ABI PRISM® GenBase™ 2.0 Database Application

User's Manual

Applied Biosystems

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Introducing GenBase

1

Chapter Overview

Introduction This chapter describes the ABI PRISM® GenBase™ 2.0 database application, the components of the ABI PRISM Genotyping Software System, and requirements for installing and starting the GenBase Database application.

In This Chapter This chapter contains the following topics:

About the GenBase Database Application

Introduction

GenBase is a database application that provides increased functionality for the ABI PRISM Genotyping Software System by providing relational database services to the Genotyper® software and the GenoPedigree™ software, as well as to users who want to search, store, or print reports from the database.

Advantages of GenBase

In conjunction with the other components of the ABI PRISM Genotyping Software System (the Genotyper software and the GenoPedigree software), the GenBase Database application allows you to:

- Import Genotyper data without reformatting.
- Store Genotyper information for retrieval and use by the GenoPedigree software.
- Store Genotyper information for retrieval and use by the Genotyper software.
- Directly export files in LINKAGE, CRI-MAP, or GENHUNTER formats.
- Quickly review Genotyper records and maintain an audit trail of reviewers and their comments.
- Import text files and integrate the data with Genotyper and GenoPedigree information.
- Exchange table data with the GenoPedigree software to create LINKAGE or GAS files.
- Construct and print detailed reports using information from the Genotyper software and the GenoPedigree software.
- Sort, search, and edit large bodies of information.

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What Is New in This Version

For those of you already familiar with the GenBase 1.0 Database application, GenBase 2.0 includes the following new features:

- Ability to directly export files in LINKAGE, CRI-MAP, and GENEHUNTER formats.
- Improved interface, with more pop-up menus in records tables to speed data entry.
- Ability to compare and review genotypes imported from the Genotyper software.
- Ability to exchange table data with the GenoPedigree software.

How GenBase Works

The GenBase Database application imports genotype records and categories from the Genotyper software, imports pedigree and disease-related data from the GenoPedigree software, and stores the data in tables specifically designed for this information. The GenoPedigree software then accesses those tables to construct graphic pedigree diagrams that can be exported to downstream applications such as LINKAGE and GAS.

The GenBase Database application also allows you to export convert table data into LINKAGE and CRI-MAP compatible files.

Note GenBase Database application was designed using 4D, and you can learn advanced database functions by reading the *4D User's Manual*. The 4D manual is not included.

Managing Genotypic and Phenotypic Data

The GenBase Database application allows you to store, manipulate, and review large batches of genotypic and phenotypic data. The primary sources for this data are two components of the ABI PRISM Genotyping Software System: Genotyper software and GenoPedigree software.

GenBase Services

The GenBase Database application also provides information to the Genotyper software and the GenoPedigree software. Both of these programs can extract table information from GenBase (see Figure 1-1).

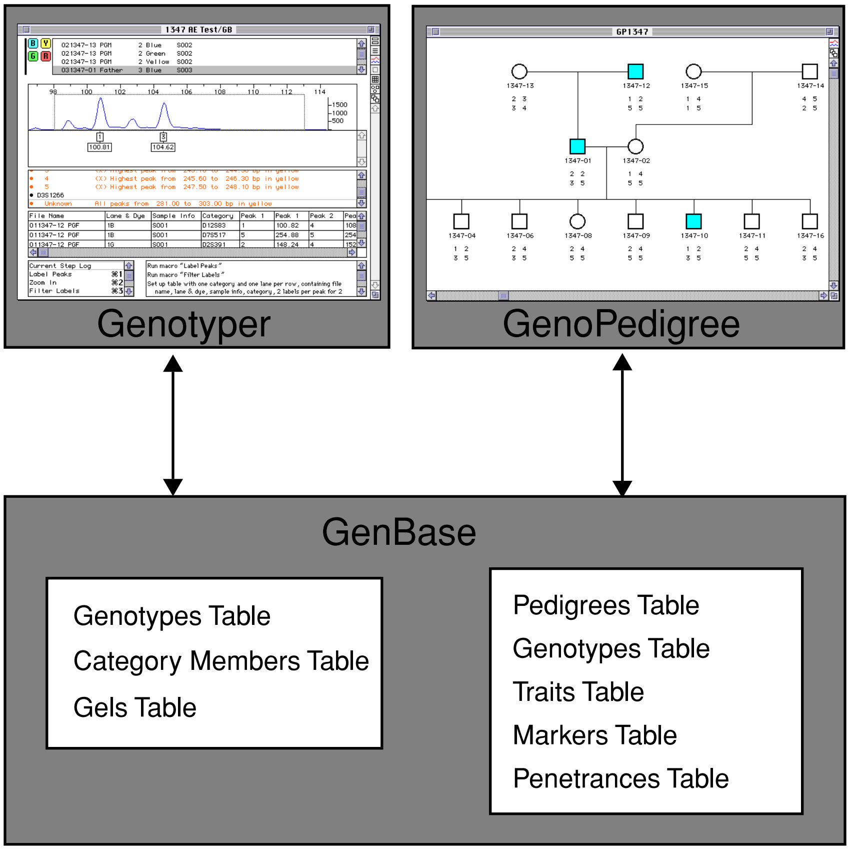
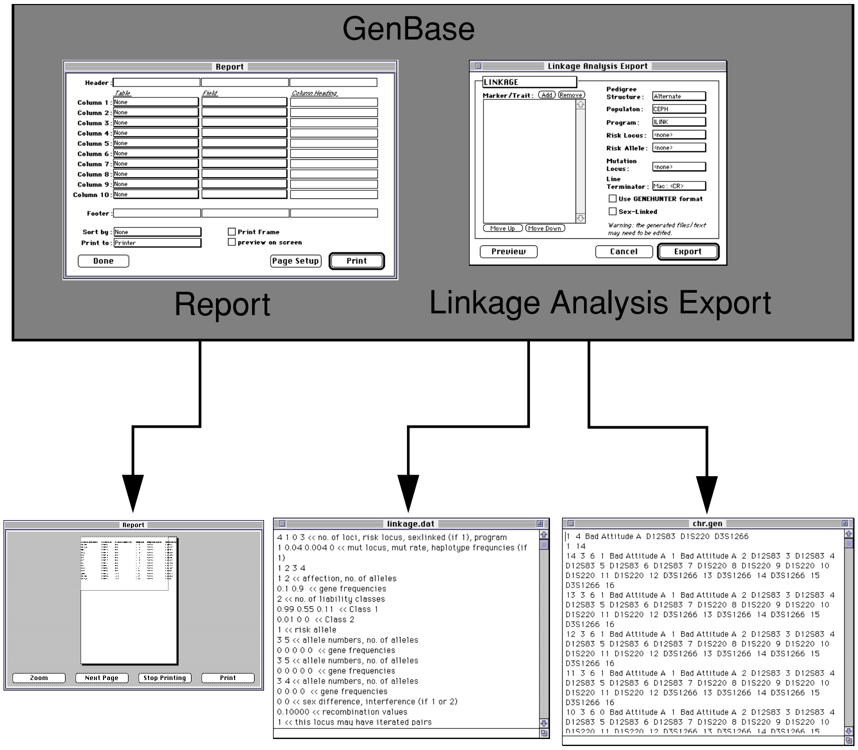


Figure 1-1 GenBase Database application interaction with the Genotyper software and the GenoPedigree software

Generating Reports and Exportable Files

You can use the GenBase Database application to produce custom reports, LINKAGE or CRI-MAP compatible files, or text files. GenBase gives you wide latitude in choosing the contents of the reports and exportable files.



Column 1 : None
Column 2 : None
Column 3 : None
Column 4 : None
Column 5 : None
Column 6 : None
Column 7 : None
Column 8 : None
Column 9 : None
Column 10 : None
Sort by : None
Print to : Printer
☐ Print Frame
☐ preview on screen

LINKAGE
Pedigree Structure : Alternate
Populaton : CEPH
Program : ILINK
Risk Locus : <none>
Risk Allele : <none>
Mutation Locus : <none>
Line Terminator : Mac : <CR>
☐ Use GENEHUNTER format
☐ Sex-Linked
Warning: the generated files/text may need to be edited.

```
4 1 0 3 << no. of loci, risk locus, sexlinked (if 1), program
1 0.04 0.004 0 << mut locus, mut rate, haplotype frequncies (if
1)
1 2 3 4
1 2 << affection, no. of alleles
0.1 0.9  << gene frequencies
2 << no. of liability classes
0.99 0.55 0.11  << Class 1
0.01 0 0  << Class 2
1 << risk allele
3 5 << allele numbers, no. of alleles
0 0 0 0 0  << gene frequencies
3 5 << allele numbers, no. of alleles
0 0 0 0 0  << gene frequencies
3 4 << allele numbers, no. of alleles
0 0 0 0  << gene frequencies
0 0 << sex difference, interference (if 1 or 2)
0.10000 << recombination values
1 << this locus may have iterated pairs
```

```
1  4  Bad Attitude A  D12S83  D1S220  D3S1266
1  14
14  3  6  1  Bad Attitude A  1  Bad Attitude A  2  D12S83  3  D12S83  4
D12S83  5  D12S83  6  D12S83  7  D1S220  8  D1S220  9  D1S220  10
D1S220  11  D1S220  12  D3S1266  13  D3S1266  14  D3S1266  15
D3S1266  16
13  3  6  1  Bad Attitude A  1  Bad Attitude A  2  D12S83  3  D12S83  4
D12S83  5  D12S83  6  D12S83  7  D1S220  8  D1S220  9  D1S220  10
D1S220  11  D1S220  12  D3S1266  13  D3S1266  14  D3S1266  15
D3S1266  16
12  3  6  1  Bad Attitude A  1  Bad Attitude A  2  D12S83  3  D12S83  4
D12S83  5  D12S83  6  D12S83  7  D1S220  8  D1S220  9  D1S220  10
D1S220  11  D1S220  12  D3S1266  13  D3S1266  14  D3S1266  15
D3S1266  16
11  3  6  1  Bad Attitude A  1  Bad Attitude A  2  D12S83  3  D12S83  4
D12S83  5  D12S83  6  D12S83  7  D1S220  8  D1S220  9  D1S220  10
D1S220  11  D1S220  12  D3S1266  13  D3S1266  14  D3S1266  15
D3S1266  16
10  3  6  0  Bad Attitude A  1  Bad Attitude A  2  D12S83  3  D12S83  4
D12S83  5  D12S83  6  D12S83  7  D1S220  8  D1S220  9  D1S220  10
D1S220  11  D1S220  12  D3S1266  13  D3S1266  14  D3S1266  15
```

Figure 1-2 Producing reports and exportable files in the GenBase Database application

Overview of the ABI PRISM Genotyping Software System

Definition

Genotyper, GeneScan®, GenBase, and GenoPedigree are four stand-alone software applications that when used together make up the ABI PRISM Genotyping Software System.

Relationship of System Components

Figure 1-3 illustrates how the ABI PRISM Genotyping Software System integrates the Genotyper software with the GenoPedigree software and linkage analysis programs through the GenBase Database application.

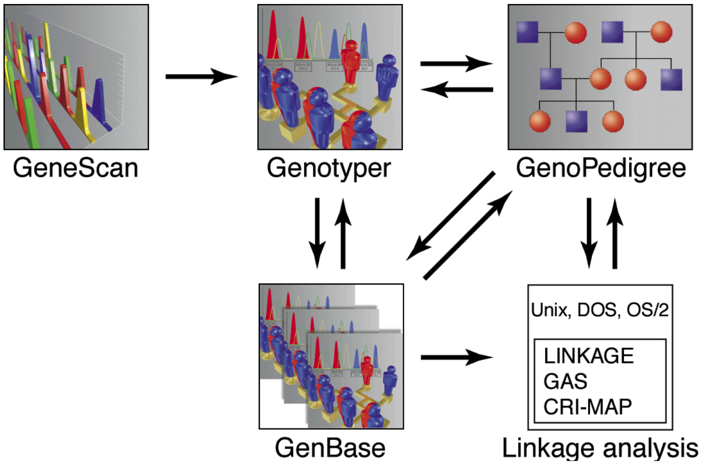


Figure 1-3 ABI PRISM Genotyping Software System

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Component Descriptions

Table 1-1 describes the components of the ABI PRISM Genotyping Software System.

Table 1-1 Genotyper system components

Component	**Description**
ABI PRISM GenBase Database application	Stores data for genotypes, pedigrees, markers, traits (diseases), and other relevant information. You can import data from or export data to the GenBase Database application from the Genotyper software and the GenoPedigree software.
ABI PRISM Genotyper software	Analyzes GeneScan files, labels fragment data with allele data, and creates tables specific to your genotyping studies.
ABI PRISM GeneScan Analysis software	Analyzes nucleic acid fragment data collected by an ABI PRISM instrument and sizes and quantifies detected fragments, storing the results in GeneScan files.
ABI PRISM GenoPedigree software	Creates pedigrees, integrates genotypic and phenotypic information of individuals in the pedigree, and prepares data for linkage analysis applications.

