TaqMan SNP Genotyping Assays

Applied Biosystems™ TaqMan™ SNP Genotyping Assays from Thermo Fisher Scientific provide a highly flexible technology for detection of polymorphisms within any genome. Applied Biosystems™ TaqMan™ Assays have the simplest workflow available and are the quickest way to generate genotyping data. Based on powerful Applied Biosystems™ TaqMan™ probe and primer chemistry and designs, and coupled to dependable Applied Biosystems™ instruments and software, these made-to-order assays produce high-confidence results. TaqMan Assays are ideal for genotyping applications, including screening, association, candidate region, candidate gene, and fine-mapping studies.

Content-rich marker selection tools simplify study design and help you select from a library of human and mouse assays. This library includes over 7 million genome-wide human assays (of which 3.5 million are HapMap SNP-based assays, 160,000 are validated assays, and over 950,000 are coding region assays) and 10,000 mouse assays. We also offer 2,700 inventoried drug metabolism genotyping assays. Additionally, with Applied Biosystems™ Custom TaqMan™ SNP Genotyping Assays you can confidentially submit target SNP sequences for any genome to create your own assays. Let TaqMan SNP Genotyping Assays accelerate the pace of your discovery by eliminating time-consuming experimental design and optimization.

Powerful, proven chemistry

Whether your genotyping studies require targeted detection of essential SNPs, or the flexibility to choose SNPs for mapping, TaqMan SNP Genotyping Assays are the technology of choice. Proven TaqMan

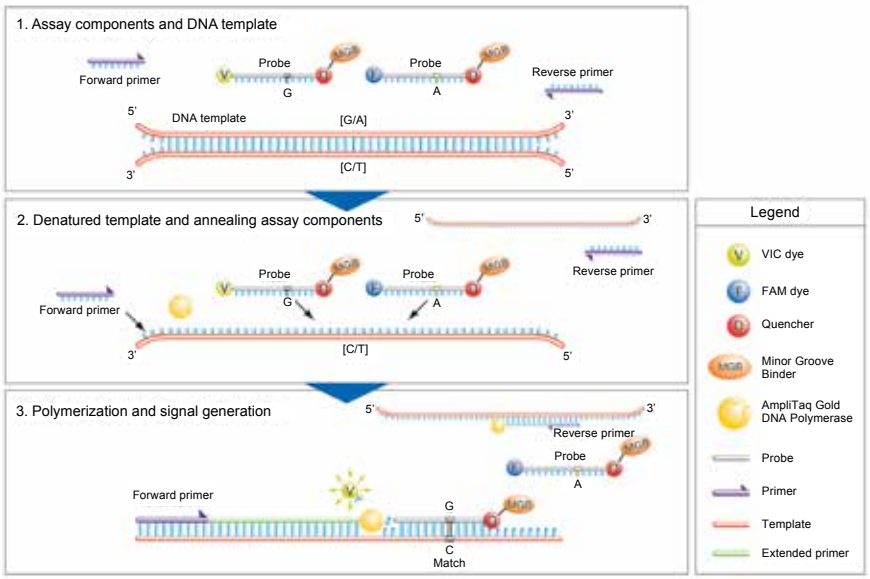

Figure 1. Allelic discrimination is achieved by the selective annealing of TaqMan MGB probes.

probes, which incorporate minor groove binder (MGB) technology at the 3′ end, deliver superior allelic discrimination. The MGB molecule binds to the minor groove of the DNA helix, improving hybridization-based assays by stabilizing the MGB probe–template complex. This increased binding stability permits the use of probes as short as 13 bases for improved mismatch discrimination and greater flexibility when designing assays for difficult or variable sequences. In addition to SNP detection, TaqMan probes can be designed to detect multiple nucleotide polymorphisms (MNPs) and insertion/deletions (indels).

Detection is achieved with proven 5′ nuclease chemistry by means of exonuclease cleavage of an allele-specific 5′ dye label, which generates the permanent assay signal (Figure 1). All MGB probes include a nonfluorescent quencher (NFQ) that virtually eliminates the background fluorescence associated with traditional quenchers, and provides a greater signal-to-noise ratio for superior assay sensitivity.

