



Product Specification

SRY, X & Y Genemer™ Fluorescent Molecular Probes

Genemer Specific TaqMan and Molecular Beacons for Quantitative PCR (QPCR)
Store at -20°C. For research use only. Not for use in diagnostic procedures for clinical purposes.

□	44-6430-01	SRY Genemer™ specific TaqMan probe 5' Fam/3' Tamra	2 nmols
□	44-6130-01	X Genemer™ specific TaqMan probe 5' Fam/3' Tamra	2 nmols
□	45-6430-01	SRY Genemer™ specific Molecular Beacon probe 5' Fam/3' Dabcyl	2 nmols
□	45-6130-01	X Genemer™ specific Molecular Beacon probe 5' Fam/3' Dabcyl	2 nmols

Background

The human sex determining region on the Y chromosome has been identified and the gene has been termed as *SRY*. Mutations in the *SRY* gene have been found in XY females. Sex reversal in XY females results from the failure of the testis determination or differentiation pathways. Some XY females with gonadal dysgenesis have lost the *SRY* gene from the Y chromosome by terminal exchange between the sex chromosome or by other deletions or mutations affecting activity (1,2).

DNA analysis for a specific region of *SRY* together with aliphoid repeat regions of the X and Y chromosome is used for accurate sex determination (in the absence of mutations involving *SRY*), and in the characterization of X-linked genetic diseases, Y chromosome anomalies such as XY females with gonadal dysgenesis, and for XO/XY mosaicism in patients with Turner syndrome. The DNA test involves the amplification of specific regions of X, Y and *SRY*. The presence of amplified product directly indicates the presence of the cognate DNA fragments on the chromosome. Normal XX females will amplify only X chromosome specific fragment showing double intensity as compared with amplification from normal XY male. *SRY* and Y fragments will only be amplified from individuals with a Y chromosome.

References

- Berta et al. (1990) Genetic evidence equating *SRY* and the testis-determining factor. *Nature* 348:448-451.
- Jager et al. (1990) A human XY female with frame shift mutation in the candidate testis-determining gene *SRY*. *Nature* 348:452-453.

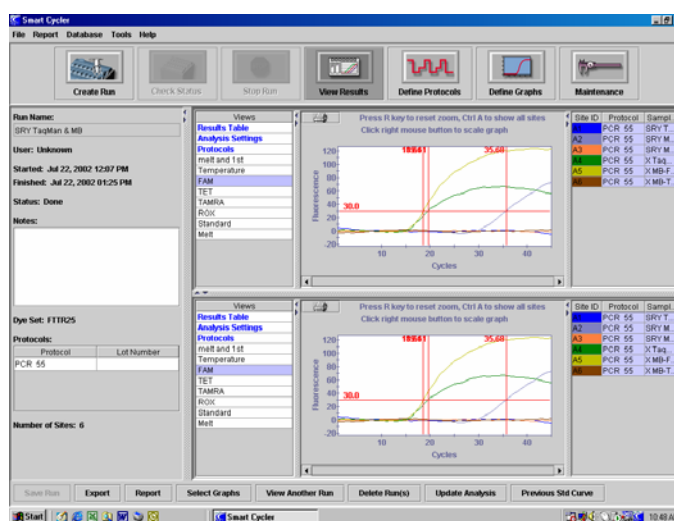
Material Supplied

One tube containing lyophilized fluorescent molecular probe. These probes are specific for Gene Link Genemer™. Please refer to item number on the top of this sheet. Each tube contains 2 nmols of the fluorescent molecular probe. The quantity supplied is sufficient for 400 QPCR of 25 µl volume.

A. Reconstitution

Stock Probe Solution: Dissolve the supplied lyophilized fluorescent molecular probe in 20 µl 1X PCR buffer. The 2 nmols of probe when dissolved in 20 µl will give a solution of 100 µM i.e. 100 pmols/µl. Store at -20°C.

Working Probe Solution: Prepare a 5 pmols/µl solution in 1X PCR buffer in **amber/dark** tube. Example: Add 38 µl 1X PCR buffer to a new tube, to this tube add 2 µl of probe stock solution. Label this tube as Probe Solution 5 pmols/µl.



Above: A screen shot from a Cepheid Real Time PCR using SRY and X Genemer™ specific TaqMan and Molecular Beacons.

Below: Gel electrophoresis pattern of loading samples to 1.5% agarose gel. Run at 90 mAmps for 2.5 hrs.

