




**Affymetrix® GeneChip® Scanner 3000 Targeted  
Genotyping System Site Preparation Guide**

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Chapter 1

**Before You Start**

Chapter 1





## About This Guide

### Contents

This site preparation guide provides you with the information you need to prepare your facility and personnel for equipment installation, laboratory set up, training on and use of the Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System (GCS 3000 TG System). It includes:

- A list of the equipment, supplies and consumables required to perform the MIP Assay Protocol.
- Recommendations and requirements for laboratory preparation.
- Resource requirements for site preparation and equipment installation.
- IT and informatics specifications.
- Guidelines for selecting the individuals who will be trained to process samples and analyze data.

### Audience

This guide is intended for:

- The individual responsible for overseeing system placement and use.
- The personnel who will schedule, manage and perform the tasks required to prepare your site for equipment installation and laboratory set up.

## Preparing for Equipment Installation and Laboratory Set Up

### Laboratory Preparation

Running the MIP Assay Protocol requires two separate laboratories. These laboratories must be prepared so that the equipment can be installed and operated correctly and safely. Careful attention to the recommendations and requirements presented in [Chapter 3, \*Site Preparation\*](#) will help ensure smooth equipment installation and laboratory set up.

### Personnel Requirements

Proper installation of and training for use of the GCS 3000 TG System requires the assistance or participation of personnel at your facility to varying degrees. In this guide, we refer to these individuals as follows:

- **Site Preparation Coordinator:** Ensures that your laboratory areas are properly prepared for equipment installation. Also confirms that the system has been installed and tested.
- **System Installation Assistant:** Assists the Affymetrix representative with unpacking and placing some of the larger pieces of equipment.
- **Facilities Personnel:** Assist with unpacking and placing the plate centrifuges.

- **Information Technology Systems Administrator:** Installs the Affymetrix server (if purchased) and performs additional tasks to connect the client computer workstations to your network.
- **Data Analyzer:** Analyzes the data collected by the GeneChip® Scanner 3000 7G 4C using GeneChip® Targeted Genotyping Analysis Software.
- **Research Associates (RAs):** Run the MIP Assay Protocol. The two individuals selected must participate in a two week training session. Training will take place at your facility.

Refer to:

- [Chapter 4, Resource Requirements](#) for more details on the tasks these individuals will perform, and on the skill sets they should possess.
- [Chapter 5, System Installation and Training](#) for more information on training.

### **Additional Reagent Kits and Other Consumables**

Reagents and other consumables included for system startup will be consumed during the training session.

#### **IMPORTANT !**

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**Be sure to order additional reagent kits and consumables prior to equipment installation to ensure uninterrupted sample processing.**

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## **Obtaining More Information and Support**

### **Related Documentation**

The following related documents are included with the GCS 3000 TG System. They are provided during the training session.

- *Affymetrix GeneChip® Targeted Genotyping System User Guide* (P/N 702164)
- *Affymetrix GeneChip® Targeted Genotyping System set of Quick Reference Cards*
- *Affymetrix GeneChip® Targeted Genotyping Analysis Software User Guide* (P/N 702126)
- *Affymetrix GeneChip® Operating Software with AutoLoader User's Guide* (P/N 701439)

Chapter 2

**Equipment, Supplies, Consumables**

Chapter 2



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## Important Notice Regarding Product Support

### ABOUT QUALIFYING FOR SUPPORT

The MIP Assay Protocol has been optimized for use on the Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System using the equipment, supplies and consumables listed in this chapter.

#### IMPORTANT !

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A double asterisk (\*\*) next to an item indicates that no substitutions can be made. To qualify for support, these particular items must be used. In addition, training will be conducted only when all of the equipment, supplies and consumables are verified as present and properly installed.

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## About the Equipment, Supplies and Consumables Required

### How the Information Is Organized

Information on the equipment, supplies and consumables required is organized as follows:

- *Equipment and Software Supplied by Affymetrix for Standard System Configuration*
- *Equipment, Supplies, and Consumables Required But Not Provided*
  - *Equipment Required But Not Provided*
  - *Supplies Required But Not Provided*
  - *Consumables Required But Not Provided*
  - *Quality Control Gels*
- *Receiving the Equipment, Supplies and Consumables*

## Equipment and Software Supplied by Affymetrix for Standard System Configuration

The following items are shipped to your facility directly from Affymetrix. A double asterisk (\*\*) next to an item indicates that no substitutions can be made.

### IMPORTANT !

Do not unpack any of the equipment supplied by Affymetrix. An Affymetrix representative will unpack, install and verify the operation of the equipment and software.

### IMPORTANT !

Store GeneChip® Universal Tag Arrays at 4°C.

**Table 2.1**  
Equipment and Software Supplied by Affymetrix for Standard System Configuration

Item	Quantity	Part Number	Laboratory Location
<b>** Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System including:</b> <ul style="list-style-type: none"> <li>- GeneChip® Scanner 3000 7G 4C with 7G Instrument Control Workstation</li> <li>- GeneChip® Scanner 3000 Autoloader with 2D barcode reader</li> <li>- GeneChip® Fluidics Station 450</li> <li>- GeneChip® Hybridization Oven 640 with carriers</li> <li>- GeneChip® Operating Software (GCOS), v1.4</li> <li>- GeneChip® TG Analysis Software, version 1.5</li> <li>- TG Pre-Amp Lab Workstation with 2D barcode reader</li> <li>- TG Post-Amp Lab Workstation with SQL Server 2000 Standard Edition license and 2D barcode reader</li> </ul>	1	00-0185	Pre and Post-Amp Labs
<b>** The following GeneChip® Universal Tag Array as appropriate:</b> <ul style="list-style-type: none"> <li>- GeneChip® Universal 3K Tag Array</li> </ul>	As required	900602 (6 pk) 900578 (96 pk)	Post-Amp Lab
<ul style="list-style-type: none"> <li>- GeneChip® Universal 5K Tag Array</li> </ul>	As required	900659 (6 pk) 900660 (96 pk)	Post-Amp Lab
<ul style="list-style-type: none"> <li>- GeneChip® Universal 10K Tag Array</li> </ul>	As required	900604 (6 pk) 900580 (96 pk)	Post-Amp Lab
<ul style="list-style-type: none"> <li>- GeneChip® Universal 25K Tag Array</li> </ul>	As required	900857 (6 pk) 900858 (96 pk)	Post-Amp Lab

## Equipment, Supplies, and Consumables Required But Not Provided

### VENDOR CONTACT INFORMATION

See [Appendix B](#) for vendor contact information.

### EQUIPMENT REQUIRED BUT NOT PROVIDED

**Table 2.2**  
Equipment Required but not Provided

Equipment	Quantity	Manufacturer/ Distributor	Part Number (U.S.)	Laboratory Location
** Thermal Cyclers – see <a href="#">Table 2.3 on page 10</a> for a list of thermal cyclers that have been validated for use with the MIP Assay Protocol				
Eppendorf® Multipurpose Centrifuge 5804	2	VWR Intl.	53513-800	1 Pre-Amp Lab 1 Post-Amp Lab
Eppendorf® Deepwell Swing Bucket Rotor with two buckets	2	VWR Intl.	77888-214	1 Pre-Amp Lab 1 Post-Amp Lab
Mini Vortexer, analog	2	VWR Intl.	58816-121	1 Pre-Amp Lab 1 Post-Amp Lab
Galaxy Mini Centrifuge (interchangeable for microtubes and strip tubes)	4	VWR Intl.	37000-700	2 Pre-Amp Lab 2 Post-Amp Lab
** Aluminum Block, 96-well	4	BioSmith	81001	Pre-Amp Lab
** Pipettes and pipette tips – see <a href="#">Table 2.4 on page 11</a> for a list of pipettes and tips that have been validated for use with the MIP Assay Protocol				
Carousel Stand for Rainin® Pipettes	4	Rainin	CR-7	2 Pre-Amp Lab 2 Post-Amp Lab
Portable Pipet-Aid (110V)	1	VWR	53498-103	Post-Amp Lab
Freezer, –20°C; deep freeze; manual defrost; 17 cu ft	2	—	—	1 Pre-Amp Lab 1 Post-Amp Lab
Refrigerator, 4°C refrigerator; 6 cu ft	2	—	—	1 Pre-Amp Lab 1 Post-Amp Lab

## Thermal Cyclers

To run the MIP Assay Protocol at a throughput of 48 assays/day, you will need 4 thermal cyclers: 2 in the Pre-Amp Lab; 2 in the Post-Amp Lab.

The MIP Assay Protocol has been optimized using GeneAmp® PCR System 9700 Thermal Cyclers (96-well, gold-plated). The additional thermal cyclers listed in [Table 2.3](#) have been validated for use with the protocol in the laboratory location specified.

### IMPORTANT !

**Note the laboratory location for the thermal cyclers listed in [Table 2.3](#). Some of these instruments can be used in the Post-Amp Lab only.**

**Table 2.3**  
Thermal Cyclers Validated for Use With the MIP Assay Protocol

Manufacturer/ Distributor	Item	Part Number (U.S.)	Laboratory Location
Applied Biosystems Thermal Cyclers	GeneAmp® PCR System 9700 Thermal Cycler, 96-well, Silver	N8050001	
	GeneAmp® PCR System 9700 Thermal Cycler, 96-well, Gold-plated	4314878	Use in both the Pre- and Post-Amp Labs
	GeneAmp® PCR System 9700 Thermal Cycler, 96-well, Aluminum	4314879	
	GeneAmp® PCR System 9700 Thermal Cycler, Dual 96-well	4343176	Use in Post-Amp Lab only
BioRad Thermal Cyclers	DNA Engine® Peltier Thermal Cycler chassis only; requires 1 Alpha Unit (see BioRad Alpha Units below)	PTC-0200	Use in Post-Amp Lab only
	DNA Engine Dyad® Peltier Thermal Cycler chassis only; requires 2 Alpha Units (see BioRad Alpha Units below)	PTC-0220	
	DNA Engine Dyad® Peltier Thermal Cycler Disciple chassis only; requires 2 Alpha Units (see BioRad Alpha Units below)	PTC-0221	
	DNA Engine Tetrad® 2 Peltier Thermal Cycler Chassis only; requires 4 Alpha Units (see BioRad Alpha Units below)	PTC-0240	
BioRad Alpha Units	Alpha™ Unit: Holds 96 x 0.2 ml tubes or one 96-well plate	ALS-1296	
	Moto Alpha™ Unit: Holds one 96-well plate with integrated motorized heated lid	ALP-2296	



## Pipettes and Pipet Tips

To run the MIP Assay Protocol, you will need the following types of pipettes. The pipettes and tips validated for use with the MIP Assay Protocol are listed in [Table 2.4](#).

