPs, mitochondrial DNA analysis and other NGS applications in human identity. For those with higher throughput requirements, we can also provide local server and cloud solutions.

- Genotype SNP markers with high sensitivity and specificity through advanced visualization
- Analysis of Unique Molecular Indexed panel data avoids interpretation issues due to stochastic PCR effects
- Flexible Import and Export possibilities enables seamless integration of downstream analysis and statistics tools
- Dedicated HID plug-in tools for specialized analysis (e.g., AQME mitochondrial DNA plug-in)



Visit QIAGEN Digital Insights for information on the CLC Genomics Workbench.

<https://digitalinsights.qiagen.com/products-overview/discovery-insights-portfolio/analysis-and-visualization/qiagen-clc-genomics-workbench/>

# Custom Bioinformatics Services



The QIAGEN Digital Insights Bioinformatics Services team is dedicated to helping your organization achieve maximum value from its data. We provide a comprehensive suite of consulting, software development, training and other professional services that match bioinformatics and integration requirements regardless of complexity or scale. We have built this organization to help your organization achieve its scientific, IT and business goals because we realize that every organization has unique needs.



Visit QIAGEN Digital Insights for more information.  
<https://digitalinsights.qiagen.com/services-overview>

## Ordering Information

### QIAseq Investigator panels

| Catalog no.      | Panel name                                 | Total number of primers* | Product no. |
|------------------|--|--------------------------|-------------|
| CDHS-15861Z-2897 | ICMP Missing Persons SNP Panel             | 2897                     | 333525      |
| CDHS-11454Z-318  | ID SNP Panel                               | 318                      | 333525      |
| CDHS-12534Z-204  | Global Ancestry SNP Panel                  | 204                      | 333525      |
| CDHS-12533Z-169  | Middle East Ancestry Panel                 | 169                      | 333525      |
| DHS-105Z         | Human Mitochondria Whole Genome Panel (12) | 222                      | 333502      |
| DHS-105Z         | Human Mitochondria Whole Genome Panel (96) | 222                      | 333505      |
| CDHS-13743-27    | Human Mitochondria Control Region Panel    | 27                       | 333525      |
| Inquire          | Microhaplotype Panel                       |                          | Inquire     |

\*The number of primers in a panel is represented by the last digits of the catalog number. For example, a custom panel with catalog number CDHS-00100Z-1256 has 1256 primers.

## Ordering Information

### Products

| Product                                       | Contents  | Cat. no. |
|---|---|----------|
| <b>QIAseq panels</b>                          |   |          |
| QIAseq Targeted DNA Custom Panel (96)         | ALL reagents (except indexes) for targeted DNA sequencing; Custom panel for 96 samples  | 333525   |
| QIAseq Targeted DNA Panel (12)                | ALL reagents (except indexes) for targeted DNA sequencing; fixed panel for 12 samples; less than 200 genes  | 333502   |
| QIAseq Targeted DNA Panel (96)                | ALL reagents (except indexes) for targeted DNA sequencing; fixed panel for 96 samples; less than 200 genes  | 333505   |
| <b>Indexes/adaptors: Illumina platform</b>    |   |          |
| QIAseq 12-Index I (48)                        | Box containing molecularly-indexed adapters and primers, enough for a total of 48 samples – for indexing up to 12 samples for targeted panel sequencing on Illumina platforms   | 333714   |
| QIAseq 96-Index I Set A (384)                 | Box containing molecularly-indexed adapters and primers, enough for a total of 384 samples – for indexing up to 96 samples for targeted panel sequencing on Illumina platforms; one of 4 sets required for multiplexing 384 samples | 333727   |
| QIAseq 96-Index I Set B (384)                 | Box containing molecularly-indexed adapters and primers, enough for a total of 384 samples – for indexing up to 96 samples for targeted panel sequencing on Illumina platforms; 2 of 4 sets required for multiplexing 384 samples   | 333737   |
| QIAseq 96-Index I Set C (384)                 | Box containing molecularly-indexed adapters and primers, enough for a total of 384 samples – for indexing up to 96 samples for targeted panel sequencing on Illumina platforms; 3 of 4 sets required for multiplexing 384 samples   | 333747   |
| QIAseq 96-Index I Set D (384)                 | Box containing molecularly-indexed adapters and primers, enough for a total of 384 samples – for indexing up to 96 samples for targeted panel sequencing on Illumina platforms; 4 of 4 sets required for multiplexing 384 samples   | 333757   |
| <b>Indexes/adaptors: Ion Torrent platform</b> |   |          |
| QIAseq 12-Index L (48)                        | Box containing molecularly-indexed adapters and primers, enough for a total of 48 samples – for indexing up to 12 samples for targeted panel sequencing on Ion Torrent platforms  | 333764   |
| QIAseq 96-Index L (384)                       | Box containing molecularly-indexed adapters and primers in arrays, enough for a total of 384 samples – for indexing up to 96 samples for targeted panel sequencing on Ion Torrent platforms   | 333777   |

see next page for a continued list



## Ordering Information – continued

| Product                              | Contents   | Cat. no.                 |
|--------------------------------------|--|--------------------------|
| <b>Quantification reagents</b>       |  |                          |
| QIAseq DNA QuantiMIZE Array Kit      | qPCR arrays for optimizing amount of input DNA and PCR cycling conditions for low-quantity DNA samples                       | 333404<br>DNAQA-002-xxx* |
| QIAseq Library Quant Array Kit       | Reagents for quantification of libraries prepared for Illumina platform; array format  | 333304<br>QSIL-003-xxx*  |
| QIAseq Library Quant Array Kit       | Reagents for quantification of libraries prepared for Ion Torrent platform; array format                                     | 333304<br>QSIT-003-xxx*  |
| <b>Analysis</b>                      |  |                          |
| CLC Genomics Workbench, Desktop Plus | 1-year subscription for a static license to use the software on a single computer; includes maintenance, upgrade and service | 832021                   |
| CLC Custom Solutions, days           | Professional services  | 832580                   |

\* Value of "xxx" depends on cycler format.

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For up-to-date licensing information and product-specific disclaimers, see the respective QIAGEN kit handbook or user manual. QIAGEN kit handbooks and user manuals are available at [www.qiagen.com](http://www.qiagen.com) or can be requested from QIAGEN Technical Services or your local distributor.

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