TaqMan SNP Genotyping Assays collection

TaqMan SNP Genotyping Assays are the world's largest collection of single-tube, ready-to-use SNP assays available. The TaqMan SNP Genotyping Assays library consists of two collections of human assays and one of mouse assays, and can be supplemented with assays designed using our Custom TaqMan SNP Genotyping Assays Service.

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Over 7 million human SNP genotyping assays

This assay group contains over 7 million genome-wide SNPs, providing unprecedented marker coverage. Included in this collection are 160,000 validated assays that have approximately 10 kb spacing across gene regions. These assays were subjected to an extensive minor allele frequency test in 2–4 ethnic populations (45 individual samples per ethnic group) and as a result, offer the highest success rate. Also included are over 600,000 assays for the detection of nonsynonymous SNPs in coding regions, including many putative functional SNPs. Visit **thermofisher.com/taqmansnp** for more information.

Over 10,000 mouse SNP genotyping assays

The Applied Biosystems™ Mouse TaqMan™ Predesigned SNP Genotyping Assays collection consists of over 10,000 assays, and can be supplemented with assays designed using our Custom TaqMan SNP Genotyping Assays Service.

TaqMan Drug Metabolism Genotyping Assays

The collection of Applied Biosystems™ TaqMan™ Drug Metabolism Genotyping Assays includes 2,700 assays that target high-value polymorphisms in 221 drug metabolism genes. These assays have proven performance in four different ethnic population samples, consisting of 45 individuals each. To enable easy identification, these assays have been mapped to the common public allele nomenclature websites where possible. Visit **thermofisher.com/taqmandme** for more information.

All TaqMan SNP Genotyping Assays are generated using next-generation algorithms from the Thermo Fisher Scientific bioinformatics pipeline. For all predesigned assays, bioinformatics evaluation of target SNP sequences includes the masking of adjacent SNPs and ambiguous bases so that assay design and subsequent performance is not affected by the poor quality of the underlying sequence. Lastly, the assay designs are aligned to the human genome using BLAST to ensure that each assay binds uniquely to the intended polymorphism. As the Custom TaqMan SNP Genotyping Assay Service is confidential and secure, you simply perform your own bioinformatics analysis prior to submitting your sequence for assay design.

Custom assay service for any possible SNP

Custom TaqMan SNP Genotyping Assays can be developed for any SNP in any organism. This service can generate assays for the detection of SNPs, MNPs, indels of up to 6 bases, or QSY™-labeled probes for multiplexing SNP assays.

Custom TaqMan SNP Genotyping Assays provide you with a complete service that includes secure and confidential ordering, assay design and manufacturing, and quality-control testing for synthesis accuracy and formulation completeness. Additionally, custom human assays are subjected to a functional test on 20 unique DNA samples.

Use the free Applied Biosystems™ Custom TaqMan™ Assay Design Tool to input and submit your sequence for assay design. This easy-to-use online resource lets you quickly submit your sequence information and start the ordering process securely and confidentially. Access the Custom TaqMan Assay Design Tool at **thermofisher.com/snpcadt**

Quality design and manufacturing

Probes and primers used in TaqMan SNP Genotyping Assays are designed using our rigorous bioinformatics pipeline. This proprietary group of algorithms has generated millions of TaqMan Assay designs by utilizing heuristic design rules deduced from both manufacturing and assay performance data. All assays are designed to perform under universal reaction conditions, as calculated probe and primer melting temperatures are consistent and include contributions from associated probe conjugates (i.e., dyes and MGB).

After manufacturing, assay components undergo extensive laboratory testing at our state-of-the-art manufacturing facility. Quality-control testing includes mass spectrometry for sequence verification and formulation assessments of probe and primer concentrations. Additionally, all human SNP genotyping assays are functionally tested to ensure allelic discrimination.

Simple workflow for quick results

TaqMan SNP Genotyping Assays constitute the simplest SNP genotyping technology available. We deliver your ready-to-use SNP genotyping assay at ambient temperature in a convenient, single-tube format. The rest is easy. Just combine the assay with Applied Biosystems™ TaqMan™ Genotyping Master Mix or TaqMan™ Universal PCR Master Mix and your purified DNA sample (Figure 2). There is no need to optimize probe, primer, salt concentrations, or temperature because all assays use universal reagent concentrations and thermal cycling conditions.