- Single channel pipettes
  - 0.1–2  $\mu\text{L}$
  - 0.5–10  $\mu\text{L}$
  - 2–20  $\mu\text{L}$
  - 20–200  $\mu\text{L}$
  - 100–1000  $\mu\text{L}$
- 12-channel pipettes
  - 0.5–10  $\mu\text{L}$  or 1–20  $\mu\text{L}$
  - 2–20  $\mu\text{L}$ , 5–50  $\mu\text{L}$ , 20–200  $\mu\text{L}$  or 20–300  $\mu\text{L}$
- 24-channel pipettes, electronic or manual
  - 2–20  $\mu\text{L}$
  - 10–100  $\mu\text{L}$

**Table 2.4**  
Pipettes and Pipet Tips Validated for Use With the MIP Assay Protocol

Manufacturer/ Distributor	Item	Quantity	Part Number (U.S.)	Laboratory Location
<b>SINGLE CHANNEL MANUAL PIPETTES</b>				
Rainin®	Pipet-Lite™ LTS™ Pipet, 0.1–2 $\mu\text{L}$	1	L-2	1 Pre-Amp Lab
	Pipet-Lite LTS Pipet, 0.5–10 $\mu\text{L}$	2	L-10	1 Pre-Amp Lab 1 Post-Amp Lab
	Pipet-Lite LTS Pipet, 2–20 $\mu\text{L}$	2	L-20	1 Pre-Amp Lab 1 Post-Amp Lab
	Pipet-Lite LTS Pipet, 20–200 $\mu\text{L}$	2	L-200	1 Pre-Amp Lab 1 Post-Amp Lab
	Pipet-Lite LTS Pipet, 100–1000 $\mu\text{L}$	2	L-1000	1 Pre-Amp Lab 1 Post-Amp Lab
Rainin Pipet Tips	LTS™ Pipette Tips, P-1000 barrier (8 racks of 96 tips each; 768 tips per box)	As required	RT-L1000F (1 box)	Pre and Post-Amp Labs
(use for single, 12-, and 24-channel Rainin pipettes)	LTS Pipette Tips, P-200 barrier (10 racks of 96 tips each; 960 tips per box)	As required	RT-L200F (1 box)	
	LTS Pipette Tips, P-10 barrier (10 racks of 96 tips each; 960 tips per box)	As required	RT-L10F (1 box)	

Manufacturer/ Distributor	Item	Quantity	Part Number (U.S.)	Laboratory Location
<b>12-CHANNEL MANUAL PIPETTES</b>				
Rainin Pipettes	Pipet-Lite LTS 12 Channel Pipet, 0.5–10 µL	2	L12-10	1 Pre-Amp Lab 1 Post-Amp Lab
	Pipet-Lite LTS 12 Channel Pipet, 2–20 µL	2	L12-20	1 Pre-Amp Lab 1 Post-Amp Lab
	Pipet-Lite LTS 12 Channel Pipet, 20–200 µL	2	L12-200	1 Pre-Amp Lab 1 Post-Amp Lab
Gilson Pipettes	Pipetman® Ultra, U12-20, P20, 1–20 µL	2	F21041	1 Pre-Amp Lab 1 Post-Amp Lab
	Pipetman Ultra, U12-300, 20–300 µL	2	F21043	1 Pre-Amp Lab 1 Post-Amp Lab
Gilson Pipet Tips	Gilson Diamond® sterilized extra long filter tips, 2-200 µL	As required	D200ST	Pre- and Post-Amp Labs
	Gilson Diamond sterilized filter tips, 30–300 µL	As required	DF300ST	
Eppendorf Pipettes	Research® Series 2100, 0.5–10 µL	2	022453947	1 Pre-Amp Lab 1 Post-Amp Lab
	Research Series 2100, 10–100 µL	2	022453963	
Eppendorf Pipet Tips	epT.I.P.S. Filter, 0.1–10 µL	As required	022491211	Pre- and Post-Amp Labs
	epT.I.P.S. Filter, 2–100 µL	As required	022491237	
<b>24-CHANNEL PIPETTES, MANUAL AND ELECTRONIC</b>				
Manual or electronic 24 channel pipettes can be used. Electronic recommended for better uniformity when mixing. A minimum and maximum quantity of electronic pipettes is listed. If using the minimum, you will have to reprogram the pipettes throughout the protocol. If using the maximum, you will not have to reprogram the electronic pipettes.				
Rainin	2–20 µL			
	- Pipet-Lite LTS 24 Channel Pipet (manual)	2	L24-20	1 Pre-Amp Lab 1 Post-Amp Lab
	- EDP3™ + Elect LTS™ Pipet 24 Channel (electronic)	2 to 4	E24-20	
	10–100 µL			
	- Pipet-Lite LTS 24 Channel Pipet (manual)	2	L24-100	1 Pre-Amp Lab 1 Post-Amp Lab
- EDP3+ Elect LTS Pipet 24 Channel (electronic)	2 to 4	E24-100		
	E3 Rapid Charge Stand	2 or 4	E3-RCS	

## SUPPLIES REQUIRED BUT NOT PROVIDED

**Table 2.5**

Supplies Required but not Provided

Small Supplies	Quantity	Manufacturer/ Distributor	Part Number	Laboratory Location
Cube Rack, polypropylene (4/pack)	2 packs	VWR Intl.	60985-444	1 Pre-Amp Lab 1 Post-Amp Lab
MicroTube Rack, 80-well (5/pack)	2 packs	VWR Intl.	10011-284	1 Pre-Amp Lab 1 Post-Amp Lab
96-place rack for 0.2 mL tubes (5/pack)	3 packs	USA Scientific	2300-9602	Pre- and Post- Amp Labs
Preparation racks for PCR thin-walled tubes (assorted colors)	1 case	Fischer Scientific	05-541-55	Pre- and Post- Amp Labs
MiniAlarm Timer/Stopwatch	2	VWR Intl.	62344-585	1 Pre-Amp Lab 1 Post-Amp Lab
Lab Coats, dedicated	8	—	—	Pre and Post-Amp Labs
Optional: Ice Bucket, red	1	VWR Intl.	35751-205	Post-Amp Lab
Note: You will need an ice bucket dedicated to the Post-Amp Lab.				
Optional: Ice Container, red (8.5" x 16" x 3")	1	VWR Intl.	35751-216	Post-Amp Lab
Note: You will need an ice container dedicated to the Post-Amp Lab.				
Optional: Ice Container, green (8.5" x 16" x 3")	1	VWR Intl.	35751-218	Pre-Amp Lab
Note: You will need an ice container dedicated to the Pre-Amp Lab.				
Calculator	2	—	—	1 Pre-Amp Lab 1 Post-Amp Lab
Scissors	2 pairs	—	—	1 Pre-Amp Lab 1 Post-Amp Lab
Distilled, deionized water supply or carboys (for cleaning the fluidics station)	—	—	—	Post-Amp Lab

## CONSUMABLES REQUIRED BUT NOT PROVIDED

### GeneChip® SNP Kits

One GeneChip SNP Kit includes reagents sufficient to process 24 reactions. The specific kit required to process your samples is determined by the assay panel you are using.

#### IMPORTANT !

Kit components are shipped in multiple boxes. Store each box as directed on the label.

### Additional Consumables

The quantities listed for consumables are sufficient to process 600 reactions including controls.

**Table 2.6**

Additional Consumables – Quantities sufficient to process approximately 600 reactions

Item	Quantity	Manufacturer /Distributor	Part Number (U.S.)	Laboratory Location
** Both of the Taq DNA Polymerases listed below are required.				
- Stratagene Taq DNA Polymerase, 200 µL/vial	As needed	Stratagene	929197 (1 vial)	Pre-Amp Lab
- Clontech TITANIUM™ Taq Polymerase	As needed	Clontech	639208 (100 rxns)	Post-Amp Lab
100 rxn size is sufficient for processing 24 reactions (with 10% extra)			639209 (500 rxns)	
** PCR Plates (96-well) and barcode labels — see <a href="#">PCR Plates on page 15</a> and <a href="#">Table 2.7 on page 16</a> for the PCR plate vendors and barcode label supplier validated for use with the MIP Assay Protocol.				
** Clear Film for Arrays (81 labels/sheet; 10 sheets per pack)	1 pack	Affymetrix		Post-Amp Lab
** MicroAmp® Clear Adhesive Films (100 films/pk)	3 pks	Applied Biosystems	4306311 (1 pack)	Pre and Post-Amp Labs
Pipettes, 5 mL, disposable, individually wrapped (200/case)	1 case	VWR	20171-046	Post-Amp Lab
Pipettes, 10 mL, disposable, individually wrapped (200/case)	1 case	VWR	20171-042	Post-Amp Lab
PCR 12-well Tube Strips with Strip Bubble Caps (80/pack)	1 pk	VWR	53509-306 (1 pack)	Pre and Post-Amp Labs
Eppendorf Color-Coded Safe-Lock Microcentrifuge Tubes, amber, 2.0 mL (500/case)	2 cases	VWR	20901-541	Post-Amp Lab
Eppendorf Color-Coded Safe-Lock Microcentrifuge Tubes, natural, 2.0 mL (500/case)	2 cases	VWR	20901-540	Pre and Post-Amp Labs
Reagent Reservoirs, 50 mL (200/case)	1 case	VWR Intl.	53504-035 (1 case)	Pre and Post-Amp Labs
Microtube Tough Spots® for 0.5–2.0 mL tubes, white (1000/roll)	1 roll	USA Scientific	9185-0000 (1 roll)	Post-Amp Lab

**Table 2.6**

Additional Consumables – Quantities sufficient to process approximately 600 reactions

Item	Quantity	Manufacturer /Distributor	Part Number (U.S.)	Laboratory Location
Corning Conical Tubes, 15 mL (500/case)	1 case	VWR Intl.	21008-670 (1 case)	Pre and Post-Amp Labs
Gloves, 100/box	6 boxes	—	—	Pre and Post-Amp Labs
Bench Pads	1 case	VWR Intl.	56616-026	Pre and Post-Amp Labs
Kimwipes® (12" x 12")	2 packs	VWR Intl.	21905-011 (1 pack)	Pre and Post-Amp Labs
Marking pens, one of the following types <ul style="list-style-type: none"> <li>• Water soluble</li> <li>• Fluorescent</li> </ul>	1 pack	Fischer Scientific	13-380-15A or 13-384	Post-Amp Lab
Marking pens, permanent, fine point, assorted colors	12 pens	—	—	6 Pre-Amp Lab 6 Post-Amp Lab
Razor blades	1 pack	—	—	Post-Amp Lab
Aluminum foil	1 roll	—	—	Post-Amp Lab
Compressed air (canned)	2 cans	—	—	Post-Amp Lab
Tacky Floor Mats	1 case	VWR Intl.	12777-112 (1 case)	Post-Amp Lab

### PCR Plates

To run the MIP Assay Protocol, you will need a supply of 96-well PCR plates with specific barcode labels. To facilitate sample tracking, the plates are referred to using specific terminology throughout the various stages of the protocol. These designations and the barcode prefix associated with each type of plate is listed below.

- Anneal Plate – ANN barcode
- Assay Plates – ASY barcode
- Label Plates – LBL barcode
- Hyb Plates – HYB barcode

#### IMPORTANT !

If you purchase plates from a vendor other than Axygen, you must purchase barcode labels from Axygen to apply to your plates. The plates supplied by Axygen come with the barcode labels already affixed to them.

**Table 2.7**

PCR Plates and Barcode Labels Validated for Use With the MIP Assay Protocol – Quantities sufficient to process approximately 600 reactions

Manufacturer/ Distributor	Item	Quantity	Part Number (U.S.)	Laboratory Location
<b>PCR PLATES</b>				
Axygen distributor	Anneal PCR Plates, 96-well thin-wall (with ANN barcode affixed; 50/case)	1 case	321-63-ANN (1 case)	Pre-Amp Lab
	Assay PCR Plates, 96-well thin-wall (with ASY barcode affixed; 50/case)	1 case	321-63-ASY (1 case)	Pre-Amp Lab
	Label PCR Plates, 96-well thin-wall (with LBL barcode affixed; 50/case)	1 case	321-63-LBL (1 case)	Post-Amp Lab
	Hybridization PCR Plates, 96-well thin-wall (with HYB barcode affixed; 50/case)	1 case	321-63-HYB (1 case)	Post-Amp Lab
Greiner (from E & K Scientific)	PCR Plate, 96 Well, Half Skirt (no barcode; 40/case)  Note: If purchasing Greiner plates, you must also purchase barcode labels from Axxygen (listed below)	8 cases	EK-19280	Pre- and Post-Amp Labs
<b>BARCODE LABELS</b>				
Axygen distributor	Barcode Labels for Sample Plates	As required	BC-LABEL-SMP	Pre-Amp Lab
	Barcode Labels for Anneal Plates		BC-LABEL-ANN	Pre-Amp Lab
	Barcode Labels for Assay Plates		BC-LABEL-ASY	Pre-Amp Lab
	Barcode Labels for Label Plates		BC-LABEL-LBL	Post-Amp Lab
	Barcode Labels for Hyb Plates		BC-LABEL-HYB	Post-Amp Lab

## QUALITY CONTROL GELS

### Training Requirements

As part of the training session, quality control (QC) gels are run on each plate of samples processed. Running QC gels helps the trainer determine the source of issues that may arise.