Related Publications

Table 1-2 describes publications that you can reference for detailed information about ABI PRISM Genotyping Software System component applications.

Table 1-2 ABI PRISM Genotyping System publications

Hard-copy Publications	
ABI PRISM GenBase User's Manual	Explains how to use a database application that stores data for genotypes, pedigrees, markers, traits (diseases), and other relevant information. You can import data from or export data to the GenBase Database application from the Genotyper software and the GenoPedigree software.
ABI PRISM Genotyper User's Manual	Explains how to use the features of the Genotyper software to analyze GeneScan files, and produce results data specific to your Genotyping application.

Table 1-2 ABI PRISM Genotyping System publications *(continued)*

Hard-copy Publications	
ABI PRISM GenoPedigree User's Manual	Explains how to use the GenoPedigree software to display, generate, analyze, import and export pedigree diagrams and data.
ABI PRISM GenoPedigree Applications Tutorials	Contains tutorial information for using the GenoPedigree software and the Genotyper System components to perform typical phenotyping applications.
ABI PRISM Genotyper Applications Tutorials	Contains tutorial information for using the Genotyper software and Genotyper System components to perform typical genotyping applications.
ABI PRISM GeneScan User's Manual	Explains how to use GeneScan Analysis software to size and quantify nucleic acid fragments detected on an ABI DNA sequencer.
CD-ROM Publications	
ABI PRISM Genotyper User's Manual	Allows you to use online navigation tools to search for information contained in the Genotyper User's Manual, and access user documentation for other Genotyper System components.
ABI PRISM GenoPedigree User's Manual	Allows you to use online navigation tools to search for information contained in the GenoPedigree User's Manual, and access user documentation for other Genotyper System components.
ABI PRISM GenBase User's Manual	Allows you to use online navigation tools to search for information contained in the GenBase User's Manual, and access user documentation for other Genotyper System components.

Knowledge Assumptions

The GenBase documentation you are using assumes that you have running copies of the Genotyper software and the GenoPedigree software with documentation, and that you are familiar with using these programs. The GenBase documentation does not assume that you are a database expert, or that you want to become one.

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Web Site Information

You can find helpful information about the ABI PRISM Genotyping Software System components by contacting the Applied Biosystems Web site at:

www.appliedbiosystems.com/techsupport

Installing the GenBase Database Application

Register Your Copy

When you register your copy of the GenBase Database application, you become eligible for telephone and field technical support from Applied Biosystems for 100 days from the date of shipment. To register, fill out the registration card included in this package and return it to Applied Biosystems.

For Applied Biosystems technical support telephone and address information, see “Technical Support” on page 1-15.

Registering also allows you to purchase upgrades for the software at a lower price than it would cost you to purchase a new, upgraded package.

IMPORTANT These privileges are available only if you have returned your registration card.

System Requirements

Table 1-3 describes the computer system required to run the GenBase Database application. These are the minimum requirements. In general, the more memory, screen size and processing power, the better.

Table 1-3 Required computer system

System Component	Requirements
CPU	A PowerPC Mac OS computer is recommended. Other models are not recommended. You will benefit from using the fastest computer available.
Operating System	Mac OS version 7.5.5 or later is recommended. Macintosh System 7.1 or later is required.
Monitor	A seventeen-inch monitor or larger is recommended, although monitor size of 640 x 480 pixels can be used. You will benefit from having a larger monitor. Color monitors are useful but not required.

Table 1-3 Required computer system *(continued)*

System Component	Requirements
Disk Drive	Because the GenBase data file is a single, large disk file, performance is dependent on having sufficient unfragmented space on your hard drive. Performance will suffer if the disk is almost full. The GenBase Database application requires a minimum of 5 MB free disk space to launch. Having a large amount of free disk space (for example, 100 MB) will help insure good performance.
Memory	The suggested memory allocation for the GenBase Database application is 6 megabytes of RAM (without virtual memory on, this increases to about 7.7 MB). Users will benefit from allocating more than 6 MB to the GenBase Database application. Up to a point, the program will run faster with more memory.

Installing GenBase

The GenBase Database application comes with the following diskettes.

- ♦ Program Diskette(s)
- ♦ Example Database Diskette(s)

IMPORTANT Do not run the GenBase Database application from the diskettes.

To install the GenBase Database application for the first time:

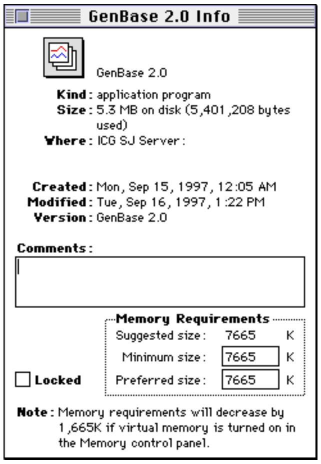
Step	Action
1	Disable virus protection software.
2	Insert the first Program diskette.
3	Double-click the GenBase installer icon.
4	Follow the on-screen directions.

continued on next page

Setting Memory Allocation

The size of your database will vary based on the number of records it contains. If you want to have many database tables open at the same time, you may need to allocate more memory to the GenBase Database application.

To change the memory allocation for the GenBase Database application:

Step	Action
1	Quit the GenBase Database application.
2	Click the GenBase application icon once to select it. 
3	Choose the Get Info command from the Finder's File menu. The Get Info box appears.  GenBase 2.0 **Kind :** application program **Size :** 5.3 MB on disk (5,401,208 bytes used) **Where :** ICG SJ Server : **Created :** Mon, Sep 15, 1997, 12:05 AM **Modified :** Tue, Sep 16, 1997, 1:22 PM **Version :** GenBase 2.0 **Comments :** **Memory Requirements** Suggested size : 7665 K Minimum size : 7665 K Preferred size : 7665 K ☐ **Locked** **Note :** Memory requirements will decrease by 1,665K if virtual memory is turned on in the Memory control panel. **Note** Memory allocation is initially set for 7,665 K. Virtual memory will reduce the minimum size by 1,665K.
4	Enter a new value for the Preferred size. **Note** In some versions of the Macintosh System, the Preferred size value is referred to as the Current size.
5	Click the close window box.
6	Restart the GenBase Database application.

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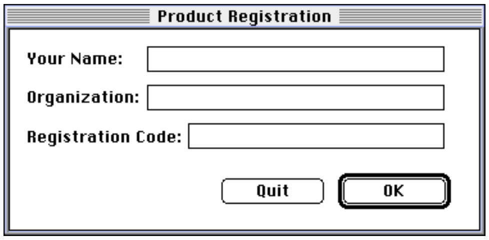
Starting GenBase

Once you have installed the GenBase Database application, you can start using the database included with the program. This database contains example data. If you want to use a database for your own application, you must create a new database.

To start the GenBase Database application:

Step	Action
1	Double-click the GenBase icon in the finder.
2	A dialog box appears and asks you to identify a database file. Select the Sample Database included with your copy of the GenBase Database application, and Click Open. To Create your own, blank database, click New. **Note** If, after the first time you open the GenBase Database application, you want to select a database other than the default, hold down the Option key when you start GenBase and select another database or a new, blank data file from the dialog that appears.
3	The first time you start the GenBase Database application, the Product Registration dialog box appears. Enter your name, organization, and registration code. 
4	Click OK. You are now ready to use the GenBase Database application.

continued on next page

Installing Updates

The upgrade kit will include instructions for installing the new version of the GenBase Database application.

IMPORTANT Do not throw out your data file. Always make a backup copy of your data file before installing an update.

Starting Up

We suggest that you first start the database with the example data file. Look at the various tables, and read through the manual, freely trying out the described features. Then, when you are comfortable with the program, start the GenBase Database application again, holding down the Option key until the file dialog appears. Then, you can begin creating a database of your own data. See Chapter 3, “Getting Started.”

IMPORTANT Be sure to read “Backing Up Your Data” on page 4-25 before putting any of your own data in the database.

Compatibility with Previous Versions

GenBase 2.0 is fully compatible with database files created with GenBase 1.0, and is able to use import and use data from all versions of the Genotyper software and the GenoPedigree software.

IMPORTANT Once you open a GenBase database file with version 2.0 of the GenBase Database application, that file becomes incompatible with GenBase 1.0.

Technical Support

Contacting Technical Support

You can contact Applied Biosystems for technical support by telephone or fax, by e-mail, or through the Internet. You can order Applied Biosystems user documents, MSDSs, certificates of analysis, and other related documents 24 hours a day. In addition, you can download documents in PDF format from the Applied Biosystems Web site (please see the section “To Obtain Documents on Demand” following the telephone information below).

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Protein Sequencing, Peptide and DNA Synthesis	corelab@appliedbiosystems.com
Biochromatography, PerSeptive DNA, PNA and Peptide Synthesis systems, CytoFluor®, FMAT™, Voyager™, and Mariner™ Mass Spectrometers	tsupport@appliedbiosystems.com
Applied Biosystems/MDS Sciex	api3-support@sciex.com
Chemiluminescence (Tropix)	tropix@appliedbiosystems.com

Hours for Telephone Technical Support

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Product	Hours
Chemiluminescence	8:30 a.m. to 5:30 p.m. Eastern Time
Framingham support	8:00 a.m. to 6:00 p.m. Eastern Time
All Other Products	5:30 a.m. to 5:00 p.m. Pacific Time

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Chemiluminescence (Tropix)	**1-800-542-2369** (U.S. only), or **1-781-271-0045**	**1-781-275-8581**
Applied Biosystems/MDS Sciex	**1-800-952-4716**	**1-650-638-6223**

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Portugal (Lisboa)	**351 (0)22 605 33 14**	**351 (0)22 605 33 15**
Russia (Moskva)	**7 095 935 8888**	**7 095 564 8787**
South East Europe (Zagreb, Croatia)	**385 1 34 91 927**	**385 1 34 91 840**
Spain (Tres Cantos)	**34 (0)91 806 1210**	**34 (0)91 806 1206**
Sweden (Stockholm)	**46 (0)8 619 4400**	**46 (0)8 619 4401**
Switzerland (Rotkreuz)	**41 (0)41 799 7777**	**41 (0)41 790 0676**

Region	Telephone Dial...	Fax Dial...
The Netherlands (Nieuwerkerk a/d IJssel)	**31 (0)180 331400**	**31 (0)180 331409**
United Kingdom (Warrington, Cheshire)	**44 (0)1925 825650**	**44 (0)1925 282502**
All other countries not listed (Warrington, UK)	**44 (0)1925 282481**	**44 (0)1925 282509**
Japan		
Japan (Hacchobori, Chuo-Ku, Tokyo)	**81 3 5566 6230**	**81 3 5566 6507**
Latin America		
Del.A. Obregon, Mexico	**305-670-4350**	**305-670-4349**

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Data Model

2

Chapter Overview

Introduction This chapter provides an overview of relational databases and how they work. It also includes a breakdown of the tables in the GenBase Database application, and each table’s unique and shared fields.

In This Chapter This chapter contains the following topics:

†11

What Is a Relational Database?

Introduction

A relational database is a tool for organizing large amount of related information is such a way that the information can be quickly searched, sorted, and printed. Relational databases differ from flat file databases (spreadsheets) in that data is organized using tables.

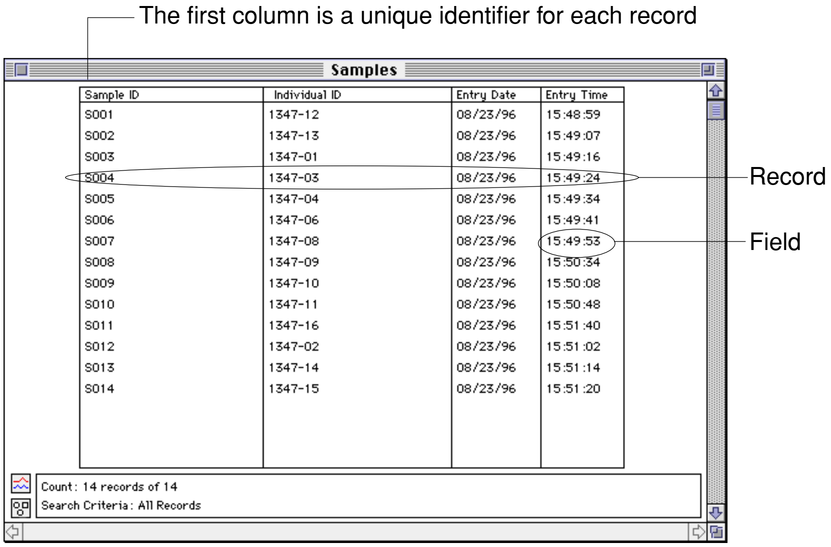
Why Use Databases?