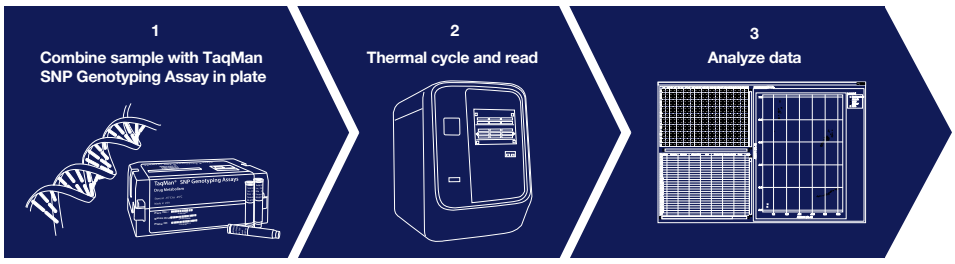

Figure 2. A simple workflow and reliable instruments combine to generate fast, high-confidence results.

After generating an endpoint read using a thermal cycler or real-time PCR instrument, no transfers, washes, or additional reagents are required, and the plate remains sealed; just read the plate and analyze the genotypes. This reduces the chance of contamination, sample mix-up, and sample loss. The simplicity of the chemistry allows you to easily automate the reaction for massively parallel genotyping studies, readily increasing the number of assays, number of samples, or both. Additionally, the analysis software allows you to auto-call genotypes, minimizing manual intervention.

Reliable real-time PCR platforms

A suite of superior Applied Biosystems instrument platforms is available for processing and analyzing TaqMan SNP Genotyping Assays (Table 1). These instruments, which meet all throughput needs and budgets, include the 7500, 7500 Fast, 7900HT Fast, ViiA™ 7, StepOne™, and StepOnePlus™ Real-Time PCR Systems, and the QuantStudio™ 3, 5, 6, 7, and 12K Flex Real-Time PCR Systems (Figure 3). Following PCR amplification, an endpoint read can be performed on any Applied Biosystems real-time PCR system. All of these dependable instruments offer the advanced multicolor detection capabilities required for highly accurate and reproducible allelic discrimination assays.

Data analysis software

The sophisticated SDS software package provided with all Applied Biosystems real-time PCR systems facilitates experimental setup, data collection, and assay performance analysis. The SDS software uses an advanced multicomponent algorithm to calculate the distinct signal contribution of each allele of a marker from the fluorescence measurements of each sample well during the assay plate read. The multicomponent data collected from the plate read are stored as SDS files, ready for genotype determination by the SDS software or optional Applied Biosystems™ TaqMan™ Genotyper Software (Figure 4).

Table 1. Applied Biosystems instrument capacities.

Instrument	Capacity
7500/7500 Fast Real-Time PCR System	96-well block (standard or Fast)
7900HT Fast Real-Time PCR System	96- and 384-well blocks (standard or Fast)
ViiA 7 Real-Time PCR System	96-well (standard or Fast), 384-well, and TaqMan Array Card blocks
StepOne Real-Time PCR System	48-well block (standard or Fast)
StepOnePlus Real-Time PCR System	96-well block (standard or Fast)
QuantStudio 3 Real-Time PCR System	96-well block (standard or Fast)
QuantStudio 5 Real-Time PCR System	96-well (standard or Fast) and 384-well blocks
QuantStudio 6 Real-Time PCR System	96-well (standard or Fast) and 384-well blocks
QuantStudio 7 Real-Time PCR System	96-well (standard or Fast), 384-well, and TaqMan Array Card blocks
QuantStudio 12K Flex Real-Time PCR System	96-well (standard or Fast), 384-well, TaqMan Array Card, and OpenArray plate blocks

Figure 3. The QuantStudio 5 Real-Time PCR System (left) and the QuantStudio 12K Flex Real-Time PCR System (right), which offers the highest throughput of all Applied Biosystems real-time PCR instruments.