- Normal female DNA should only amplify X specific fragment.
- Normal male DNA should amplify all fragments (SRY, X & Y)

Normal PCR amplified fragment size		
SRY	X chromosome	Y chromosome
422 bp	130 bp	170 bp

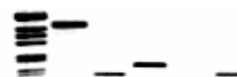


Figure 1. SRY, X and Y PCR amplification gel profile. Lane 1, molecular weight marker. Lanes 2-4 male DNA, lanes 5-7 female DNA. Lanes 2 and 5 SRY amplification, lanes 3 and 6 X amplification, lanes 4 and 7 Y amplification. Note the absence of amplification of SRY and Y from female DNA (lanes 5 & 7).



B. QPCR* File**Protocol for SRY, X and Y DNA Genotyping**

The following PCR* profile has been optimized for SRY, X and Y specific product amplification using the specific Genemer™.

QPCR Profile		
Denaturation	94 °C	30 sec.
Annealing	55 °C	30 sec.
Elongation	72 °C	1 min.

C. QPCR* Typical Setup

QPCR Setup for 25 µl Reaction Volume		
Component	Volume	Final Concentration
10 x PCR Buffer	2.5 µl	
2.0 mM dNTP mix (each)	2.5 µl	
25 mM MgCl ₂	3 µl	3mM of total Mg ⁺⁺
Primer Mix (10 pmol/µl each)	1.25 µl	[0.5 pmol/µl final or 500 nM]
Probe (MB or TaqMan) 5 pmol/µl	1 µl	[0.2 pmol/µl final or 200 nM]
Template	1 µl	
Taq Polymerase	0.5 µl	
H ₂ O	13.25 µl	

Ordering Information

Genemer™ (Selected List) Primer pair for gene or mutation specific amplification. Special optimized conditions may be required for certain amplifications.

Product	Size	Catalog No.	Price, \$
Fragile X (spanning triple repeat region)	10nmols	40-2004-10	100.00
Huntington Disease (spanning triple repeat region)	10nmols	40-2025-10	100.00
Myotonic Dystrophy (spanning triple repeat region)	10nmols	40-2026-10	100.00
Friedreich Ataxia	10nmols	40-2027-10	100.00
Factor V	10nmols	40-2035-10	100.00
Factor VIII (Hemophilia)	10nmols	40-2036-10	100.00
STS (Steroid Sulfatase)	10nmols	40-2023-10	100.00
HGH (Human Growth Hormone)	10nmols	40-2024-10	100.00
Sickle Cell	10nmols	40-2001-10	100.00
RhD (Rh D gene exon 10 specific)	10nmols	40-2002-10	100.00
Rh EeCc (Rh Ee and Cc exon 7 specific)	10nmols	40-2003-10	100.00
Gaucher* (various mutations)	10nmols	40-2047-10	100.00
Cystic Fibrosis* (various mutations)	10nmols	40-2029-10	100.00
SRY (sex determining region on Y)	10nmols	40-2020-10	100.00
X alphoid repeat	10nmols	40-2021-10	100.00
Y alphoid repeat	10nmols	40-2022-10	100.00

***Please visit www.genelink.com for other GENEMER™ not listed here**

Genemer™ Control DNA (Selected List)

Product	Size	Catalog No.	Price, \$
Fragile X, various CGG triple repeat region control DNA	10nmols	40-2004-XX	175.00
Huntington Disease various CAG triple repeat region control DNA	10nmols	40-2025-XX	175.00
Myotonic Dystrophy various CTG triple repeat region control DNA	10nmols	40-2026-XX	175.00
Friedreich Ataxia, various GAA triple repeat region control DNA	10nmols	40-2027-XX	175.00

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GeneProber™ (Selected List)

Product	Size	Catalog No.	Price, \$
Fragile X GeneProber™ Products Products and kits for non-radioactive genotyping CGG triple repeat spanning region	Various	40-2004-XX	Check website
Huntington Disease Gene Prober™ Products Products and kits for non-radioactive genotyping CAG triple repeat spanning region	Various	40-2025-XX	Check website
Myotonic Dystrophy Gene Prober™ Products Products and kits for non-radioactive genotyping CTG triple repeat spanning region	Various	40-2026-XX	Check website
Friedreich's Ataxia Gene Prober™ Products Products and kits for non-radioactive genotyping GAA triple repeat spanning region	Various	40-2026-XX	Check website

*The polymerase chain reaction (PCR) process is covered by patents owned by Hoffmann-La Roche. A license to perform is automatically granted by the use of authorized reagents.
Prices subject to change without notice

All Gene Link products are for research use only.