Databases provide the following benefits to people who need centralized control of current information:

- Redundancy controlled and reduced
- Inconsistent data eliminated
- Data easily shared
- Standards enforced
- Security restrictions applied
- Integrity maintained
- Conflicting requirements balanced

Database Structure

It is easy to think of a relational database as a collection of spreadsheets having related rows. In database terminology, these spreadsheets are referred to as tables. Like spreadsheets, tables contain rows and columns. Rows in database tables are referred to as records, and each record contains one or more fields. (see Figure 2-1).


The first column is a unique identifier for each record

Record

Field

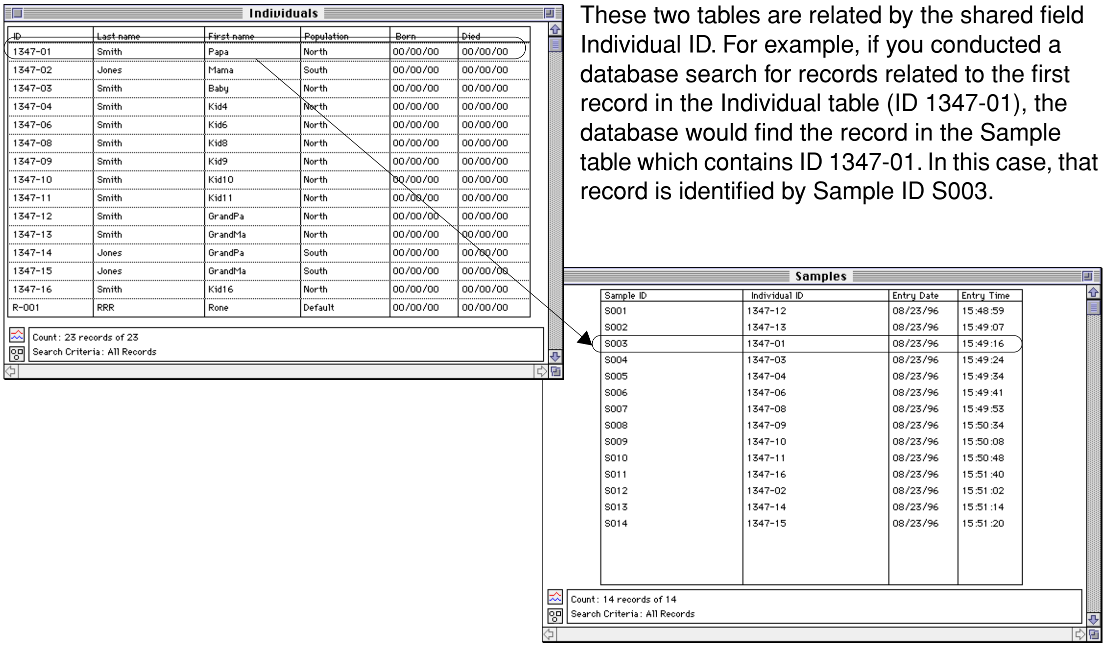
Sample ID	Individual ID	Entry Date	Entry Time
S001	1347-12	08/23/96	15:48:59
S002	1347-13	08/23/96	15:49:07
S003	1347-01	08/23/96	15:49:16
S004	1347-03	08/23/96	15:49:24
S005	1347-04	08/23/96	15:49:34
S006	1347-06	08/23/96	15:49:41
S007	1347-08	08/23/96	15:49:53
S008	1347-09	08/23/96	15:50:34
S009	1347-10	08/23/96	15:50:08
S010	1347-11	08/23/96	15:50:48
S011	1347-16	08/23/96	15:51:40
S012	1347-02	08/23/96	15:51:02
S013	1347-14	08/23/96	15:51:14
S014	1347-15	08/23/96	15:51:20

Count: 14 records of 14
Search Criteria: All Records

Figure 2-1 Example table

Fields can be either unique to that particular table or shared between multiple tables.

Records in each table are related to records in other tables by shared fields. For example, if one table in a database contains individual ID numbers, and another table contains sample records which contain the ID of the individual who contributed the sample, the two tables are related (see Figure 2-2).



ID	Last name	First name	Population	Born	Died
1347-01	Smith	Papa	North	00/00/00	00/00/00
1347-02	Jones	Mama	South	00/00/00	00/00/00
1347-03	Smith	Baby	North	00/00/00	00/00/00
1347-04	Smith	Kid4	North	00/00/00	00/00/00
1347-06	Smith	Kid6	North	00/00/00	00/00/00
1347-08	Smith	Kid8	North	00/00/00	00/00/00
1347-09	Smith	Kid9	North	00/00/00	00/00/00
1347-10	Smith	Kid10	North	00/00/00	00/00/00
1347-11	Smith	Kid11	North	00/00/00	00/00/00
1347-12	Smith	GrandPa	North	00/00/00	00/00/00
1347-13	Smith	GrandMa	North	00/00/00	00/00/00
1347-14	Jones	GrandPa	South	00/00/00	00/00/00
1347-15	Jones	GrandMa	South	00/00/00	00/00/00
1347-16	Smith	Kid16	North	00/00/00	00/00/00
R-001	RRR	Rone	Default	00/00/00	00/00/00

```
Count : 23 records of 23
Search Criteria : All Records
```

Sample ID	Individual ID	Entry Date	Entry Time
S001	1347-12	08/23/96	15:48:59
S002	1347-13	08/23/96	15:49:07
S003	1347-01	08/23/96	15:49:16
S004	1347-03	08/23/96	15:49:24
S005	1347-04	08/23/96	15:49:34
S006	1347-06	08/23/96	15:49:41
S007	1347-08	08/23/96	15:49:53
S008	1347-09	08/23/96	15:50:34
S009	1347-10	08/23/96	15:50:08
S010	1347-11	08/23/96	15:50:48
S011	1347-16	08/23/96	15:51:40
S012	1347-02	08/23/96	15:51:02
S013	1347-14	08/23/96	15:51:14
S014	1347-15	08/23/96	15:51:20

```
Count : 14 records of 14
Search Criteria : All Records
```

These two tables are related by the shared field Individual ID. For example, if you conducted a database search for records related to the first record in the Individual table (ID 1347-01), the database would find the record in the Sample table which contains ID 1347-01. In this case, that record is identified by Sample ID S003.

Figure 2-2 Related tables

It is this relationship between tables that distinguishes a relational database from a collection of spreadsheets and what makes a relational database so useful.

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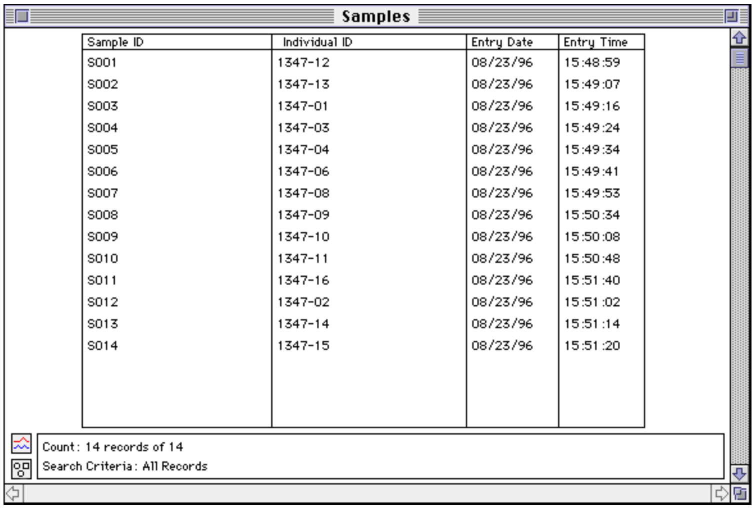
Example Database

Let's say, for example, that you have a list of samples taken from individuals with the date and time of each sample (see Figure 2-3).



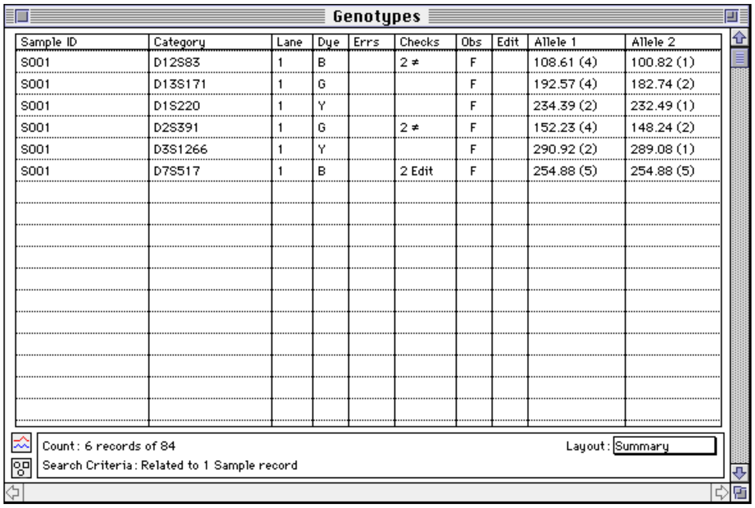
Sample ID	Individual ID	Entry Date	Entry Time
S001	1347-12	08/23/96	15:48:59
S002	1347-13	08/23/96	15:49:07
S003	1347-01	08/23/96	15:49:16
S004	1347-03	08/23/96	15:49:24
S005	1347-04	08/23/96	15:49:34
S006	1347-06	08/23/96	15:49:41
S007	1347-08	08/23/96	15:49:53
S008	1347-09	08/23/96	15:50:34
S009	1347-10	08/23/96	15:50:08
S010	1347-11	08/23/96	15:50:48
S011	1347-16	08/23/96	15:51:40
S012	1347-02	08/23/96	15:51:02
S013	1347-14	08/23/96	15:51:14
S014	1347-15	08/23/96	15:51:20

Count: 14 records of 14
Search Criteria: All Records

Figure 2-3 Database table (Samples)

This information cannot be kept in a simple spreadsheet, but if you want to quickly find all the genotype records for a particular Sample ID, you need to relate samples to their genotype records. Then you can create a table that stores a list of Genotype records.

Figure 2-4 shows the genotype records for Sample ID S001.



Sample ID	Category	Lane	Dye	Errs	Checks	Obs	Edit	Allele 1	Allele 2
S001	D12S83	1	B		2 ≠	F		108.61 (4)	100.82 (1)
S001	D13S171	1	G			F		192.57 (4)	182.74 (2)
S001	D1S220	1	Y			F		234.39 (2)	232.49 (1)
S001	D2S391	1	G		2 ≠	F		152.23 (4)	148.24 (2)
S001	D3S1266	1	Y			F		290.92 (2)	289.08 (1)
S001	D7S517	1	B		2 Edit	F		254.88 (5)	254.88 (5)

Count : 6 records of 84

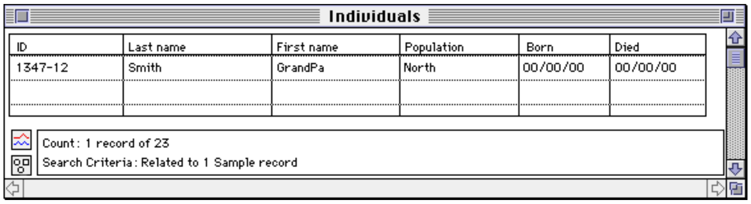
Layout : Summary

Search Criteria : Related to 1 Sample record

Figure 2-4 Genotypes of Sample ID S001

In both of these tables (Figure 2-3 and Figure 2-4), each row begins with the unique identifier Sample ID. In both tables, there is only one row for each sample.

Finally, for this example, we need a table that stores information about the individuals who contributed these samples (Figure 2-5). The unique identifier is Individual ID. Tables are typically named after their unique identifier.