Your laboratory must have gel electrophoresis capability and a gel imaging system. Gel recommendations are listed below.

### QC Gel Recommendations

We recommend running two QC gels per assay:

- While learning to run the MIP Assay Protocol
- For troubleshooting purposes

## Precast Gel Recommendations

We recommend using one of the following precast gels and gel system:

### Biorad Precast Gels and Gel Box System

**Table 2.8**

Recommended Biorad Precast Gels and Gel Box System

Item	Part Number	For Training	Ongoing
Wide Mini ReadySub-Cell GT Cell	170-4489	Two cells required to concurrently run two gels	Two cells required to concurrently run two gels
ReadyAgarose 96 Plus 3% TBE Gel	161-3062	8 gels	1 gel/24 assays

### Invitrogen Precast Gels and Gel System

**Table 2.9**

Recommended Invitrogen Precast Gels and Gel System

Item	Part Number	For Training	Ongoing
Power supply and gel tray recommendations: <ul style="list-style-type: none"> <li>• One Mother Base</li> <li>• Three Daughter Bases</li> </ul>	Mother Base: EB-M03 Daughter Bases: EB-D03	One Mother Base and Three Daughter Bases	One Mother Base and Three Daughter Bases
E-Gel 48 4% Agarose (8 gels/box)	G8008-04	8 boxes	2 gels/24 assays

## Self-Cast Gel Recommendations

If casting your own gels, we recommend following these guidelines:

- Use acrylamide or a high-resolution agarose.
- For agarose gels, we recommend preparing a 4% Agarose SFR gel using Agarose SFR by Amresco, part number J234-100G.

## Receiving the Equipment, Supplies and Consumables

### **Instruments, Equipment and Supplies**

The unpacking, installation and verification of all instruments, computer workstations, other equipment, and supplies will be done by a representative from Affymetrix.

### **Consumables**

Store all consumables as stated on the product insert.



Chapter 3

**Site Preparation**

Chapter 3



## Laboratory Requirements

### Overview

To successfully use the Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System (GCS 3000 TG System), two fully separated laboratories are required. We refer to these two areas as the Pre-Amp Lab and the Post-Amp Lab.

The purpose of having two separate laboratories is to prevent sample contamination, the most likely source being previously amplified PCR product. In the MIP Assay Protocol, common primers are used to amplify all assay products. The resulting product must be contained to the Post-Amp Lab. Any PCR product that gets back into the Pre-Amp Lab can contaminate your entire process.

#### IMPORTANT !

---

**To minimize the possibility of sample contamination, we strongly recommend you protect the Pre-Amp Lab from external PCR products.**

**The minimum separation required between the Pre-Amp Lab and the Post-Amp Lab is a solid wall and a door. Rooms that are further separated in distance by intervening labs or floors is optimal.**

**See [Preventing Sample Contamination on page 27](#) for a list of recommendations.**

---

If the rooms are adjoining, the Pre-Amp Lab must have a separate entrance and exit, and the Post-Amp Lab must have a separate exit. This facilitates a single-direction workflow from the Pre- to the Post-Amp Lab.

## Pre-Amp Lab

### Recommended Square Footage and Bench Space

For the Pre-Amp Lab, we recommend the following:

- Approximately 105 sq ft
- Two lab benches (approximately 72" L X 30" D each)

#### NOTE

---

**The square footage and layout for the Pre-Amp Lab are suggestions only.**

---

### Computer Network Connections

One computer, the TG Pre-Amp Lab Workstation, will be installed in this lab. Its approximate location in relation to instruments and the work area is shown in [Figure 3.1](#). This computer must have a network connection to the server that is part of the GCS 3000 TG System. Be sure that there is a network connection in the immediate vicinity for this computer.

## Decontamination Recommendations

Before using the GCS 3000 TG System for the first time, ensure that the area designated as the Pre-Amp Lab is free of PCR products. We strongly recommend using a PCR DNA degradation solution such as *DNAZap* by Ambion to decontaminate the area.

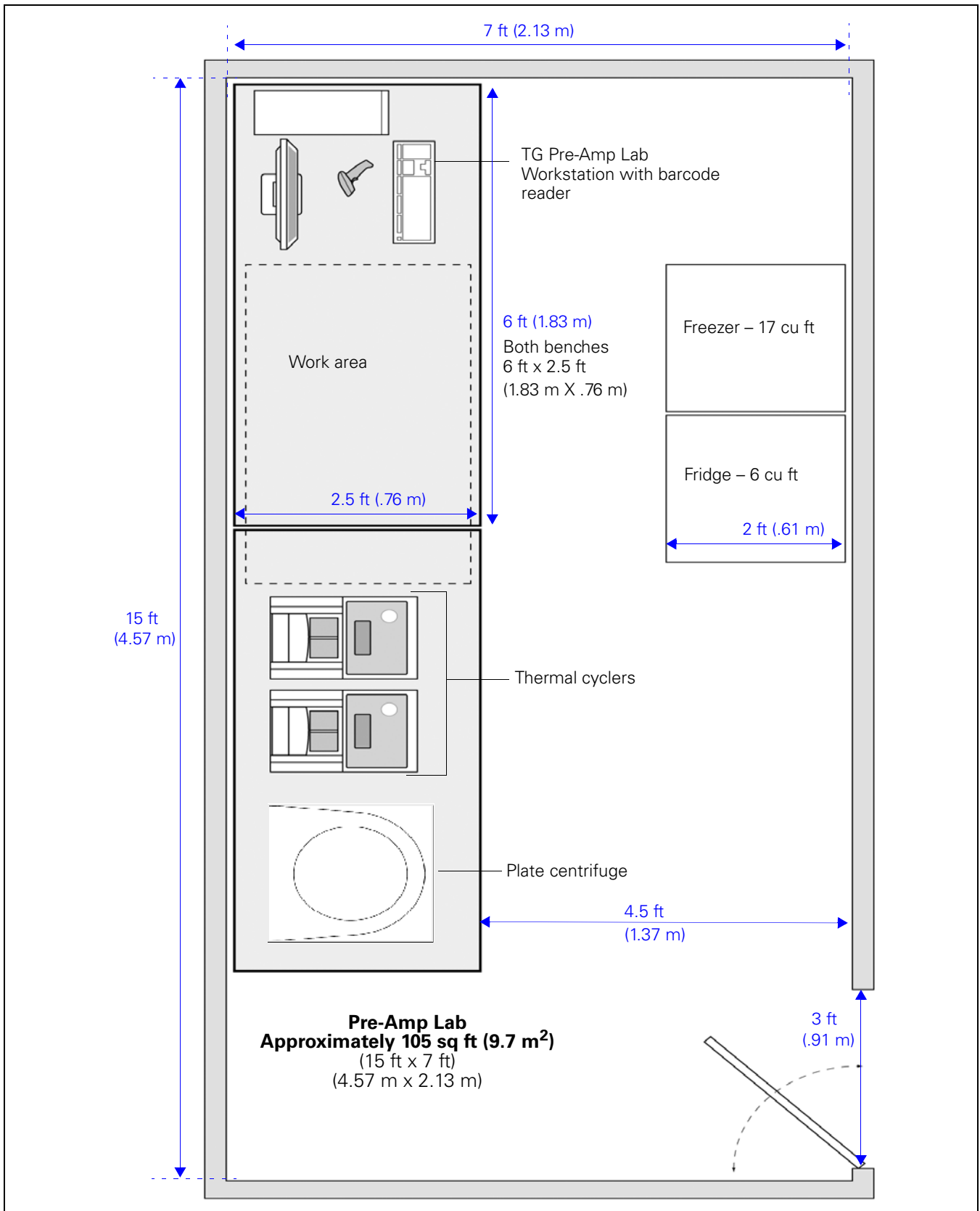
We also recommend keeping the Pre-Amp Lab free of PCR products by decontaminating it on a regular basis.

See [Appendix B](#) for vendor contact information.

## Equipment Layout Recommendations

[Figure 3.1](#) shows the recommended placement of equipment and the work area. The equipment that will be located in this area includes:

- The TG Pre-Amp Lab Workstation (includes a barcode reader)
- Two thermal cyclers
- Freezer, 17 cu ft
- Refrigerator, 6 cu ft
- Plate centrifuge



**Figure 3.1**  
 Pre-Amp Lab Space Recommendations and Suggested Layout  
 (Miscellaneous equipment such as pipettes are not shown in this figure)

## Post-Amp Lab

### Recommended Square Footage and Bench Space

For the Post-Amp Lab, we recommend the following:

- Approximately 247 sq ft
- Five lab benches (approximately 72" L X 30" D each)

---

**NOTE** 

**The square footage and layout for the Post-Amp Lab are suggestions only.**

---

### Computer Network Connections

Two computer network connections are required for each of the following workstations:

- Instrument Control Workstation
- TG Post-Amp Lab Workstation

The suggested locations of these workstations in relation to instruments and the work area are shown in [Figure 3.2](#). Network connections in the immediate vicinity must be available for both of these workstations.