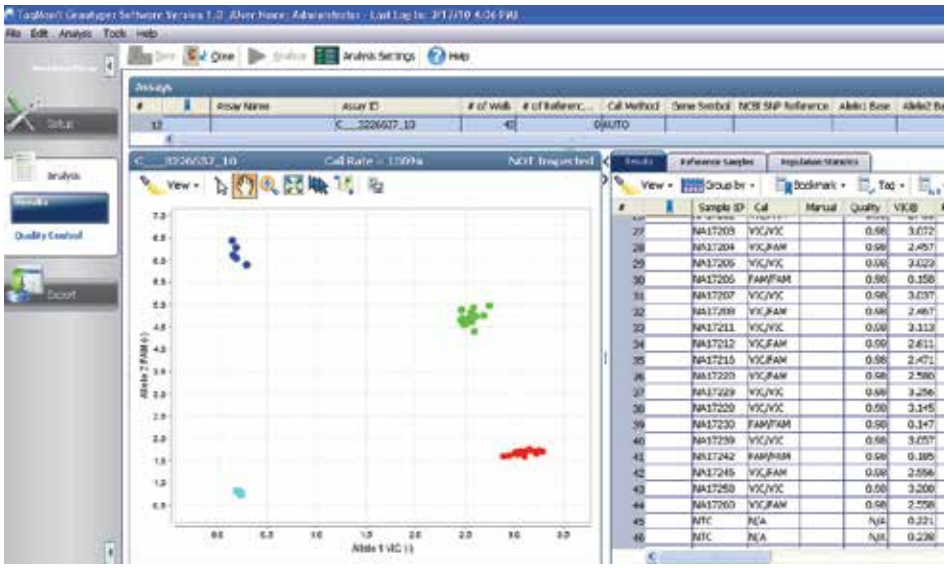

Figure 4. TaqMan Genotyper Software automatically determines sample genotypes and displays data.

TaqMan Genotyper Software is a great resource for fast and accurate genotype calling. It is a free SNP genotyping data analysis tool for use with TaqMan SNP Genotyping Assays performed in 48-, 96-, or 384-well microtiter plates or on Applied Biosystems™ TaqMan™ OpenArray™ Plates. It has a state-of-the-art genotype-calling algorithm, an intuitive user interface, and enhanced study-based analysis features. The software enables multi-plate data analysis for high-throughput workflows and improved accuracy in genotype calling; versatile export features and comprehensive quality-control features facilitate streamlining of the entire workflow. TaqMan Genotyper Software can be downloaded at **thermofisher.com/taqmangenotyper**

Simple ordering

Selecting and ordering TaqMan SNP Genotyping Assays is as simple as "point and click." Use SNPbrowser Software to select the most informative SNPs for your genotyping studies. As you identify SNPs of interest, simply upload your selected TaqMan SNP Genotyping Assays to our online ordering tool.

The TaqMan Assay online ordering tool (Figure 5) enables you to search, select, and order from our catalog of over 7 million made-to-order predesigned TaqMan SNP Genotyping Assays. You can search for SNPs using any of several criteria: National Center for Biotechnology Information (NCBI) gene ID, NCBI SNP reference ID (rs#), or gene symbol. You can further refine your search by using SNP type (i.e., intragenic, 5′ or 3′ UTR, chromosome, etc.).

Our Custom TaqMan SNP Genotyping Assays supply you with SNPs that are not available from our predesigned assay collection, including those from any nonhuman organism. This service designs assays for all possible SNP, MNP, and indel targets but without the up-front bioinformatic preparation used for the predesigned made-to-order assays. Our complementary Custom TaqMan Assay Design Tool conveniently formats your target sequence for submission to our manufacturing facilities. To order custom assays, simply prepare your target sequence according to the Design and Ordering Guide, and upload your submission file at **thermofisher.com/snpcadt**