ID	Last name	First name	Population	Born	Died
1347-12	Smith	GrandPa	North	00/00/00	00/00/00

Count : 1 record of 23

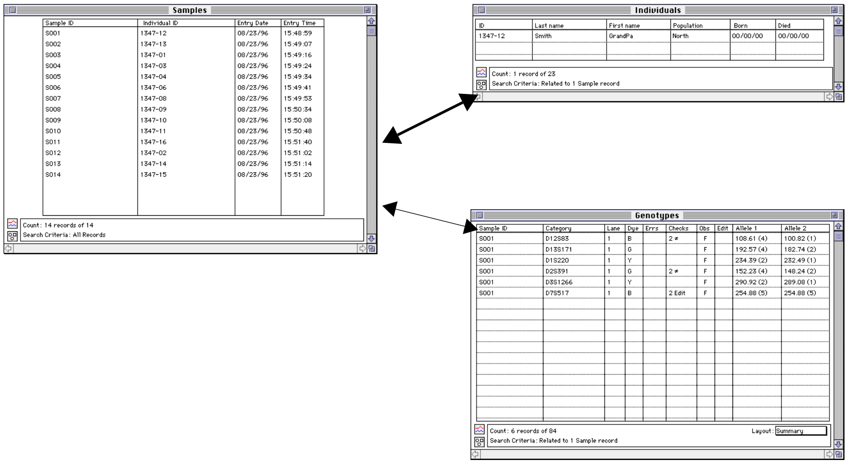
Search Criteria : Related to 1 Sample record

Figure 2-5 Individuals who contributed sample S001

We now have a small relational database. Records in the Individual table (Figure 2-5) are related to samples in the Samples table, which are related to the Genotypes table.

Example Data Model

A data model is a picture of your database tables that shows how they relate to one another (see Figure 2-6).



Sample ID	Individual ID	Entry Date	Entry Time
S001	1347-12	08/23/96	15:48:59
S002	1347-13	08/23/96	15:49:07
S003	1347-01	08/23/96	15:49:16
S004	1347-03	08/23/96	15:49:24
S005	1347-04	08/23/96	15:49:34
S006	1347-06	08/23/96	15:49:41
S007	1347-08	08/23/96	15:49:53
S008	1347-09	08/23/96	15:50:34
S009	1347-10	08/23/96	15:50:08
S010	1347-11	08/23/96	15:50:48
S011	1347-16	08/23/96	15:51:40
S012	1347-02	08/23/96	15:51:02
S013	1347-14	08/23/96	15:51:14
S014	1347-15	08/23/96	15:51:20

Count: 14 records of 14
Search Criteria: All Records

ID	Last name	First name	Population	Born	Died
1347-12	Smith	GrandPa	North	00/00/00	00/00/00

Count: 1 record of 23
Search Criteria: Related to 1 Sample record

Sample ID	Category	Lane	Dye	Errs	Checks	Obs	Edit	Allele 1	Allele 2
S001	D12S83	1	B		2 ≠	F		108.61 (4)	100.82 (1)
S001	D13S171	1	G			F		192.57 (4)	182.74 (2)
S001	D1S220	1	Y			F		234.39 (2)	232.49 (1)
S001	D2S391	1	G		2 ≠	F		152.23 (4)	148.24 (2)
S001	D3S1266	1	Y			F		290.92 (2)	289.08 (1)
S001	D7S517	1	B		2 Edit	F		254.88 (5)	254.88 (5)

Count: 6 records of 84
Layout: Summary
Search Criteria: Related to 1 Sample record

Figure 2-6 Data model example

Note that the Individual table is directly related to the Sample table, but not the Genotype table. For example, if you executed a database search for records related to Individual ID 1347-12, the search would find the first record in the Sample table (Sample ID S001) but no records from the Genotype table. However, if you searched for records related to Sample ID S001, the search would find the record 1347-12 in the individual table, as well as the six records in the Genotype table displayed in the figure above.

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Building a Database

Information can typically be entered into a database in one of two ways. The first is by manually paging through tables and typing in (or selecting choices from pop-up menus or checkboxes) the information you want in each individual field.

The second way is by importing data from other database programs, front-end programs such as the Genotyper software or the GenoPedigree software, or from text files. Obviously, importing data is much quicker, since tables are filled in automatically. However, it is important that the data be correctly formatted for the relevant table. For example, if a table has three fields per record, and the data you are importing has four fields per record, the imported values will end up in the wrong fields. Text files are particularly likely to be in an incorrect format, while Genotyper and GenoPedigree are designed to send correctly formatted data to the GenBase Database application.

Using the Database

The point of building a database is to be able to logically organize large volumes of data, and then use (manipulate) that data. Typically, database programs provide the following data manipulation tools:

- *Queries*–Find individual or groups of related fields or records based on multiple criteria
- *Sorts*–Sort the entire database or a subset of the database (for example, sorting a particular table) based on multiple criteria
- *Report Generation Functions*–Print or create exportable files from tables or collections of records. These reports may be based the results of a Query or Sort function, and can be preformatted by the application, or based on a custom report format.

Backing Up

Frequent backups are an important part of using a database. They assure you a way to recover your work quickly. Ideally, your backups should be kept on a volume that is separate from your work disk.

Saving Your Work

There are no save commands in most database programs. When you execute an action, the results are immediate and irreversible. This is another good reason to keep a current (read daily) backup of your work.

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Database Size

There are, generally speaking, no limits on the size of your database other than your available hard disk space. However, the size of your database will effect the speed and responsiveness of your database program.

If you have a large database, you must also be careful about executing broad searches. If you aren’t, you could spend several minutes waiting for the database program to search and sort a large number of matching records.

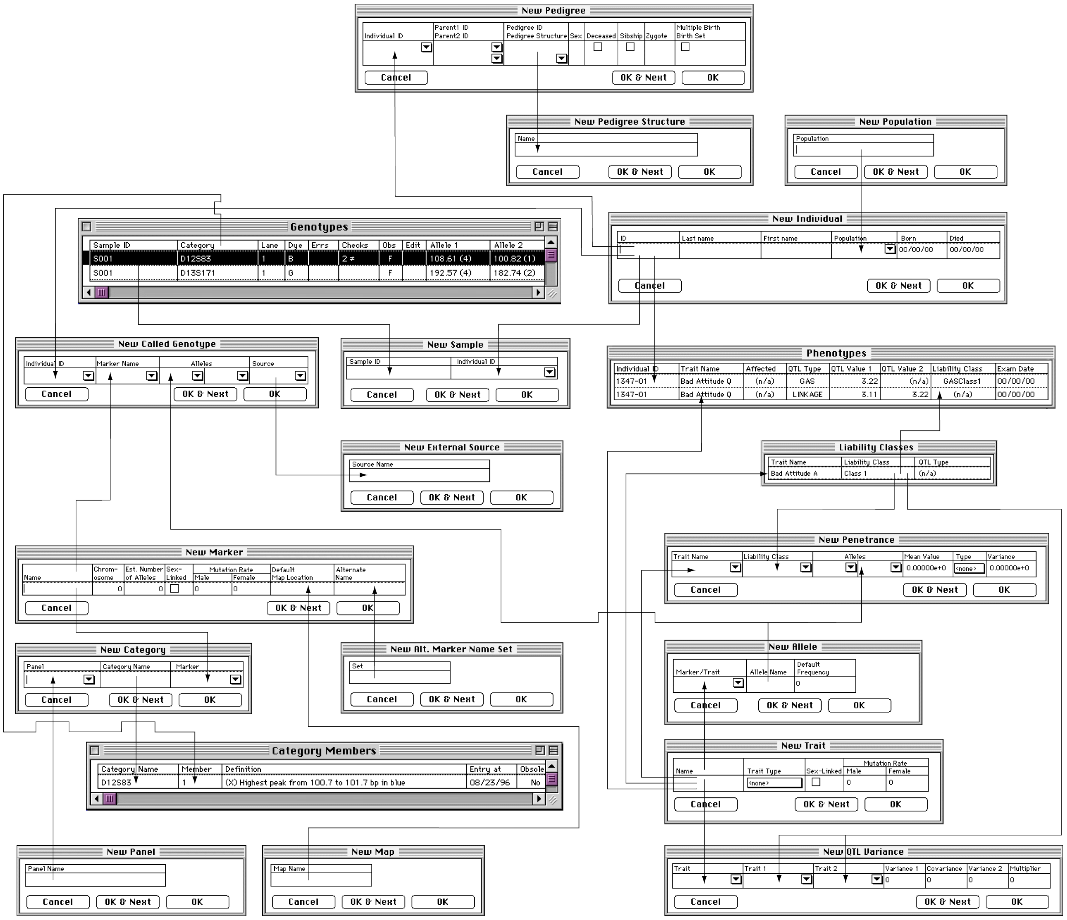
The GenBase Database Application

Introduction

The GenBase Database application is designed for data from the ABI PRISM Genotyping Software System. GenBase can store Genotyper records, as well as data from the GenoPedigree software. You can also import tab delimited text records into GenBase tables, however, the responsibility of verifying the format of that data is yours.

GenBase Organization

Like most other databases, the GenBase Database application is organized around tables (for a complete description of GenBase tables, see “About GenBase Tables” on page 2-15). Most of these tables are related through shared fields. Figure 2-7 is a representation of the GenBase database. The tables are represented by their Add Record dialog boxes. Arrows illustrate which fields are shared between tables.



Genotypes

Sample ID	Category	Lane	Dye	Errs	Checks	Obs	Edit	Allele 1	Allele 2
S001	D12S83	1	B		2 ≠	F		108.61 (4)	100.82 (1)
S001	D13S171	1	G			F		192.57 (4)	182.74 (2)

Phenotypes

Individual ID	Trait Name	Affected	QTL Type	QTL Value 1	QTL Value 2	Liability Class	Exam Date
1347-01	Bad Attitude Q	(n/a)	GAS	3.22	(n/a)	GASClass1	00/00/00
1347-01	Bad Attitude Q	(n/a)	LINKAGE	3.11	3.22	(n/a)	00/00/00

Liability Classes

Trait Name	Liability Class	QTL Type
Bad Attitude A	Class 1	(n/a)

Category Members

Category Name	Member	Definition	Entry at	Obsole
D12S83	1	(X) Highest peak from 100.7 to 101.7 bp in blue	08/23/96	No

Figure 2-7 GenBase Database application organization

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GenBase Tools

The GenBase Database application provides the following tools for manipulating database information:

- *Find*–The Find command allows you to search tables for individual records. You can then search the selected records based on new criteria to further narrow your search.
- *Find Genotypes*–A find command tailored specifically to the Genotypes table. The command allows you to search for specific review comments, specific reviews, or other genotype criteria.
- *Find Called Genotypes*–A find command tailored specifically for the Called Genotype table. The command allows you to search for records using criteria unique to the Called Genotypes table.
- *Sort*–The Sort command allows you to sort records in an individual table.
- *Find Related Records*–Allows you to find records related to the selected record across the entire database.
- *Reports*–The Report command allows you to build multiple column reports from all open tables and send them to the printer or create tab delimited text files.

Building GenBase Databases

Building a database, in most cases, consists of the following tasks:

- *Setting Up Tables*–Filling out database tables by either manually entering information, or by importing text files.
- *Importing Records*–Using ABI PRISM Genotyping applications to move Genotyper or GenoPedigree records into the database.

It isn't necessary to follow this order. You can import records without setting up tables, but generally it is easier to set up tables first.

Setting Up Tables

You can set up GenBase tables–filling empty tables with information–in one of the following ways:

- *Manual Entry*–This involves going through each table and manually filling in fields for each record.
- *Importing Tab Delimited Text Files*–These files can be imported directly into GenBase tables. However, because this data wasn't generated by a Genotyping application, there is no guarantee that it will be properly formatted for GenBase tables.

Importing Records

There are three basic types of records that can be imported into the GenBase Database application.

- *Importing Genotyper Data*–Genotyper data is already formatted for use in the GenBase Database application, and can be sent into the Genotype table from the Genotyper software.
- *Importing Pedigrees and Loci Data*–GenoPedigree data is correctly formatted for GenBase tables, and can be modified and sent back to the GenoPedigree software for export to downstream applications.
- *External Source Data*–You can also import genotype data from sources other than the Genotyper software directly into the Called Genotype tables. Because this information doesn't come from the ABI PRISM Genotyper software, verifying that data is in the correct format is up to you.

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Using GenBase

The GenBase Database application is an extremely flexible application, and there are many different ways to set up and use the database. However, in general, you will follow the path set out in Figure 2-8.