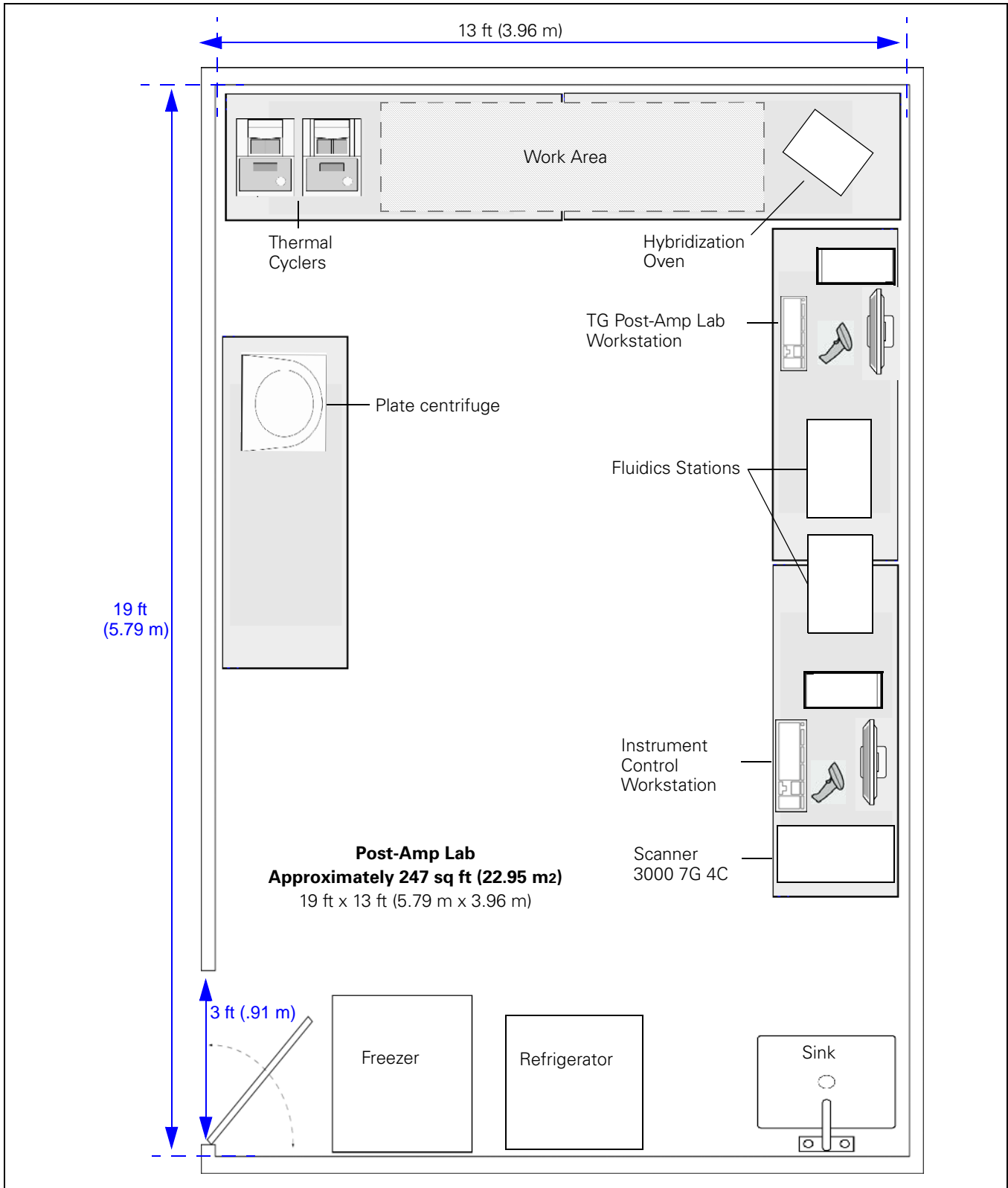
### DI Water Supply and Sink

The fluidics station requires maintenance on a weekly and monthly basis. DI water and a sink to dispose of waste is required for both of these procedures. As such, we recommend a sink and a source of DI water be located in this lab.

### Equipment Layout Recommendations

[Figure 3.2](#) shows the recommended placement of equipment and work areas. The equipment that will be located in this area includes:

- GeneChip® Scanner 3000 7G 4C
- Instrument Control Workstation (controls the scanner and the fluidics station)
- TG Post-Amp Lab Workstation
- Two Fluidics Stations 450
- Affymetrix Hybridization Oven
- Two thermal cyclers
- Plate centrifuge
- Freezer, 17 cu ft
- Refrigerator, 6 cu ft



**Figure 3.2**  
 Post-Amp Laboratory Space Recommendations and Suggested Layout  
 (Miscellaneous equipment such as pipettes not shown in this figure)

# Affymetrix Instrument Site Requirements

## Overview

The following information is intended to provide you with a brief overview of the site requirements for your Affymetrix instruments and computer workstations.

Please have space available for these system components. The dimensions and weight of each component are listed below.

## Receipt and Installation

Do not unpack the Affymetrix instruments or computer workstations. Unpacking should only be performed by an authorized Affymetrix representative.

## Electrical Requirements

All instruments run on 120 Vac ( $\pm 10\%$ ) in North America and Japan, and on 220 Vac ( $\pm 10\%$ ) in Europe.

## GeneChip® Scanner 3000 7G 4C

The Scanner 3000 7G 4C has no special power requirements. It does not require a dedicated power supply.

### Physical Dimensions and Weight

- Footprint: 13" W x 22" D x 18" H (33 cm x 56 cm x 46 cm)
- Weight: approximately 70 lb (31.8 kg)

## Instrument Control Workstation

### Physical Dimensions and Weight

- Computer Monitor
  - Footprint: 20.25" W x 11" D x 21" H (52 cm x 28 cm x 54 cm)
  - Weight: approximately 26 lb (11.8 kg)
- Computer Tower
  - Footprint: 7.25" W x 18" D x 17" H (19 cm x 46 cm x 44 cm)
  - Weight: approximately 30 lb (13.6 kg)

## GeneChip® Fluidics Station 450

### Physical Dimensions and Weight

- Footprint: 31.1" W x 20" D x 15" H (79 cm x 48 cm x 38 cm)
- Weight: approximately 76 lb (34.5 kg)



## GeneChip® Hybridization Oven

### Physical Dimensions and Weight

- Footprint: 18" W x 15" D x 19" H (45.7 cm x 38 cm x 48.3 cm)
- Weight: approximately 50 lb (22.7 kg)

## Preventing Sample Contamination

### Recommendations

To prevent sample contamination, we strongly suggest that everyone entering the Pre- and Post-Amp Labs follow these guidelines:

- Keep the Pre-Amp Lab free of PCR products by cleaning it on a regular basis with a PCR DNA degradation solution such as DNAZap by Ambion. See [Appendix B](#) for vendor contact information.
- Do not move equipment between the Pre- and Post-Amp Labs. Keep dedicated equipment in both labs.
- Always assume that all surfaces in the Post-Amp Lab are contaminated with amplified PCR product, and that all items that enter the Post-Amp Lab become contaminated with product.
- Never bring any items that have been in the Post-Amp Lab into the Pre-Amp Lab.
- Keep dedicated lab coats for the Pre-Amp Lab and the Post-Amp Lab.
- Use proper gowning procedures.
- Always wear gloves when handling equipment or samples in either lab.
- Print separate copies of the protocol for each lab.
- If both labs are connected by a door, do not return to the Pre-Amp Lab via the Post-Amp Lab.
- Place tacky mats at the entrance and exit of the Post-Amp Lab.
- If available, have positive airflow in the Pre-Amp Lab relative to the Post-Amp Lab.
- If possible, restrict Pre-Amp Lab access to authorized personnel only. An example of unauthorized personnel would be cleaning staff.



Chapter 4

**Resource Requirements**

Chapter 4



## Overview of Resource Requirements

This section includes a description of the resources required to successfully install the required equipment and use the Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System. These resources include:

- Site Preparation Coordinator
- System Installation Assistant
- Facilities Personnel
- Research Assistants
- Data Analyzer
- Information Technology (IT) Systems Administrator

### SITE PREPARATION COORDINATOR

We ask that you designate one individual to act as the Site Preparation Coordinator. This individual is responsible for ensuring that the lab requirements described in [Chapter 3, Site Preparation](#) have been met. Before we can schedule a date for equipment installation, the Site Preparation Coordinator must complete, sign and return the [Confirmation of Site Preparation](#) form located in [Appendix A](#).

Once installation is complete, the Site Preparation Coordinator and the Affymetrix representative performing the installation complete and sign the [Confirmation of Equipment Installation and Laboratory Set Up](#) form ([Appendix A](#).) Each party retains a copy of the completed form.

### SYSTEM INSTALLATION ASSISTANT

One able-bodied individual is required to help unpack and place some of the larger pieces of equipment. This individual must be trained to use proper lifting techniques.

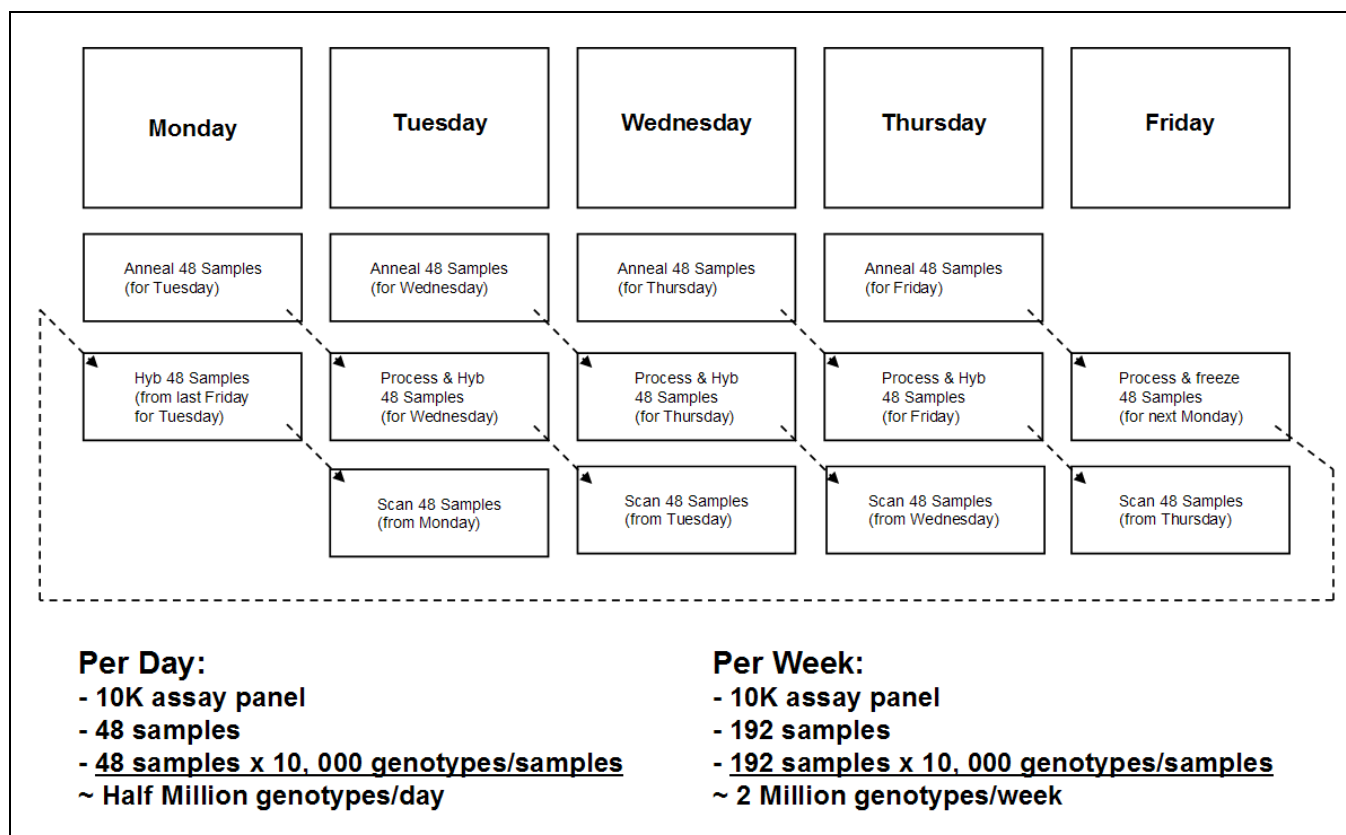
### FACILITIES PERSONNEL

Two individuals from your Facilities group are required to unpack, lift and place the plate centrifuges. These instruments weigh approximately 80 lbs each; therefore, these individuals must be trained to use proper lifting techniques.

## RESEARCH ASSISTANTS

To process samples at maximum throughput, two full-time employees are required. Maximum throughput (including recommended controls) is 192 assays per 5-day work period (refer to [Figure 4.1](#)). In this document, we refer to these individuals as Research Assistant A (RA-A) and Research Assistant B (RA-B.) The RAs work together in an overlapping fashion to run the MIP Assay Protocol.

The following table shows how the 5 high-level steps of the protocol are divided between the RAs. It also shows the approximate amount of time required to perform each step. These estimates assume that the RAs have successfully completed training for this product and are comfortable performing all aspects of the protocol.



**Figure 4.1**  
Standard Workflow

Chapter 5

**System Installation and Training**

Chapter 5





## Overview

### Prerequisites

Before a date can be scheduled for equipment installation and laboratory set up, Affymetrix must:

- Verify that your laboratory areas have been prepared based on the guidelines presented in [Chapter 3, Site Preparation](#).
- Receive a completed copy of the [Confirmation of Site Preparation](#) in [Appendix A](#). Included is confirmation that the resources listed in [Chapter 4, Resource Requirements](#) have been selected and are available.