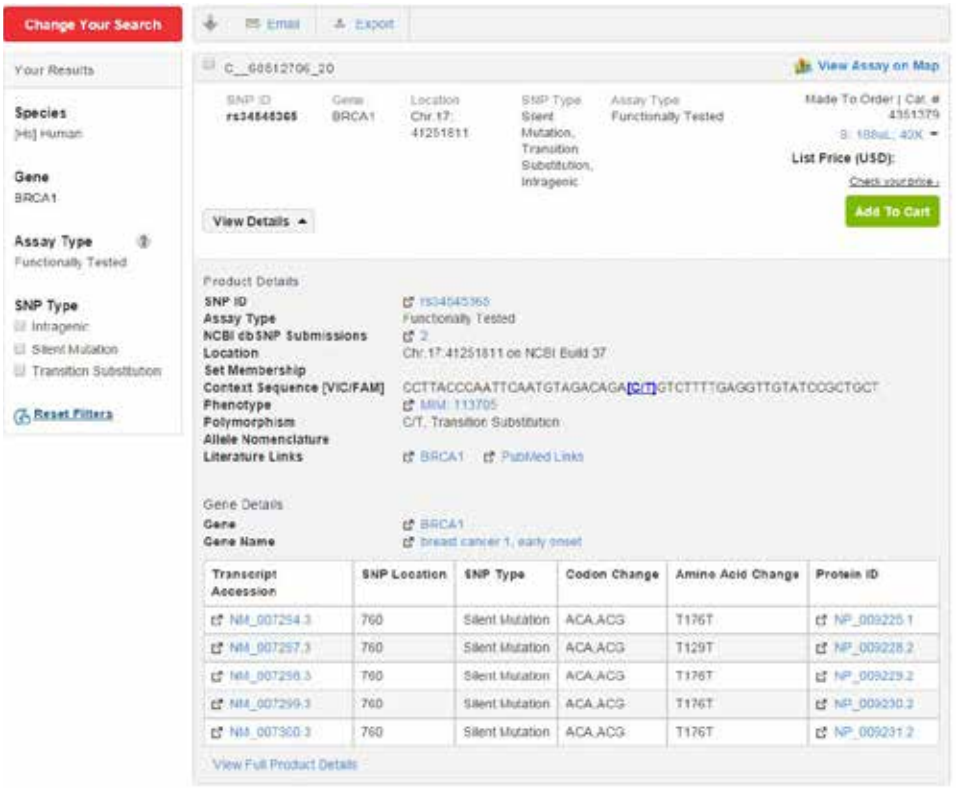

Figure 5. Our TaqMan Assay search and order tool makes online ordering easy. For convenient online ordering and multiple search options for all our genotyping assays, including keyword, batch, and location searches, visit **thermofisher.com/taqmansnp**

Ordering information

Size	Human assays (Cat. No.)	Nonhuman assays (Cat. No.)	Number of SNPs	No. of 5 µL reactions (384-well plate)	No. of 25 µL reactions (96-well plate)	Assay mix formulation	Assay type
Predesigned TaqMan SNP Genotyping Assays for Human and Mouse							
Small	4351379	4351384*	>4.5 million	1,500	300	40X	made-to-order
Medium	4351376	4351382*	>4.5 million	5,000	1,000	40X	made-to-order
Large	4351374	4351380*	>4.5 million	12,000	2,400	80X	made-to-order
Custom TaqMan SNP Genotyping Assays							
Small	4331349	4332077	∞	1,500	300	40X	made-to-order
Medium	4332072	4332075	∞	5,000	1,000	40X	made-to-order
Large	4332073	4332076	∞	12,000	2,400	80X	made-to-order
TaqMan Drug Metabolism Genotyping Assays							
Small	4362691	NA	2,700	750	150	20X	inventoried

All assays are quality-control tested using a mass spectrometer to verify sequence and yield. All assays have a VIC™ dye–labeled probe, a FAM™ dye–labeled probe, and two target-specific primers. All assays, excluding Custom TaqMan SNP Genotyping Assays, undergo bioinformatics evaluation of target SNP sequences.

Functional testing against 20 unique genomic DNA samples is performed on all custom and predesigned made-to-order human TaqMan SNP Genotyping Assays. Validation testing against four populations (45 samples/population) was performed on all 160,000 validated TaqMan SNP Genotyping Assays, and all TaqMan Drug Metabolism Genotyping Assays.

* Over 10,000 mouse assays available.

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Find out more at **thermofisher.com/taqmansnp**

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