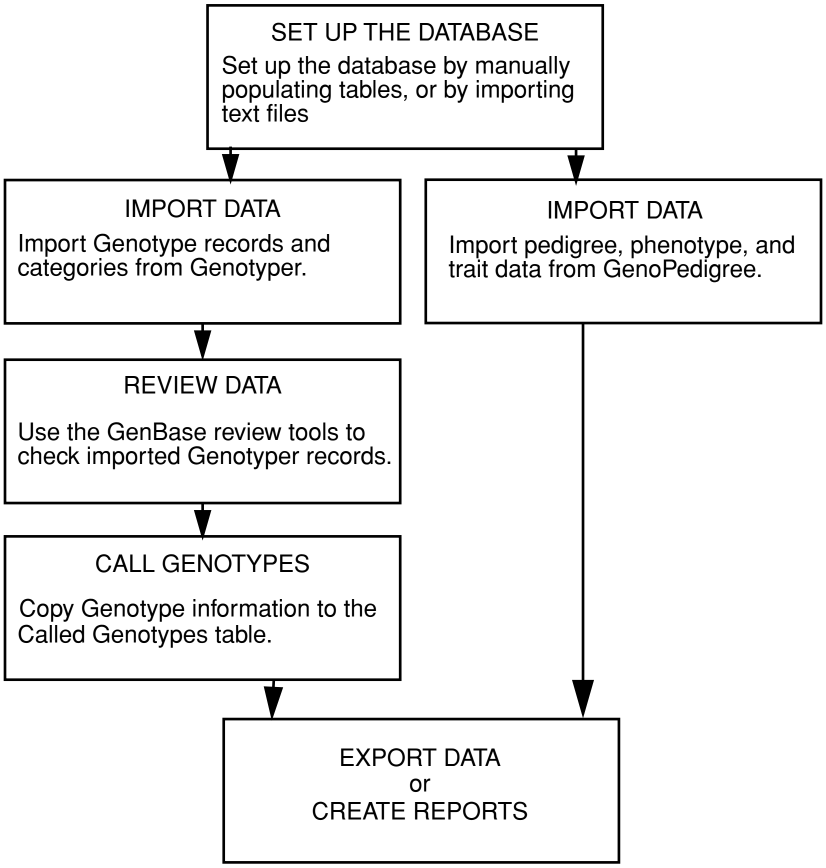


Figure 2-8 Using the GenBase Database application

About GenBase Tables

Introduction

This section contains a list of GenBase tables, a description of each, and a list of their unique and shared fields.

GenBase Tables

Table 2-1 contains a complete list of tables used by the GenBase Database application, including each table's shared fields.

Table 2-1 GenBase tables

Table	Contents	Shared Fields
Alleles	Defines alleles. You can specify allele frequencies for a particular population.	Marker/Trait Name Allele Name
Alternate Marker Name Sets	List of sets of marker-name synonyms. (Alternate marker names are recorded in the Markers table.)	Name Set
Called Genotypes	List of final genotype results from individuals. May be derived from the Genotyper software or from external sources.	Individual ID Marker Allele
Categories	Lists categories and associates them with a panel and marker.	Panel Name Marker Name Category Name
Category Members	Stores category definitions from the Genotyper software. This table cannot be modified in the GenBase Database application.	Category Name
External Sources	List of sources (other than the Genotyper software) of called genotype data.	Source Name
Gels	Names of gel files used in imported Genotype records.	None
Genotypes	Stores genotype results from the Genotyper software. Can be displayed as Layout or Summary.	None
Liability Classes	Defines liability classes for traits.	Trait Name Liability Class
Maps	List of names of maps. (Map locations are recorded in the Markers table.)	Map Name

Table 2-1 GenBase tables *(continued)*

Table	Contents	Shared Fields
Markers	Defines markers. You can specify a location (for a particular Map) and alternate names (for a particular Alternate Marker Name Set).	Marker Name Map Name Alternate Name
Panels	List of sets of Genotyper categories already imported into the GenBase Database application.	Panel Name Marker Name
Pedigree Structures	List of names of hypotheses for Pedigree relationships.	Pedigree Name
Pedigrees	Pedigree information for an individual that relates to a Pedigree hypothesis (Pedigree Structure).	Individual ID Structure Name
Penetrances	Penetrance data for trait, liability class, and genotypes.	Trait Name Liability Class Allele
Phenotypes	Trait information for individuals.	Individual ID Trait Name
Populations	List of populations being studied.	Population
QTL Variances	Covariance for two quantitative traits or variance for a single quantitative trait in LINKAGE QTL format.	Variance
Samples	Matches Sample IDs to individuals (there can be multiple DNA samples for each individual).	Sample ID Individual ID
Traits	Defines traits being studied.	Trait Name

continued on next page

Fields

The GenBase Database application fields can be broken down into the following types:

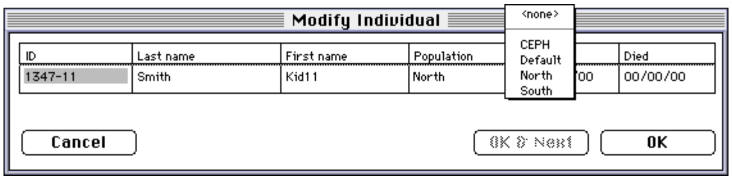
- *Unique*–This field only exists in one table, which means you cannot conduct queries based on this field.
  - For example, the Male Mutation Rate of a particular trait does not appear in any other table than Traits, and you cannot conduct database searches using this value.
- *Shared Fields*–A field that appears in more than one table, and can be used to find records in multiple tables that relate to that field.
  - For example, if you searched for records related to a record in the individual table, you may find records in several different tables related to the record in question by the individual’s name.

Related Records

The GenBase Database application uses the term *related records* to designate records that share fields with other tables. There are several instances where these relations are important.

Entering Data

When you manually enter data in the GenBase Database application, related fields generally use pop-up menus for data entry to ensure that only correct data is entered. For example, if you enter an individual (see Figure 2-9), you must assign that individual a population already designated in the Population table.

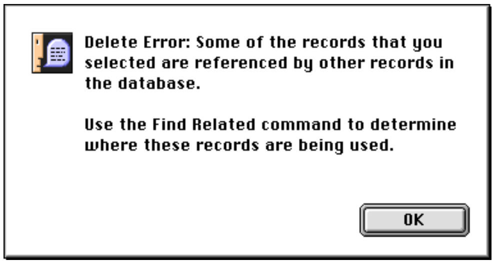


ID	Last name	First name	Population		Died
1347-11	Smith	Kid11	North	00	00/00/00

Figure 2-9 Creating a record with related field

Deleting Data

The GenBase Database application prevents you from deleting records that share fields with other records. GenBase does this to prevent you from creating broken cross-references within the database. When you try to delete such a record, you see an error message similar to the one in Figure 2-10. In some tables (for example, the Genotype table) only users with database access are allowed to delete records.



Delete Error: Some of the records that you selected are referenced by other records in the database.

Use the Find Related command to determine where these records are being used.

Figure 2-10 Delete error message

In order to delete the record in question, you must first delete (or modify) all related records.

Importing Text Files

When you import text files into a GenBase table, new records are created for shared fields if the file contains a different entry from the available choices.

For example, Figure 2-9 shows the available populations in the database, and limits your manual entries to those choices. However, if you import a text file containing an entry for population that doesn't match one of the existing choices (for example, East), the GenBase Database application creates a record in the Population table for East.

The GenBase Database application also creates a record if you misspell an existing entry. In the example above, if the text file contains "Nroth" instead of "North," GenBase assumes you want to create a new population "Nroth" and creates a record in the Population table.

Finding Related Records

The GenBase Database application provides a simple way to find all the records that are related to the record you have selected. Choose Find Related Record from the Record menu, and a dialog appears telling you which tables contain related records. When you open those tables, only the related records are displayed.

Getting Started

3

Chapter Overview

Introduction This chapter is intended to allow you to start using the GenBase Database application with the included Example Database. This database should not be used to store your own data, but does give you an opportunity to experiment with program features and commands.

This chapter also includes a breakdown of the GenBase user interface and procedures for setting up your user name and assigning yourself a password.

In This Chapter This chapter contains the following topics:

Topic	See Page
Getting Started	3-2
GenBase Interface	3-5
Exploring the Example Database	3-11

Getting Started

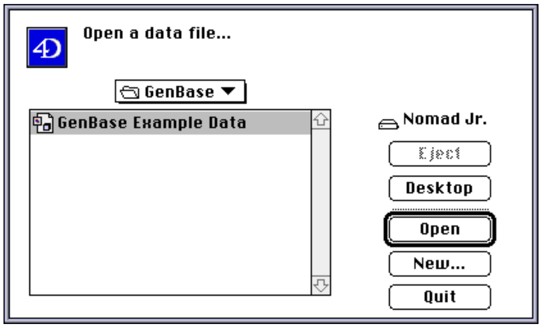
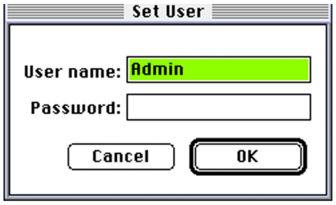
Opening the Included Database

You can start using the GenBase Database application immediately by opening the Example Database and experimenting with the various commands and features of GenBase without fear of losing your data.

When you are ready to set up and use your own database, see Chapter 4, “Preparing the Database.”

IMPORTANT Do not use the Example Database for your own data.

To open the Example Database:

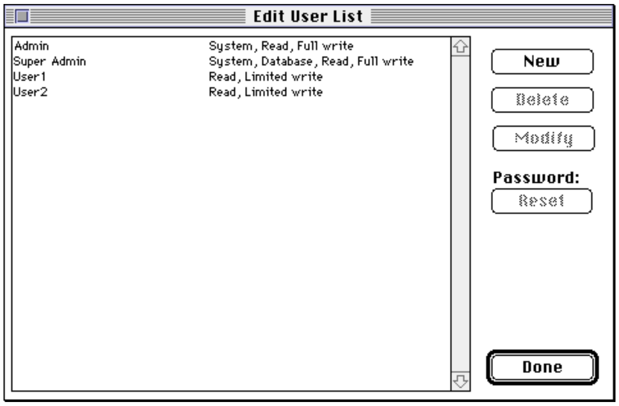
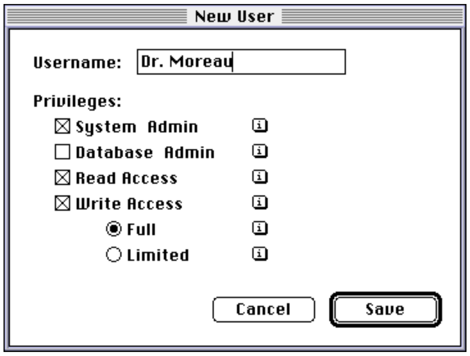
Step	Action
1	While holding down the Option key, double-click the GenBase icon.Continue to hold down the option key until the following dialog appears: 
2	In the Open dialog box, find the Example Database and click Open.
3	Enter Admin in the User Name field (leave the Password blank) and press Return.  User name: Admin Password:
4	The GenBase Database application is now open. You have Admin access, which allows you to edit the user tables and read/write to data tables.

continued on next page

Editing the User List

You can choose to change your name from the default (Admin), or you can assign yourself different access. The access required to set up tables and to use the database is Read and Full write. You must have System Admin access to edit the user list.

To assign yourself the correct access:

Step	Action
1	Select Edit User List from the Admin Menu (requires System Admin access). The following dialog appears:  Admin System, Read, Full write Super Admin System, Database, Read, Full write User1 Read, Limited write User2 Read, Limited write
2	Click New. The following dialog appears:  Username: Dr. Moreau Privileges: ☒ System Admin ☐ Database Admin ☒ Read Access ☒ Write Access ● Full ○ Limited **Note** For more information about the selections, click the “i” button next to the access.

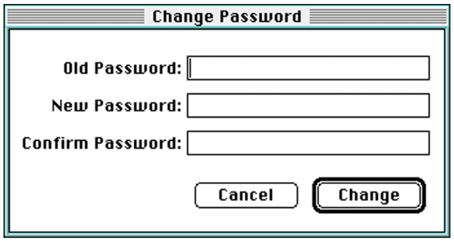
To assign yourself the correct access: *(continued)*

Step	Action
3	Enter your name.
4	Click Save, then Done in the Edit User dialog box.

Setting Your Password

The GenBase Database application provides the option of password protection for each user name.

To assign yourself a password:

Step	Action
1	Select Change Password from the Admin menu. The following dialog box appears: 
2	Leave the Old Password field blank, and enter your password in the New Password field. **Note** Be sure to write down the password. You can not recover a lost password. If your password is lost, a user with System Admin access must reset (clear) the password.
3	Re-enter your password in the Confirm Password field.
4	Click Change to accept the password. **Note** If you incorrectly entered your password in the confirmation field, the GenBase Database application will alert you, and you must re-enter your password.

GenBase Interface

Introduction

The GenBase Database application is a relational database which manages information through tables. Tables are the primary tools for entering, viewing and modifying data. You may want to configure certain tables prior to importing data from the Genotyper software, the GenoPedigree software, or text files.

The major interface structures in GenBase are

- *The Main Window*–Main interface from which you can establish links with the Genotyper and GenoPedigree software, as well as opening GenBase tables.
- *Data Tables*–Formatted tables for storing Genotyper or GenoPedigree data. You can print or create tab delimited text files directly from the tables.
- *The Genotype Record*–Table that only contains genotype records imported from the Genotyper software. From this table you can review the imported records and append your comments directly to the record.