### When

When all of the prerequisites have been met, the schedule for equipment installation and laboratory set up will be confirmed. Installation and set up will take approximately one to two days.

### Who

Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System installation is performed by the following individuals:

- Affymetrix representatives (with the assistance of your designated System Installation Assistant and Facilities personnel.) See [System Installation](#) below for more information.
- Your designated Systems Administrator (with assistance from Affymetrix IT staff as required.) See [Informatics Hardware Installation on page 36](#) for a description of the tasks involved.

## System Installation

### Who

Affymetrix representatives will unpack, place, install and verify operation of:

- GeneChip® Scanner 3000 7G 4C
- 7G Instrument Control Workstation
- The TG Post-Amp Workstation
- The TG Pre-Amp Workstation
- All other equipment and supplies

Your designated System Installation Assistant, Facilities personnel, and IT Systems Administrator will help unpack, place, and install these items.

## Informatics Hardware Installation

Below is a summary of the Information Technology (IT) tasks that must be performed to install the Affymetrix server (if purchased) and computers. Your Systems Administrator will be given full server access (administrator login and password.)

The software required for the GCS 3000 TG System is installed on the server (if purchased) and on the computers before you receive them. The maximum time required for computer and server installation is approximately three full days.

### Server Installation

If a server was purchased, your Systems Administrator must:

- Install the server on your network.
- Configure host name parameters, Domain Name Service, and IP address settings to incorporate the server into your local area network.
- Install any anti-virus and backup software specific to your site.

### Computer Installation

Your Systems Administrator must:

- Install the computers on your network.
- Configure computer names, Domain Name Service, and IP address settings.
- Install any anti-virus and backup software specific to your site

Routine backups of the server should be performed. Affymetrix IT staff will work with your IT personnel to determine an appropriate backup method that will co-exist with your existing backup strategy.

### Security Settings

The Affymetrix representative will ask what application-level security setting should be used. Your options are:

- Allow all users on the Domain to access the software.
- Allow only specific users to access the software.

The individual who will manage the list of allowable users must be identified.

### Instrument Control Workstation

You may want to create additional user accounts and change the name of the Instrument Control Workstation.

---

**IMPORTANT !**

**Changing the workstation name and adding additional user accounts must be coordinated with the Affymetrix Instrument Service Engineer installing this instrument. If not done properly, these changes can interfere with proper instrument operation.**

---

## Training

**NOTE** 

The standard training program is described below. The training program for your site may vary.

### STANDARD TRAINING PROGRAM

The standard training program is one week in duration. Two rounds of the assay are completed with a total of 48 arrays processed (24 arrays per round). Support personnel will be on-site for the first three days to conduct hands-on training, followed by an independent run of the assay by the trainees. Two trainees are required to be in attendance for the full five days. GeneChip<sup>®</sup> SNP Kits for training must be ordered in advance.

#### Goals

The goals of this training program are to:

- Familiarize users with the lab set up required.
- Provide an understanding of the assay workflow and process.
- Ensure that the system is operating correctly.
- Train users on the use of the GeneChip<sup>®</sup> Targeted Genotyping Analysis Software (GTGS).
- Provide information on analysis and data collection.

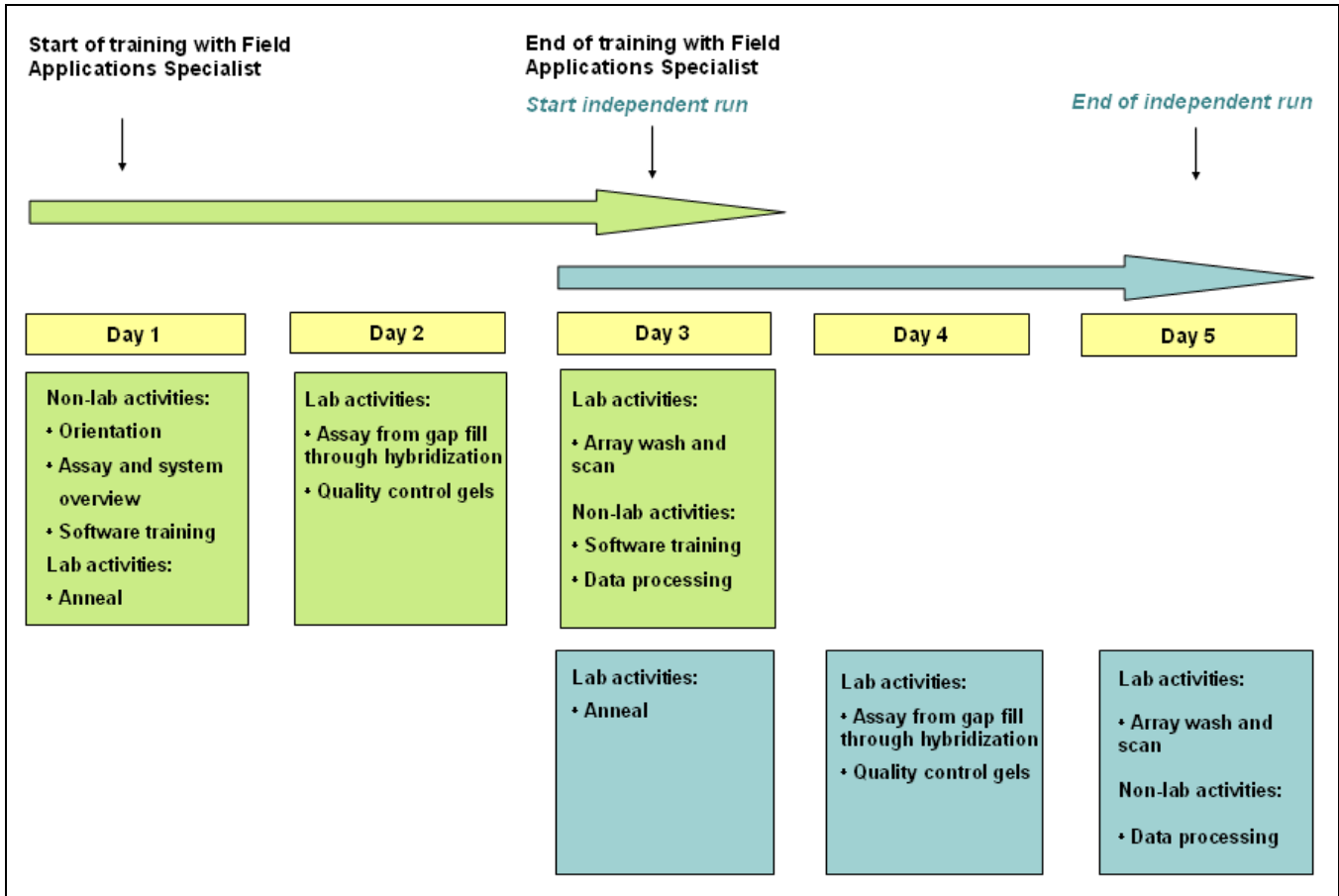
#### Schedule of Activities

The activities listed for the standard training program are guidelines only.

As shown in [Figure 5.1](#), the training begins with an overview of the MIP Assay Protocol and how the assay works. Also included is an overview of the GTGS as it is used to set up and run experiments. In the afternoon, you will enter the lab and begin your first experiment.

The trainer will demonstrate each step of the protocol, and the trainees will repeat the procedure after each demonstration.

Instruction on using the GTGS to perform data analysis is interspersed throughout the training period.



**Figure 5.1**  
Standard Training Program

## SAMPLE REQUIREMENTS

### For Training

During training, your RAs learn how to perform the MIP Assay Protocol by running standard DNA with a standard assay panel and GeneChip® Universal Tag Arrays supplied by Affymetrix.

### Criteria for Genomic DNA

Guidelines for your own genomic DNA samples are as follows:

- Concentration
  - For best results, we recommend normalizing all samples to a single concentration of 150 ng/μL. This concentration matches that of the Kit Control DNA included in each GeneChip SNP Kit.
  - We also recommend using PicoGreen to determine your sample concentration.

### IMPORTANT !

---

**We do not recommend sample concentrations below 100 ng/μL.**

---

- Amount of DNA
  - For assay panels less than or equal to 5K: 2 μg/assay (volume 15 μL/assay)
  - For assay panels greater than 5K: 4 μg/assay (volume 30 μL/assay)
- Buffer: 1X TE buffer
- Extraction and purification methods:
  - QIAGEN; QIAamp DNA Blood Maxi Kit
  - Phenol-chloroform extraction
  - Gentra PUREGENE

### General Requirements

Genomic DNA samples to be processed using the MIP Assay Protocol must:

- Not be amplified.
- Be free of inhibitors.
 

The extraction and purification method should render genomic DNA that is generally salt-free. High concentrations of certain salts can inhibit enzyme reactions.
- Not be contaminated with other human genomic DNA sources or with genomic DNA from other organisms.
- Not be degraded.

## **Sources of Genomic DNA**

The following sources of genomic DNA have been successfully processed using the MIP Assay Protocol in our laboratories:

- Blood
- Cell line
- Semen
- Tissue including brain, liver and spleen

Chapter 6

**IT and Informatics Specifications**

Chapter 6





## Hardware Components

### 7G Instrument Control Workstation

The Instrument Control Workstation is installed in the Post-Amp Lab. It controls the scanner and the fluidics station. This workstation includes:

- Microsoft® Windows XP operating system with Service Pack 1 or 2
- Applications:
  - GeneChip® Operating Software (GCOS)
  - Microsoft® Office Professional
  - Norton Antivirus™

### TG Post-Amp Lab Workstation

The TG Post-Amp Lab Workstation includes:

- Standard configuration
- Microsoft Windows XP operating system with Service Pack 2
- Applications:
  - Microsoft® SQL Server 2000, personal edition
  - Java 2 Platform, Standard Edition, release 1.4.2
  - Apache Tomcat Web Server 4.9.1
  - GeneChip® Targeted Genotyping Analysis Software (GTGS) (Java-based)
  - Microsoft Office Professional
  - Norton Antivirus
  - GCOS Runtime Libraries

### TG Pre-Amp Lab Workstation

The TG Pre-Amp Lab Workstation includes:

- Microsoft Windows XP operating system with Service Pack 2
- Applications:
  - Microsoft Office Professional
  - Norton Antivirus
  - Java Runtime environment
  - GTGS (Java-based)
  - GCOS Runtime Libraries

## Upgrading to Microsoft Windows XP Service Pack 2

To upgrade the workstations to Microsoft® Windows XP Service Pack 2, follow the instructions included with GTGS and GCOS.

## Network Requirements

### Server

Purchasing an Affymetrix TG server is optional. If purchased, network requirements are:

- Standard ethernet connection
- Does not require Internet connectivity to operate

### Client Computers

- Standard ethernet connections
  - One in the Pre-Amp Lab (TG Pre-Amp Lab Workstation)
  - Two in the Post-Amp Lab (TG Post-Amp Lab Workstation and Scanner 3000 7G 4C Workstation)

---

**IMPORTANT !**

**All computers must be connected to the same Microsoft Windows NT domain.**

---

## About the Database

### Backing Up the Database

The GTGS database is automatically backed up on a routine basis to another hard drive on the same computer. This backup can be copied to another location.

### What is Stored on the System Database or Server

The analysis results generated by GTGS are stored on the TG Post-Amp Lab Workstation (or Affymetrix TG server if purchased.) The .cel files, which are parsed when transferred from GCOS into GTGS, remain on the Instrument Control Workstation.

### Space Required

The amount of space required to store the data and other information related to 1,000,000 genotypes is approximately 600 MB.

## Backup Recommendations

### What Files Should be Backed Up

Regular backups of the database are performed automatically. Should you lose the database and the backup, your projects can be recovered if you also save to file the following information:

- Sample plate and sample info files
- The assay panel file (on the CD ROM included with the assay panel)
- From the scanner:
  - .dat files
  - .cel files
- From GTGS for each project:
  - Array data experiment list
  - The experiments table from the saved cluster results
  - The genotypes file
  - The genotype settings file

## Data Formats and Genotype Call Codes

### DATA INPUT FORMATS

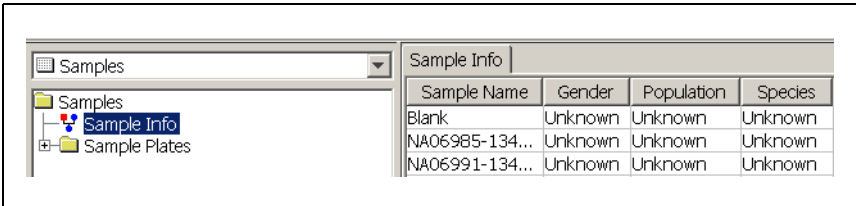
Sample data is input into the software using:

- Sample Info Files (optional)
- Sample Plate Files (required)

### Sample Info File Format Required (for Genomic DNA Sample Data)

Sample Info files are optional. The information they contain includes the sample name, gender, population and species.

If you decide not to create Sample Info files, the gender, population and species for each sample will be designated as Unknown when sample plate files are imported (Figure 6.1).

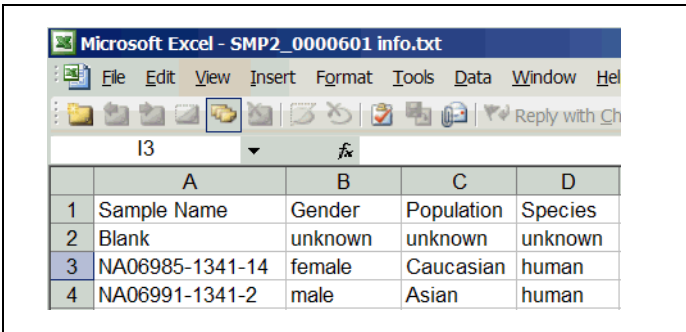


Sample Name	Gender	Population	Species
Blank	Unknown	Unknown	Unknown
NA06985-134...	Unknown	Unknown	Unknown
NA06991-134...	Unknown	Unknown	Unknown

**Figure 6.1**  
Sample Info Icon and Table

Guidelines for creating Sample Info files are:

- Input must be in the form of a tab delimited text file created in a text editor such as Microsoft® Office Excel®.
- Use the column headings and format shown in [Figure 6.2](#).
- Samples names:
  - Maximum length is 50 characters.
  - Cannot contain the following characters: \ / : ; \* ? " < > | ' , { } [ ]
  - Can appear only once in a Sample Info file
- Gender can be Male, Female or Unknown.



The screenshot shows a Microsoft Excel spreadsheet titled "SMP2\_0000601 info.txt". The spreadsheet has four columns labeled A, B, C, and D. The data is as follows:

	A	B	C	D
1	Sample Name	Gender	Population	Species
2	Blank	unknown	unknown	unknown
3	NA06985-1341-14	female	Caucasian	human
4	NA06991-1341-2	male	Asian	human

**Figure 6.2**  
Example of Sample Info File

### Sample Plate Format Required (for Genomic DNA Sample Data)

Sample plate files contain information about your samples and where they are located on sample plates. This information includes well designation, sample name, sample type and concentration.

Guidelines for creating Sample Plate files are:

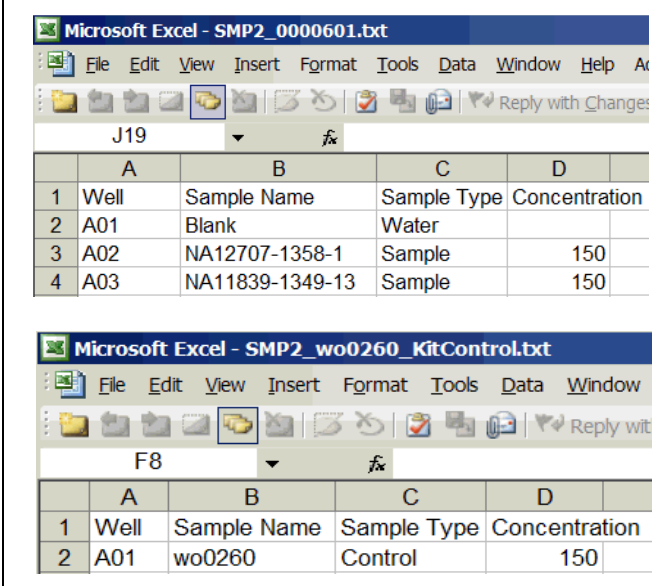
- Input must be in the form of a tab delimited text file created in a text editor such as Microsoft Office Excel.
- Use the column headings and format shown in [Figure 6.3 on page 47](#). Sample types can be sample, control and water.
- Each text file should correspond to one sample plate only.
- Well designations must be based on a 96-well plate format (A1 through H12.)
- The sample plate does not need to be full. Do not include empty wells in the file.
- Samples name guidelines:
  - Must be unique for each sample
  - Maximum length is 50 characters
  - Cannot contain the following characters: \ / : ; \* ? " < > | ' , { } [ ]

#### IMPORTANT !

**Assign a unique name to each sample. GTGS compares samples based on sample name. If different samples are assigned the same name, and if those samples are clustered together, the repeatability metrics calculated by GTGS will be incorrect.**

**NOTE**

For text files containing control DNA information, we recommend including the lot number in the filename. For example, **SMP2\_wo0260\_KitControl.txt**. You can use this filename as a the sample plate barcode later in the protocol.



Microsoft Office Excel® file with sample plate layout. Must be saved as a tab-delimited text file (.txt).

	A	B	C	D
1	Well	Sample Name	Sample Type	Concentration
2	A01	Blank	Water	
3	A02	NA12707-1358-1	Sample	150
4	A03	NA11839-1349-13	Sample	150

File with control DNA information. Must be saved as a tab-delimited text file (.txt).

	A	B	C	D
1	Well	Sample Name	Sample Type	Concentration
2	A01	wo0260	Control	150

**Figure 6.3**

Example of Text Files with Sample Plate Layout and Control DNA Information

## DATA OUTPUT FORMATS

Formats available for exporting genotype data and settings are:

- Standard – one assay per row (Figure 6.4)
- Long – one genotype per row (Figure 6.5)
- Short – one genotype per row; less information than the Long format; reports genotypes in letter format (Figure 6.6)
- Assay Allele Signals (Figure 6.7)

The assay allele signals output includes the signals used to calculate the genotypes. For example, you can use this information to recreate the scatter plots shown in the assays or experiments tabs on a saved cluster result.

### NOTE

In the figures below, the Auto and Manual Status columns refer to the status of the experiment (array), not to the genotype.

									Experiment Name	(a)4002716-68467	(a)4000639-64620	(a)4
									Status	Auto Pass	Auto Pass	Autc
									Sample Location	SMP2_0000601:A01	SMP2_0000601:A02	SMF
									Sample Name	NA12753-1447-2	NA12707-1358-1	NA1
									Sample Repeat Count	1	1	1
									Experiment Call Rate %	100	96.875	
Assay Index	Assay Id	External Id	Target Allele	Genome Map	Chrom Name	Chrom Position	Gene	Assay Call Rate %				
0	191049	rs9653	G/T	NCBI build 34	12	53075423	ZNF385	100		2	2	
1	191063	rs10789	A/C	NCBI build 34	12	119391862	DNCL1	97.826087		1	1	
2	191079	rs766719	G/T	NCBI build 34	12	16780580	-	100		2	2	
3	191094	rs3851650	A/C	NCBI build 34	12	115254812	-	100		1	2	
4	191102	rs4883251	G/T	NCBI build 34	12	9412026	-	100		2	2	
5	191134	rs7970250	A/C	NCBI build 34	12	43984966	TMEM16F	100		2	2	
6	191153	rs7979887	A/C	NCBI build 34	12	129649602	RIMB2_HUMAN	100		0	1	
7	191163	ssahaSNP.101015307	A/C	NCBI build 34	12	123365583	-	100		2	2	
8	191179	ssahaSNP.101033282	A/C	NCBI build 34	12	126897822	-	98.913043		1	1	
9	191212	ssahaSNP.101070950	A/C	NCBI build 34	12	9403900	-	95.652174		0	1	
10	191226	ssahaSNP.101087962	G/T	NCBI build 34	12	115252153	-	100		2	2	
11	191259	ssahaSNP.101113721	A/C	NCBI build 34	12	122199765	-	97.826087		2	2	
12	191310	ssahaSNP.101161132	A/C	NCBI build 34	12	75209608	-	100		1	0	

**Figure 6.4**  
Example of Genotypes Standard Data Output Format

	A	B	C	D	E	F	G	H	I
1	Assay Id	External Id	Target Allele	Experiment Name	Sample Location	Sample Name	Exp. Status	Genotype	Call Confidence
2	191049	rs9653	G/T	(a)4002716-68467	SMP2_0000601:A01	NA12753-1447-2	Auto Pass	2	0.969789135
3	191049	rs9653	G/T	(a)4000639-64620	SMP2_0000601:A02	NA12707-1358-1	Auto Pass	2	0.998832335
4	191049	rs9653	G/T	(a)4000639-64658	SMP2_0000601:A03	NA11839-1349-13	Auto Pass	2	0.999462632
5	191049	rs9653	G/T	(a)4000639-64642	SMP2_0000601:A04	NA10859-1347-2	Auto Pass	2	0.999684294

**Figure 6.5**  
Example of Genotypes Long Data Output Format

	A	B	C	D	E	F	G
1	Sample Name	Experiment Name	Gene	External Id	Assay Id	Allele 1	Allele 2
2	NA12753-1447-2	(a)4002716-68467	ZNF385	rs9653	191049	T	T
3	NA12707-1358-1	(a)4000639-64620	ZNF385	rs9653	191049	T	T
4	NA11839-1349-13	(a)4000639-64658	ZNF385	rs9653	191049	T	T
5	NA10859-1347-2	(a)4000639-64642	ZNF385	rs9653	191049	T	T
6	NA07034-1341-11	(a)4000639-64661	ZNF385	rs9653	191049	T	T

**Figure 6.6**  
Example of Genotypes Short Data Output Format

	A	B	C	D	E	F	G	H	I
1	Assay Id	External Id	Experiment Name	Sample Location	Sample Name	Exp. Status	Assay Status	Assay Allele	Is Assay Reverse
2	191049	rs9653	(a)4002716-68467	SMP2_0000601:A01	NA12753-1447-2	Auto Pass	Pass	A/C	TRUE
3	191049	rs9653	(a)4000639-64620	SMP2_0000601:A02	NA12707-1358-1	Auto Pass	Pass	A/C	TRUE
4	191049	rs9653	(a)4000639-64658	SMP2_0000601:A03	NA11839-1349-13	Auto Pass	Pass	A/C	TRUE
5	191049	rs9653	(a)4000639-64642	SMP2_0000601:A04	NA10859-1347-2	Auto Pass	Pass	A/C	TRUE

J	K	L	M	N	O	P
Measured	A Normalized Signal	C Normalized Signal	G Normalized Signal	T Normalized Signal	Contrast	Signal Sum
0	7747.2056	292.53174	35.734016	-21.031044	-0.863433	8039.737
0	8399.323	-59.862076	-43.79608	81.44976	-1.030214	8339.461
0	8300.163	-34.280968	48.506363	-0.014109294	-1.022568	8265.882
0	8744.991	24.794477	21.638216	162.45306	-0.989423	8769.785

**Figure 6.7**  
Example of Assay Allele Signals Output

## GENOTYPE CALL CODES

Table 6.1 contains the genotype call codes you should use to determine your genotypes.