The Main Window

When you open the GenBase Database application, the main window appears with a Summary table that provides information about the database. Clicking on a table name in the Summary opens the relevant table in the software. The buttons on the lower left corner link the Genotyper software or the GenoPedigree software to the GenBase Database application if the applications are currently open.

Table 3-1 describes the menu bar options on the main window.

Table 3-1 Main window menu bar options

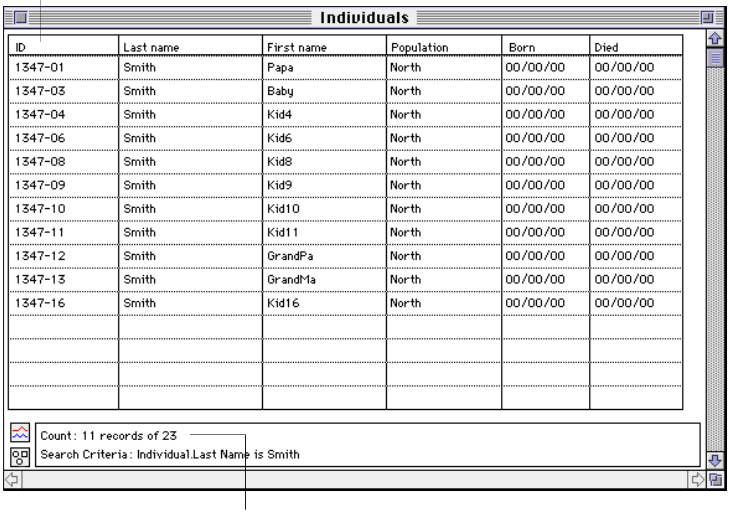
Menu Item	Description
File	Contains basic Macintosh options limited to setting the user, printing individual tables, or quitting the software.
Edit	Contains basic Macintosh options such as Cut, Copy, Paste and Undo. **Note** Most tasks in the GenBase Database application cannot be Undone.
Table	Opens tables that allow you to enter and view relational database information.

Table 3-1 Main window menu bar options *(continued)*

Menu Item	Description
Records	Contains commands for adding or modifying new records, sorting and searching routines, generating and printing complex reports, and importing or exporting records.
Special	Contains special routines for organizing, viewing, and editing database information.
Admin	Contains access to the user list that allows you to edit a user's level of access, or to change the current user's password.
Windows	Contains commands to bring open windows to the front.

Data Tables

The GeneScan Analysis software uses tables to store, sort, and display database information. These tables provide easy access to data fields, and allow you to quickly search and modify data. Advanced users can access more complex data relationships to build custom search, sort, and print procedures. You can change existing records or manually add new records in most tables.



ID	Last name	First name	Population	Born	Died
1347-01	Smith	Papa	North	00/00/00	00/00/00
1347-03	Smith	Baby	North	00/00/00	00/00/00
1347-04	Smith	Kid4	North	00/00/00	00/00/00
1347-06	Smith	Kid6	North	00/00/00	00/00/00
1347-08	Smith	Kid8	North	00/00/00	00/00/00
1347-09	Smith	Kid9	North	00/00/00	00/00/00
1347-10	Smith	Kid10	North	00/00/00	00/00/00
1347-11	Smith	Kid11	North	00/00/00	00/00/00
1347-12	Smith	GrandPa	North	00/00/00	00/00/00
1347-13	Smith	GrandMa	North	00/00/00	00/00/00
1347-16	Smith	Kid16	North	00/00/00	00/00/00

Count: 11 records of 23

Search Criteria: Individual.Last Name is Smith

Each row in the table is a record, which is broken down into one or more fields (six in this example). Each record is distinguished by the first field, or unique identifier (in this example, ID).

Number of records shown out of the total for the table. If only certain records are shown, the search criteria also is displayed (in this case, individuals whose last names are Smith).

Records

Each row in a table is an individual record. Each record contains one or more fields, which may be unique to the table or shared with other tables. If two records in different tables have the same entry in a shared field, those two records are related.

You can modify, add, or delete records in most tables. These commands are located in the Record menu and are grayed out if the commands are unavailable for the particular table. If the commands are available, double clicking a record opens the Modify dialog box (if the record has an entry) or the Add Record dialog box (if the record is empty).

Fields

Most tables are related to other tables by shared fields. For example, the Sample table assigns sample numbers to specific individual IDs, which are the same as the IDs in the individual table (see "Finding Related Records" on page 3-21).

Shared fields often dictate how data can be entered in the tables. In the previous example, you could not assign a Sample to an individual unless that individual had been entered in the Individual table.

Unique fields are only present in one table. No other records can be related to a unique field.

Entering Data

In order for you to use the database for your own data, you must fill in tables manually, or import preformatted tables from other applications. For an explanation of the proper set-up procedure and the purpose of each table, see Chapter 4, "Preparing the Database."

Exporting Tables

Data in tables can be exported to the Genotyper software and the GenoPedigree software, as well as to tab-delimited text files. The GenBase Database application also gives you the option of converting data into CRI-MAP, LINKAGE, or GENEHUNTER files.

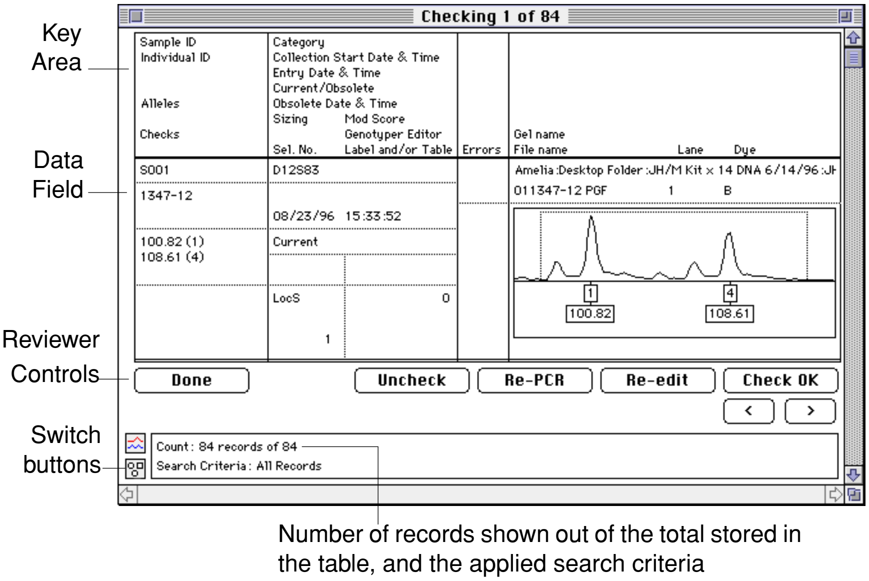
continued on next page

The Genotype Record

Genotype records imported from the Genotyper software are stored in the Genotype table. The Genotype table cannot be edited manually. You can only add records to this table from the Genotyper software.

This table is also used to review records. Selecting Check Genotypes from the Special menu allows you to review genotype records for accuracy (see Figure 3-1 below and Table 3-2 on page 3-9). Your recommendations become part of the record (see "Checking Imported Genotype Records" on page 5-17). You can search this table based on reviewer comments or their recommendations using the Find Genotype command in the Record menu (see "Find Genotypes" on page 3-19).

Access the Genotype table through the Table menu or by clicking the table name in the Main window.



Checking 1 of 84

Sample ID
Individual ID
Alleles
Checks

Category
Collection Start Date & Time
Entry Date & Time
Current/Obsolete
Obsolete Date & Time
Sizing Mod Score
Genotyper Editor
Sel. No. Label and/or Table

Errors

Gel name
File name Lane Dye

S001
1347-12
100.82 (1)
108.61 (4)

D12S83
08/23/96 15:33:52
Current
LocS 0
1

Amelia:Desktop Folder:JH/M Kit x 14 DNA 6/14/96:JH
011347-12 PGF 1 B

Count: 84 records of 84
Search Criteria: All Records

Figure 3-1 Genotype record

Table 3-2 Genotype record fields

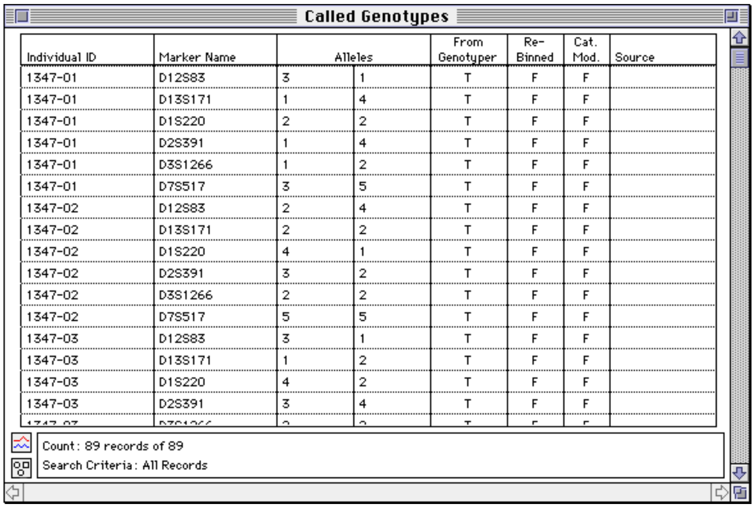
Field	Description
Key Area	Describes the field entries in the data area below.
Data Area	Provides data about the Genotype record, including sample and individual ID, who checked the data, and a display of individual sample and Allele peaks.
Reviewer Controls	Grayed out buttons are for use during review of data only.
Application Switch Buttons	Brings either the Genotyper software or the GenoPedigree software to the foreground, if the programs are already open.
Count	Number of records shown out of the total number stored in the table.
Search Criteria	If only selected records are shown (less than the total number in the table), this field shows the criteria by which they were selected.
Layout Pull-down	Allows you to switch between the graphical and summary views of the Genotypes table.

The Called Genotype Table

After you review Genotype records, you can copy the record to the Called Genotype table. Called Genotypes are the end result of data review, and should be created only when you are sure the Genotype records have been properly reviewed.

You can also import data directly into the Called Genotype table, or add new records manually using the New (⌘-N) command in the Record Menu. Data imported into the Called Genotype table is called “External Source Data.” You can designate the source in the External Source table, which shares the Source Name field with the Called Genotype table (Table 3-2 on page 3-10).

Importing External Source data or manually creating new Called Genotypes circumvents the review process the GenBase Database application provides for Genotype data imported from the Genotyper software. Whenever possible, use only genotype records from the ABI PRISM family of Genotyping applications.



Called Genotypes

Individual ID	Marker Name	Alleles		From Genotyper	Re-Binned	Cat. Mod.	Source
1347-01	D12S83	3	1	T	F	F	
1347-01	D13S171	1	4	T	F	F	
1347-01	D1S220	2	2	T	F	F	
1347-01	D2S391	1	4	T	F	F	
1347-01	D3S1266	1	2	T	F	F	
1347-01	D7S517	3	5	T	F	F	
1347-02	D12S83	2	4	T	F	F	
1347-02	D13S171	2	2	T	F	F	
1347-02	D1S220	4	1	T	F	F	
1347-02	D2S391	3	2	T	F	F	
1347-02	D3S1266	2	2	T	F	F	
1347-02	D7S517	5	5	T	F	F	
1347-03	D12S83	3	1	T	F	F	
1347-03	D13S171	1	2	T	F	F	
1347-03	D1S220	4	2	T	F	F	
1347-03	D2S391	3	4	T	F	F	

Count : 89 records of 89
Search Criteria : All Records

Figure 3-2 Called Genotype table

When called, the records copied from the Genotype table are not deleted.

Note Not all of the Genotype data is moved to the Called Genotype table.

The GenBase Gear Window

You may see the GenBase Gear window open and close at the bottom of your screen periodically (Figure 3-3). The GenBase Database application displays this window when it is writing records to the disk. You don't need to interact with it.

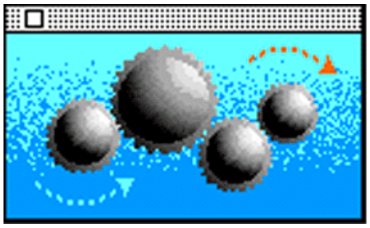


Figure 3-3 GenBase Gear window

Exploring the Example Database

Introduction

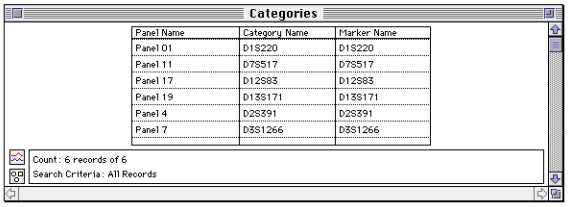
This section describes standard the GenBase Database application commands to use on the example database followed by examples of procedures.