**Table 6.1**  
Genotype Call Codes

Allele/Call Code	0	1	2	5
AC	AA	AC	CC	NO CALL
AG	AA	AG	GG	NO CALL
AT	AA	AT	TT	NO CALL
CG	CC	CG	GG	NO CALL
CT	CC	CT	TT	NO CALL
GT	GG	GT	TT	NO CALL
<b>General</b>	<b>0</b>	<b>1</b>	<b>2</b>	<b>5</b>
XY	XX	XY	YY	NO CALL



## About the Software

### SYSTEM SOFTWARE

The following software is supplied with the GCS 3000 TG System.

#### **GeneChip® Targeted Genotyping Analysis Software**

GTGS allows users to perform:

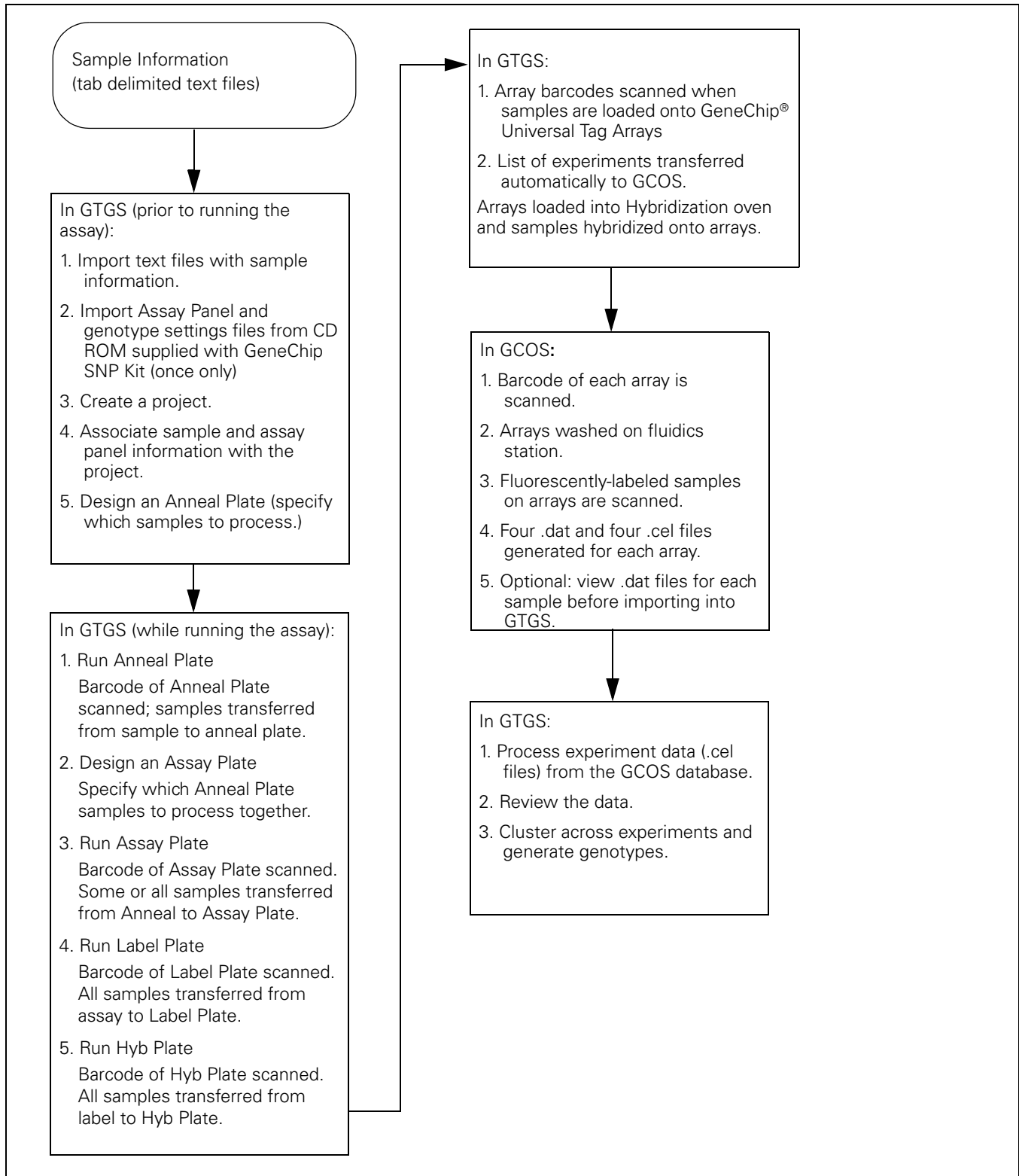
- User management
- Project design
- Sample tracking
- Automated experiment validation
- Clustering and genotyping

#### **GeneChip® Operating Software**

GCOS version 1.4:

- Controls the Scanner 3000 7G 4C and the GeneChip® Fluidics Station 450
- Generates and allows users to view .dat files (image files) for an experiment
- Generates .cel files (intensity files)

## DATA FLOW FROM SAMPLE TO GENOTYPE



**Figure 6.8**  
Data Flow from Sample to Genotype

## Data Analysis Frequently Asked Questions

**Q:** How automated is SNP scoring?

**A:** GTGS automatically assigns genotype calls which cannot be manually overridden within the software. However, genotype settings can be adjusted to make genotype assignment more or less stringent.

**Q:** Can our software distinguish between assays that passed, failed, or were borderline?

**A:** Yes.

**Q:** Can data from other vendor's software be imported into GTGS and reanalyzed?

**A:** No.

**Q:** Can GTGS test Mendelian inheritance (as a measure of genotyping accuracy)?

**A:** Yes.

**Q:** What is the largest data set that can be genotyped?

**A:** The software has been verified to genotype data sets as large as 5000 experiments of a 20K assay panel (100 million genotypes) in a single GTGS project.

**Q:** What happens as data accumulates in the database? Is it ever emptied?

**A:** No. This version of the GCS 3000 TG System does not support automatic archiving. You can delete data if desired. See [Backup Recommendations on page 45](#) for more information.

**Q:** Can genotypes be extracted at any time?

**A:** Yes. You do not need to wait until the end of a project. However, these genotypes should be treated as a snapshot only. Genotype data is better if clustering is done at the end of a project.

**Q:** How many samples are required for clustering?

**A:** We recommend clustering data from at least 80 unique samples. Clustering results are not as meaningful when fewer than 80 samples are used.

**Q:** Can GCS 3000 TG System data be consolidated with existing data?

**A:** GTGS exports only genotypes. You must decide how you will integrate your data.

**Q:** How is the database backed up?

**A:** A nightly backup is made to the other physical hard drive of the TG Post-Amp Workstation. You can edit the SQL backup procedure, or copy the backup elsewhere as needed.



Appendix **A**

**Forms**

Appendix *A*



## List of Forms

The forms included in this appendix are:

- *Confirmation of Site Preparation*

This form must be completed, signed and returned to Affymetrix before a date can be set for installation of your Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System (GCS 3000 TG System).

This form is located [on page 58](#).

- *Site Preparation Checklist*

This form is located [on page 59](#).

- *Confirmation of Equipment Installation and Laboratory Set Up*

This form must be completed and signed by your Affymetrix representative and by your Site Preparation Coordinator upon completion of system installation and verification. Each party retains a copy of this form.

This form is located [on page 61](#).

- *Workstation Installation and Integration Information and Questionnaire*

This form must be completed, signed and returned to Affymetrix before a date can be set for installation of your GCS 3000 TG System.

This form is located [on page 63](#).

## Confirmation of Site Preparation

Before equipment installation and laboratory set up for the Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System can be scheduled, you must have two areas designated as the Pre-Amp Lab and the Post-Amp Lab. These areas must be setup per the guidelines listed in the *Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System Site Preparation Guide*.

Site preparation must be confirmed by your organization or company's designated Site Preparation Coordinator. This individual must complete the *Site Preparation Checklist*, sign this form where indicated, and return the form and checklist to your local Affymetrix representative.

---

I verify that the site will be prepared prior to installation and training per the specifications listed in the *Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System Site Preparation Guide* and the attached *Site Preparation Checklist*.

Company/Organization Name:

\_\_\_\_\_

Confirmed By:

\_\_\_\_\_

(Print Name)

Confirmed By:

\_\_\_\_\_

(Authorized Signature)

Date: \_\_\_\_\_

Telephone Number: \_\_\_\_\_



## Site Preparation Checklist

Item	√ if ready	Date	Comments (Also use Notes on page 3)
Instruments received from Affymetrix.			
Thermal cyclers approved for use are available. See <a href="#">Thermal Cyclers on page 10</a> .			
You have ordered additional consumables including GeneChip® SNP Kits, GeneChip® Universal Tag Arrays, and both Taq polymerases (from Stratagene and Clontech).  All consumables have been properly stored.			
Two physically separated labs have been designated and setup per the specifications listed in the <i>Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System Site Preparation Guide</i> .			
An IT Systems Administrator has been selected and advised of the system installation requirements.			
Two Research Assistants have been selected and are available for training.			
An individual to do data analysis has been selected and is available for training.			
A System Installation Assistant and two Facilities individuals have been designated to help unpack and place some of the equipment.			
<b>In the Pre-Amp Lab</b>			
One –20°C freezer has been installed			
One 4°C refrigerator has been installed			
<b>Post-Amp Lab</b>			
One –20°C freezer has been installed			
One 4°C refrigerator has been installed			



## Confirmation of Equipment Installation and Laboratory Set Up

Before a training session can be scheduled, all of the equipment and the computer hardware and software required for the Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System must be installed and verified. The installation must be confirmed by your designated Site Preparation Coordinator and by the Affymetrix representative performing the installation.

To confirm that the lab areas have been properly setup and that a training session can now be scheduled, complete this form with the required signatures. Each party should retain a copy of this form.

List any comments or outstanding installation issues that must be resolved prior to training under [Notes on page 62](#).

---

We, the undersigned, verify that all of the equipment, and the computer hardware and software required for the Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System have been installed and are ready for operation.

Company/Organization Name:

\_\_\_\_\_

Site Preparation Coordinator:

\_\_\_\_\_

(printed)

Site Preparation Coordinator:

\_\_\_\_\_

(signature)

Date: \_\_\_\_\_

Telephone Number: \_\_\_\_\_

Affymetrix Representative: \_\_\_\_\_  
(printed)

Affymetrix Representative: \_\_\_\_\_  
(signature)

Date: \_\_\_\_\_



## Workstation Installation and Integration Information and Questionnaire

Please complete the questionnaire at the end of this document and return it to your Affymetrix representative.

### INSTALLATION INFORMATION FOR THE SYSTEM ADMINISTRATOR

System installation must be performed in the sequence specified by the Affymetrix representative. Do not change computer configurations unless requested by the Affymetrix representative (this includes changing machine names, patching Microsoft® Windows software, adding them to the domain, and configuring antivirus protection.)

The TG Post-Amp Lab and Instrument Control Workstation names should not be changed after installation. Both of these workstations act as servers, and the client computers must know the server names.

Prior to system installation, please have active network ports available in the intended locations of the three computers. Two standard power outlets are needed for each computer. Please have a power strip available.

The following computers will have to be added to the same Microsoft Windows NT domain as the intended users of the system prior to the workstation setup.

- 7G Instrument Control Workstation
- TG Pre-Amp Lab Workstation
- TG Post-Amp Lab Workstation

Affymetrix GeneChip® Targeted Genotyping Analysis Software (GTGS) is distributed by clicking the following URL: *http://<post-amp workstation name>/genol/index.html*.

If a proxy server is utilized by clients for HTTP connections on the local LAN, an appropriate entry must be made in the proxy server, DNS Server, and/or the “hosts” file on all client machines. This entry allows clients to connect to the TG Post-Amp Lab Workstation web server by machine name.

### ACCESS TO THE SYSTEM DATABASE AND APPLICATIONS

Be aware that users are capable of running the system software from any machine that can access the application web link, provided that a relatively recent version of Java Runtime exists on that machine. Running the software from a computer other than the TG Pre-Amp and TG Post-Amp Lab Workstations is NOT supported by Affymetrix, and the applications may not run properly.

Two security level options are available for allowing users to access system software and data

- Microsoft Windows NT authentication, specific NT users

When GTGS launches, the software assumes the user is the username logged into the current Microsoft Windows session. Users must select a username and enter a password when the computer starts up. In this configuration, application access is restricted in the following ways:

- You must create a Microsoft Windows NT user group called **GTGSusers**. Authorized users must be added or removed as necessary. Only **GTGSusers** and the database administrator have access to the TG Post-Amp Workstation database. **GTGS** will not launch unless run by an authorized Windows NT user group member.
- You can use Windows security features to prevent unauthorized users from accessing computers logged in as an authorized user. For example, a password-protected screen saver can turn on after a period of inactivity. You can also ask users to log out of the current Windows session, so that at next use **GTGS** records the name of the user who is actually using the software.
- DB authentication, honor system  
When **GTGS** launches, users select their name from a pull-down menu (no password). In this mode, certain database operations associate the currently-selected user to those operations for traceability purposes. Users themselves can create new users and inactivate old users as needed. This system assumes that users will log in as themselves. It does not prevent unauthorized users from accessing **GTGS** and database.

## **MICROSOFT WINDOWS AND FIREWALL**

**GTGS** and **GCOS** have been validated to work with Windows XP Service Pack 1 and Service Pack 2. To update the system to Windows XP Service Pack 2 (which includes Windows Firewall), follow the instructions that are included with **GTGS** and **GCOS**.

If you decide to set a Windows updating preference, it must be done **AFTER** **GTGS** and **GCOS** have been installed.

Automatic Java updates should remain disabled for the system workstations.

## **THIRD PARTY SOFTWARE**

The software on each workstation includes:

- Norton Antivirus™ (not activated)
- Microsoft Office® Professional (not activated)

If you decide to activate Norton Antivirus, be sure to follow the instructions provided with **GTGS**.

Automatic Java updates should remain disabled for all system workstations.

**The following questions must be answered before the Affymetrix GeneChip® Scanner 3000 Targeted Genotyping System is installed.**

Site Name:

City/ State:

Date:

**Contacts**

Name of contacts that will be involved with system installation.

Name of IT personnel:

Phone number:

Email address:

Name of Bioinformatics Specialist personnel:

Phone number:

Email address:

Name of Scientist or Primary End User personnel:

Phone number:

Email Address:

1. What is the Windows NT Domain name that this system will reside on?  
The name is: \_\_\_\_\_
2. What is the Windows NT Domain name that the users will reside on?  
The name is: \_\_\_\_\_
3. Is it required for the computer names to be updated to conform to corporate naming standards before they can be placed on the corporate domain?  
\_\_\_ Yes  
\_\_\_ No
4. Is a proxy server required for the workstations for HTTP connections on the local LAN?  
\_\_\_ Yes, and I will configure each client computer appropriately.  
Proxy Server info: \_\_\_\_\_ .  
DNS Server info: \_\_\_\_\_ .  
IP Address/setup info: \_\_\_\_\_ .  
\_\_\_ No proxy server is required for the system workstations
5. Do you want to restrict user access to GTGS and database?  
\_\_\_ Yes. I will create the Windows NT usergroup **GTGSusers**, and I will designate someone to maintain the list of users who can access the system software.

\_\_\_ No. Grant access to all users who can access the URL  
http://<post-amp workstation name>/geno/index.html

6. GCS 3000 TG System Workstations are preconfigured with a local user account called **Administrator** with administrative privileges, and with **NO** password. A user who logs in as this user on the TG Post-Amp Lab Workstation can access GTGS and database, if “DB\_authentication” mode is used. Do you want to change this account on the TG Pre-Amp and TG Post-Amp Lab Workstations **AFTER** system installation is complete?

\_\_\_ Yes, the password has been changed to \_\_\_\_\_

\_\_\_ No, this account doesn't need to be changed.

7. How will the Affymetrix representative be able to configure and test the installation while logged in as a domain user?

\_\_\_ I will arrange for a Windows NT domain user to be present to log in to all the machines whenever requested by the Affymetrix representative.

\_\_\_ I will grant the Affymetrix representative access to a domain account for the duration of the installation process.

Username \_\_\_\_\_

Password \_\_\_\_\_

## STAFF REQUIREMENT DURING THE WEEK OF INSTALLATION

1. IT personnel available to assist with client install (login privileges and resolution of network configuration issues)
2. Scanner systems not to be used during the installation
3. Scientists aware of installation
4. Bioinformatics available for contact or locating any staff members

I verify that the above information is correct and will be configured prior to the arrival of the Affymetrix representative who will install the software on the server and the necessary clients.

The necessary IT, Bioinformatics, and Scientists (end-users) will be available to work with Affymetrix staff during the week of installation.

NOTE: If Affymetrix incurs any delays, and is required to do more work while on-site due to the above not being complete prior to arrival, we will require a Purchase Order number to charge you for the additional on-site time and expenses.

\_\_\_\_\_  
Name

\_\_\_\_\_  
Date



Appendix **B**

**Vendor Contact Information**

Appendix **B**



## Vendor Contact Information

**NOTE** 

Customers outside the United States should check each vendor's website for additional contact information.

**Table B.1**  
Vendor Contact Information

Company	Address	Telephone
<b>Affymetrix</b> <a href="http://www.affymetrix.com">www.affymetrix.com</a>	3380 Central Expressway Santa Clara, California 95051	U.S. 1-888-362-2447 U.K. +44 (0)1628 552550 Japan +81-3-5730-8222
<b>Applied Biosystems</b> <a href="http://www.appliedbiosystems.com">www.appliedbiosystems.com</a>	850 Lincoln Centre Foster City, California 94404	1-800-327-3002 1-650-638-5800
<b>Axygen Scientific, Inc.</b> <a href="http://www.axygen.com">www.axygen.com</a>	33210 Central Ave. Union City, California 94587	1-800-4-AXYGEN 1-510-494-8900
<b>Distributors for Barcoded 96-Well PCR Thermal Cycler Plates and Barcode Labels (Anneal, Assay, Label, Hyb)</b> For distributors in countries not listed, refer to: <a href="http://www.axygen.com/Distributors">www.axygen.com/Distributors</a>		
	<b>U.S.A.</b> E & K Scientific, Inc. 3575 Thomas Road Santa Clara, California 95054 <a href="http://www.eandkscientific.com">www.eandkscientific.com</a>	Tel: 1-800-934-8114 or 1-408-378-2013 Fax: 1-408-378-2611
	<b>Australia</b> Radiometer Pacific P/L PO Box 47 Nunawading Vic 3131 <a href="http://oceania1.radiometer.com">oceania1.radiometer.com</a>	Tel: [61] 39-7063-655 Fax: [61] 39-7063-755
	<b>Canada</b> Ultident Scientific 4850 Chemin Bois Franc Suite 100 St. Laurent, Quebec H4R 2G7 <a href="http://www.ultident.com">www.ultident.com</a>	Tel: 514-335-3433 Fax: 514-335-0992
	<b>Germany</b> G.Kisker GbR Produkte f.d. Biotechnologie Postfach 1329 48543 Steinfurt <a href="http://www.kisker-biotech.com">www.kisker-biotech.com</a>	Tel: [49] 02551-864310 Fax: [49] 02551-864312
	<b>Japan</b> Funakoshi Co Ltd. 9-7. Hongo 2-Chome Bunkyo-Ku Tokyo 113-0033 <a href="http://www.funakoshi.co.jp">www.funakoshi.co.jp</a>	Tel: [81] 3-5259-5901 Fax: [91] 3-5259-1005

**Table B.1**  
Vendor Contact Information

Company	Address	Telephone
	<p><b>Singapore</b> Bio Laboratories 10 Ubi Crescent. Lobby A #06-02. Ubi Techpark 408564 <a href="http://www.biolab.com.sg">www.biolab.com.sg</a></p>	<p>Tel: [65] 6846-7577 Fax: [65] 6846-7477</p>
	<p><b>United Kingdom</b> Thistle Scientific Ltd. DFDS House Goldie Road Uddingston. Glasgow G71 6NZ</p>	<p>Tel: [44] 169-833-8844 Fax: [44] 169-833-8880</p>
<p><b>Bio-Rad Laboratories</b> <a href="http://www.biorad.com">www.biorad.com</a></p>	<p>2000 Alfred Nobel Dr. Hercules, California 94547</p>	<p>Life Science Research Group 1-800-424-6723 1-510-741-1000</p>
<p><b>BioSmith Biotech</b> <a href="http://www.biosmith.com">www.biosmith.com</a></p>	<p>3649 Conrad Ave. San Diego, California 92117</p>	<p>1-800-929-7894 1-858-270-8389</p>
<p><b>Clontech, a TAKARA BIO Company</b> <a href="http://www.clontech.com/clontech">www.clontech.com/clontech</a></p>	<p>1290 Terra Bella Avenue Mountain View, CA 94043 USA</p>	<p>1-800-662-2566 1-650-919-7300</p>
<p><b>Coriell Cell Repositories</b> <a href="http://locus.umdj.edu/ccr/">http://locus.umdj.edu/ccr/</a></p>	<p>403 Haddon Avenue Camden, NJ 08103</p>	<p>Tel: 1-800-752-3805 Fax: 1-856-757-9737</p>
<p><b>Eppendorf</b> U.S. only: <a href="http://www.eppendorfn.com">www.eppendorfn.com</a> Also available through distributors</p>	<p>One Cantiague Road P.O. Box 1019 Westbury, NY 11590-0207</p>	<p>1-800-645-3050</p>
<p><b>Gilson</b> <a href="http://www.pipetman.com">www.pipetman.com</a></p>	<p>3000 W. Beltline Hwy. P.O. Box 620027 Middleton, WI 53562-0027</p>	<p>1-800-GILSON1 or 1-608-836-1551</p>
<p><b>Invitrogen Life Technologies</b> <a href="http://www.invitrogen.com">www.invitrogen.com</a></p>	<p>1600 Faraday Ave. P.O. Box 6482 Carlsbad, California 92008</p>	<p>1-760-603-7200</p>
<p><b>Rainin Instrument, LLC</b> <a href="http://www.rainin.com">www.rainin.com</a></p>	<p>7500 Edgewater Dr. P.O. Box 2160 Oakland, California 94621</p>	<p>1-800-472-4646 1-510-564-1600</p>
<p><b>Stratagene</b> <a href="http://www.stratagene.com">www.stratagene.com</a></p>	<p>11011 North Torrey Pines Rd. La Jolla, California 92037</p>	<p>1-858-535-5400</p>
<p><b>USA Scientific, Inc</b> <a href="http://www.usascientific.com">www.usascientific.com</a></p>	<p>P.O. Box 3565 Ocala, Florida 34478</p>	<p>U.S.: 1-800-522-8477 International: 1-352-237-6288</p>
<p><b>VWR International</b> <a href="http://www.vwr.com">www.vwr.com</a></p>	<p>1310 Goshen Pkwy. West Chester, Pennsylvania 19380</p>	<p>1-800-932-5000</p>

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