- *Changing Data*–Modifying existing table records
- *Adding Data*–Adding records to tables
- *Checking Data*–Reviewing Genotype records using GenBase's review functions
- *Exporting Data*–Creating LINKAGE or CRI-MAP files from table data
- *Searching the Database*–Using GenBase's database tools to search table records and the database
- *Sorting Records*–Using the GenBase Sort command to reorder tables
- *Printing Records*–Creating custom printed reports from GenBase table data

Changing Data

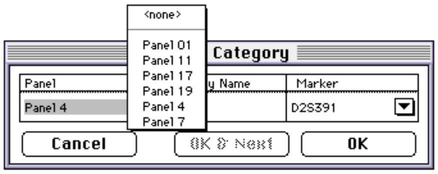
You can change information in most fields of the GenBase tables. The procedure for each table is similar. What changes are the actual fields and whether the interface uses pull-down menus or text entry fields. For this example, we'll change a record in the Categories table.

To change a record in the categories table:

<table>
<tr><th>Step</th><th>Action</th></tr>
<tr><td>1</td><td>Open the Categories table by selecting it from the Table menu. The following window appears:


<table>
<tr><th>Panel Name</th><th>Category Name</th><th>Marker Name</th></tr>
<tr><td>Panel 01</td><td>D1S220</td><td>D1S220</td></tr>
<tr><td>Panel 11</td><td>D7S517</td><td>D7S517</td></tr>
<tr><td>Panel 17</td><td>D12S83</td><td>D12S83</td></tr>
<tr><td>Panel 19</td><td>D13S171</td><td>D13S171</td></tr>
<tr><td>Panel 4</td><td>D2S391</td><td>D2S391</td></tr>
<tr><td>Panel 7</td><td>D3S1266</td><td>D3S1266</td></tr>
</table>
Count : 6 records of 6
Search Criteria : All Records
</td></tr>
</table>

To change a record in the categories table: *(continued)*

Step	Action
2	Double-click the record you want to modify (for example, Panel 01). or Highlight a record and select Modify (⌘-M) from the Record menu. The following dialog box appears:  Panel Panel 4 y Name Marker D2S391
3	Select a new Panel from the Panel pop-up menu. **Note** Pop-up menus are used for fields that are dependent on entries in other tables. For example, the Panel field uses a pop-up menu because the new entry must be a panel from Panels table. If there were no entries in the Panels table, the pop-up menu would contain no choices.
4	Type in a new Category Name in the Category Name field.
5	Select a Marker from the Marker pop-up menu.
6	Click OK to accept your changes.

Adding Data

You can manually add records to most tables in the database. You will enter data in the record fields in one of three ways.

- *Text Fields*–Only restriction to data entered in these fields is the total number of characters (usually 12 to 15) or integers allowed.
- *Pop-up Menus*–Typically these fields include options based on fields on other tables. For example, in the Categories table, you can only choose from a selection of Panels that have already been entered in the Panels table.
- *Checkboxes*–Y or N entry fields.

To add records to a table:

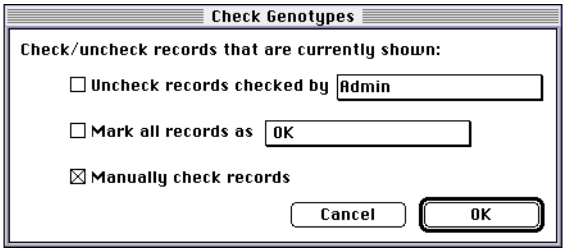
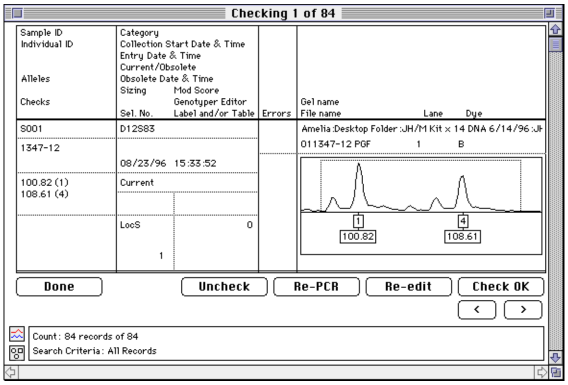
Step	Action
1	Select Categories from the Tables menu.
2	Select New Record (⌘-N) from the Records menu. The following dialog box appears: 
3	Select a new Panel from the Panel pop-up menu. **Note** Pop-up menus are used for fields that are dependent on entries in other tables. For example, the Panel field uses a pop-up menu because the entry must be a panel from the Panels table. If there were no entries in the Panels table, the pop-up menu would contain no choices, and you could not create a record.
4	Type in a new Category Name in the Category Name field.
5	Select a Marker from the Marker pop-up menu.
6	Click OK to accept the new record, OK & Next to create more records.

Checking Imported Genotyper Records

The GenBase Database application allows you to import Genotyper records and have one or more people review them for accuracy. Once the records have been reviewed, you can add them to the Called Genotype table to create printed reports or to export the data to downstream applications (LINKAGE or CRI-MAP).

You can search the Genotyper table by reviews, or by their comments (see “Find Genotypes” on page 3-19).

To check imported Genotypes:

Step	Action
1	Open the Genotypes table by selecting Genotypes from the Tables menu.
2	Select Check Genotypes from the Special menu. The following window appears:  Check Genotypes Check/uncheck records that are currently shown: ☐ Uncheck records checked by Admin ☐ Mark all records as OK ☒ Manually check records
3	Click OK. The following window appears. **Note** In this case, the defaults in the window are appropriate. The unchecked selections allow you to quickly check or uncheck all Genotyper records.  Checking 1 of 84 Sample ID: S001 Individual ID: 1347-12 Alleles: 100.82 (1) 108.61 (4) Category: D12S83 Entry Date & Time: 08/23/96 15:33:52 Current/Obsolete: Current Sizing: LocS Mod Score: 0 Sel. No.: 1 Gel name: Amelia :Desktop Folder :JH/M Kit x 14 DNA 6/14/96 :JH File name: 011347-12 PGF Lane: 1 Dye: B Count : 84 records of 84 Search Criteria : All Records

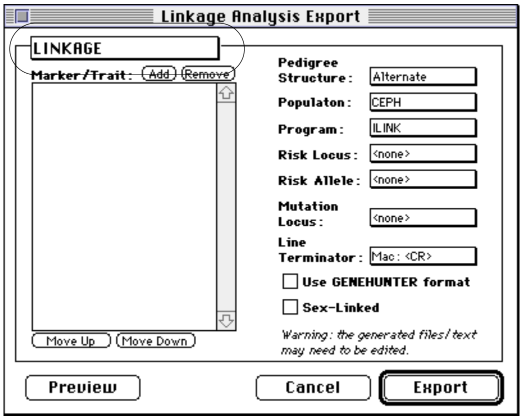
To check imported Genotypes: *(continued)*

Step	Action
4	Review the Genotyper record for accuracy. When finished, choose one of the following options: ♦ Exit the review by clicking Done. ♦ Remove your review comments by clicking Uncheck. ♦ Recommend that the record be re-run by clicking Re-PCR. ♦ Recommend the record be re-edited in the Genotyper software by clicking Re-edit. ♦ Recommend the Genotyper record be moved to the called Genotypes table by clicking Check OK. ♦ Move on to the next/previous record by clicking the arrows.
5	Your user name and recommendation are entered on each record you review.

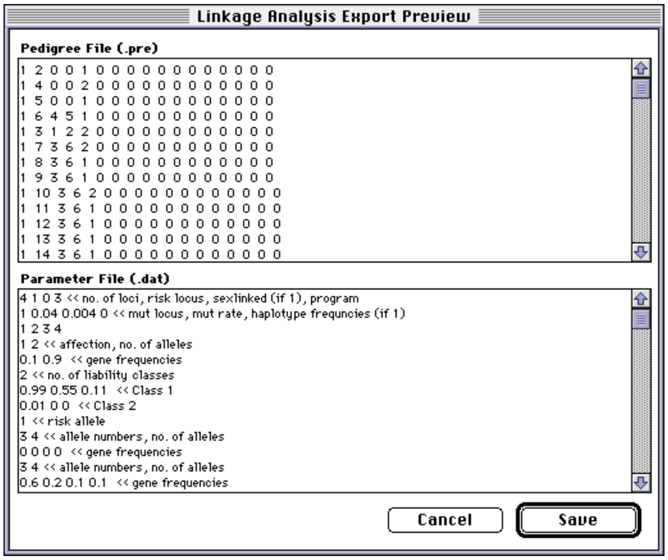
Creating Exportable Files

You can export data by creating LINKAGE or CRI-MAP files using the Linkage Analysis Export command from the Special menu. You can then generate custom files containing pedigree and locus information.

To create exportable files:

Step	Action
1	Select Linkage Analysis Export from the Special menu. The following dialog box appears:  Linkage Analysis Export LINKAGE Pedigree Structure: Alternate Populaton: CEPH Program: ILINK Risk Locus: <none> Risk Allele: <none> Mutation Locus: <none> Line Terminator: Mac: <CR> ☐ Use GENEHUNTER format ☐ Sex-Linked *Warning: the generated files/text may need to be edited.*
2	Select an export format from the pop-up menu (circled in the figure above). The default is LINKAGE.

To create exportable files: *(continued)*

Step	Action
3	Add Markers or Traits by clicking Add (above the scroll box).
4	Select additional information from the pop-up menus.
5	To see the file before exporting it, click preview. The following dialog box appears: 

```
1 2 0 0 1 0 0 0 0 0 0 0 0 0 0 0 0
1 4 0 0 2 0 0 0 0 0 0 0 0 0 0 0 0
1 5 0 0 1 0 0 0 0 0 0 0 0 0 0 0 0
1 6 4 5 1 0 0 0 0 0 0 0 0 0 0 0 0
1 3 1 2 2 0 0 0 0 0 0 0 0 0 0 0 0
1 7 3 6 2 0 0 0 0 0 0 0 0 0 0 0 0
1 8 3 6 1 0 0 0 0 0 0 0 0 0 0 0 0
1 9 3 6 1 0 0 0 0 0 0 0 0 0 0 0 0
1 10 3 6 2 0 0 0 0 0 0 0 0 0 0 0 0
1 11 3 6 1 0 0 0 0 0 0 0 0 0 0 0 0
1 12 3 6 1 0 0 0 0 0 0 0 0 0 0 0 0
1 13 3 6 1 0 0 0 0 0 0 0 0 0 0 0 0
1 14 3 6 1 0 0 0 0 0 0 0 0 0 0 0 0
```

```
4 1 0 3 << no. of loci, risk locus, sexlinked (if 1), program
1 0.04 0.004 0 << mut locus, mut rate, haplotype frequncies (if 1)
1 2 3 4
1 2 << affection, no. of alleles
0.1 0.9  << gene frequencies
2 << no. of liability classes
0.99 0.55 0.11  << Class 1
0.01 0 0  << Class 2
1 << risk allele
3 4 << allele numbers, no. of alleles
0 0 0 0  << gene frequencies
3 4 << allele numbers, no. of alleles
0.6 0.2 0.1 0.1  << gene frequencies
```

Step	Action
6	If necessary, edit the information in both windows as you would a text file.
7	Click Save to save the file in the format you have selected. The GenBase Database application creates both a.pre and.dat Linkage file.

continued on next page

Searching the Database

You can search through data on individual tables using the Find command, or search Genotyper records using the Find Genotypes command. You can also search the entire database for records related to the one selected by using the Find Related Records command.

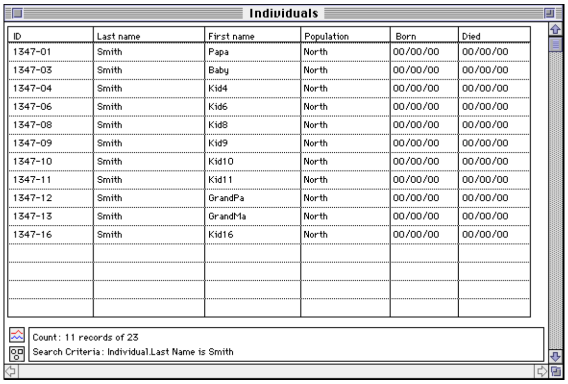
Using the Find Command

Before you begin the search, use the radio buttons to select whether to clear the list before a search, to search within an already refined list, or add the results of your search to an already created list.

To use the Find command:

Step	Action
1	Open the table in which you want to find records. **Note** You can have multiple tables open when executing the Find command. However, only the active window will be searched.
2	Select Find (⌘-F) from the Record menu. The Find dialog box appears.  Find Find Individuals where: Individual Last Name is Smith List: ● Create New ○ Search Within ○ Append
3	Select the primary field to search from the first pop-up menu. **Note** Only fields available to the open table can be searched.
4	Select the secondary field to search from the second pop-up menu.
5	Select the search operator. The values in this pop-up menu vary depending on the type of field selected in the second pop-up menu.
6	Enter the value to search for.

To use the Find command: *(continued)*

<table>
<tr><th>Step</th><th>Action</th></tr>
<tr><td>7</td><td>Using the radio buttons, select whether to:
♦ <i>Clear list, then search</i> – Searches the entire table
♦ <i>Search within list</i> – Searches within the current list of records displayed in the table
♦ <i>Append to list</i> – Searches the entire table, then appends the records to the current list of records</td></tr>
<tr><td>8</td><td>Click Search.</td></tr>
<tr><td>9</td><td>The records that match the search criteria are shown.
Note The search criteria used is listed at the bottom of the screen.


<table>
<tr><th>ID</th><th>Last name</th><th>First name</th><th>Population</th><th>Born</th><th>Died</th></tr>
<tr><td>1347-01</td><td>Smith</td><td>Papa</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-03</td><td>Smith</td><td>Baby</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-04</td><td>Smith</td><td>Kid4</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-06</td><td>Smith</td><td>Kid6</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-08</td><td>Smith</td><td>Kid8</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-09</td><td>Smith</td><td>Kid9</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-10</td><td>Smith</td><td>Kid10</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-11</td><td>Smith</td><td>Kid11</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-12</td><td>Smith</td><td>GrandPa</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-13</td><td>Smith</td><td>GrandMa</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-16</td><td>Smith</td><td>Kid16</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
</table>
Count : 11 records of 23
Search Criteria : Individual.Last Name is Smith</td></tr>
<tr><td>10</td><td>The table remains in this configuration as long as the program is open. To show all records, select Show All (⌘-J) from the Record menu.</td></tr>
</table>

Find Genotypes

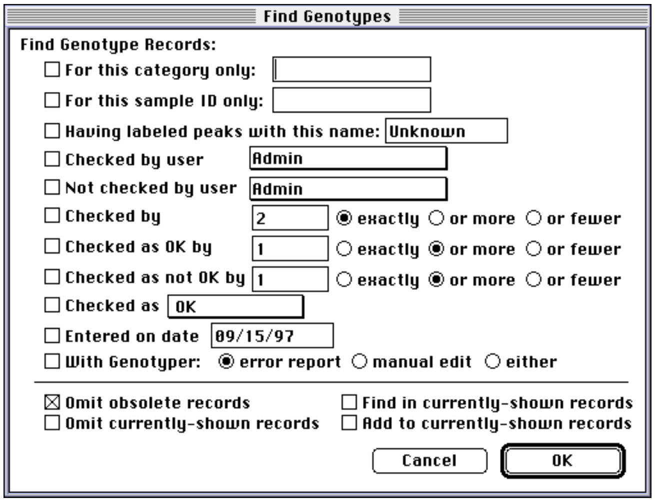
The Find Genotypes command allows searches of the Genotypes table based on criteria that includes reviewer name and review recommendation.

For example, to find genotype records with the following criteria, construct a search to find the matching Genotypes:

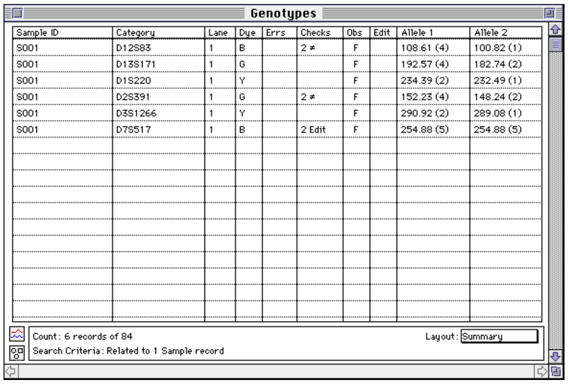
- Records for sample S001
- Records that at least one reviewer marked for “Re-PCR”

The Find Genotypes command is only available when the Genotypes table is the active window.

To find genotypes:

Step	Action
1	Open the Genotypes table by clicking the table name in the Main window or by selecting Genotypes from the Table menu.
2	Select the Find Genotype Records in the User menu. The following window appears:  Find Genotype Records: ☐ For this category only: ☐ For this sample ID only: ☐ Having labeled peaks with this name: Unknown ☐ Checked by user Admin ☐ Not checked by user Admin ☐ Checked by 2 ● exactly ○ or more ○ or fewer ☐ Checked as OK by 1 ○ exactly ● or more ○ or fewer ☐ Checked as not OK by 1 ○ exactly ● or more ○ or fewer ☐ Checked as OK ☐ Entered on date 09/15/97 ☐ With Genotyper: ● error report ○ manual edit ○ either ☒ Omit obsolete records ☐ Find in currently-shown records ☐ Omit currently-shown records ☐ Add to currently-shown records
3	Check the Checked by User Box, and select User 1 from the pop-up menu.
4	Check the Checked As box, and select Re-PCR from the pop-up menu.
5	Check the With Genotyper box, and click the “manual edit” radio button.

To find genotypes: *(continued)*

Step	Action
6	Click OK. The records that match the criteria are displayed in the Genotypes table. 

Sample ID	Category	Lane	Dye	Errs	Checks	Obs	Edit	Allele 1	Allele 2
S001	D12S83	1	B		2 ≠	F		108.61 (4)	100.82 (1)
S001	D13S171	1	G			F		192.57 (4)	182.74 (2)
S001	D1S220	1	Y			F		234.39 (2)	232.49 (1)
S001	D2S391	1	G		2 ≠	F		152.23 (4)	148.24 (2)
S001	D3S1266	1	Y			F		290.92 (2)	289.08 (1)
S001	D7S517	1	B		2 Edit	F		254.88 (5)	254.88 (5)

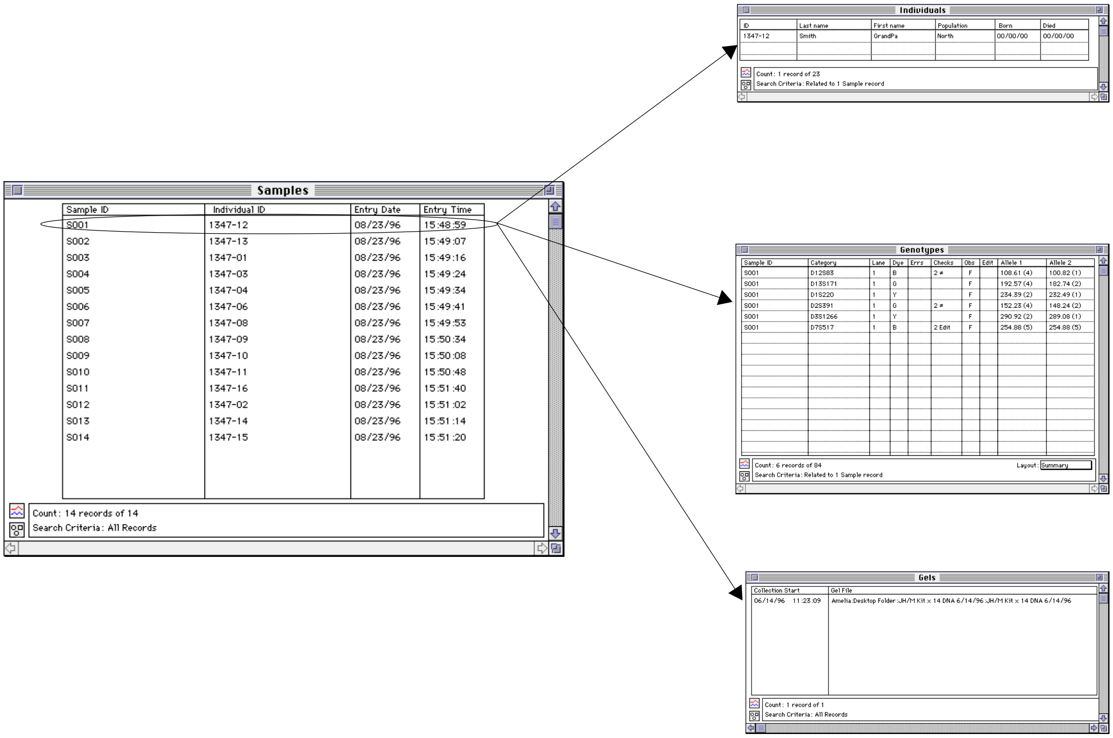
Count : 6 records of 84

Layout : Summary

Search Criteria : Related to 1 Sample record

Finding Related Records

The Find Related Records command searches the entire database for records that are directly related to the unique identifier of the selected record. Figure 3-4 shows records related to the Sample S001.



Sample ID	Individual ID	Entry Date	Entry Time
S001	1347-12	08/23/96	15:48:59
S002	1347-13	08/23/96	15:49:07
S003	1347-01	08/23/96	15:49:16
S004	1347-03	08/23/96	15:49:24
S005	1347-04	08/23/96	15:49:34
S006	1347-06	08/23/96	15:49:41
S007	1347-08	08/23/96	15:49:53
S008	1347-09	08/23/96	15:50:34
S009	1347-10	08/23/96	15:50:08
S010	1347-11	08/23/96	15:50:48
S011	1347-16	08/23/96	15:51:40
S012	1347-02	08/23/96	15:51:02
S013	1347-14	08/23/96	15:51:14
S014	1347-15	08/23/96	15:51:20

Count: 14 records of 14
Search Criteria: All Records

ID	Last name	First name	Population	Born	Died
1347-12	Smith	GrandPa	North	00/00/00	00/00/00

Count: 1 record of 23
Search Criteria: Related to 1 Sample record

Sample ID	Category	Lane	Dye	Errs	Checks	Obs	Edit	Allele 1	Allele 2
S001	D12S83	1	B		2 ≠	F		108.61 (4)	100.82 (1)
S001	D13S171	1	G			F		192.57 (4)	182.74 (2)
S001	D1S220	1	Y			F		234.39 (2)	232.49 (1)
S001	D2S391	1	G		2 ≠	F		152.23 (4)	148.24 (2)
S001	D3S1266	1	Y			F		290.92 (2)	289.08 (1)
S001	D7S517	1	B		2 Edit	F		254.88 (5)	254.88 (5)

Count: 6 records of 84
Search Criteria: Related to 1 Sample record
Layout: Summary

Collection Start	Gel File
06/14/96 11:23:09	Amelia:Desktop Folder:JH/M Kit x 14 DNA 6/14/96:JH/M Kit x 14 DNA 6/14/96

Count: 1 record of 1
Search Criteria: All Records

Figure 3-4 Related records

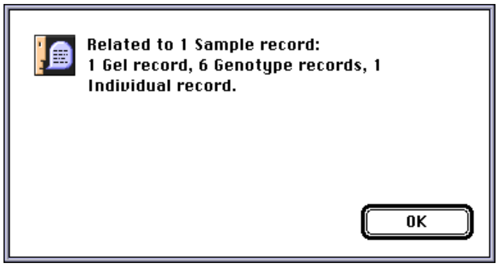
These tables do not automatically open when you select Find Related Records. You must open the relevant table manually (after you conduct a search, a dialog appears informing you of the records the GenBase Database application found). Once open, the tables display only the records related to the search.

Note The number of displayed records and the search criteria is always displayed at the bottom of the table.

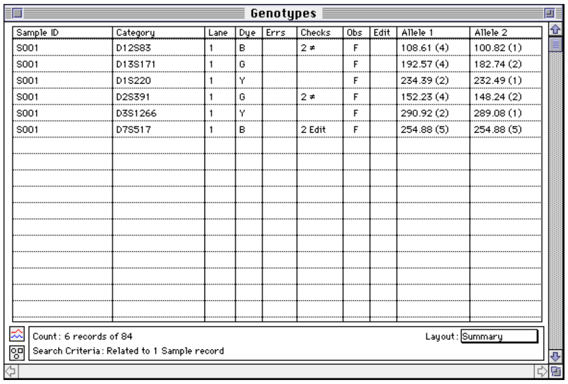
This command applies to the following tables:

- Sample
- Genotype
- Pedigree
- Called Genotype
- Individual Trait
- Individual
- Genotype Summary
- Category
- Category Member
- Panels
- Markers
- Gels

To find related records:

Step	Action
1	Select the record or records for which you want to find related records. **Note** This can be single or multiple genotype records, or entries from a table.
2	Select Find Related Record from the Records menu.
3	A summary of the related records is displayed in the following dialog box:  Related to 1 Sample record: 1 Gel record, 6 Genotype records, 1 Individual record.

To find related records: *(continued)*

Step	Action
4	Close the dialog box, and open the designated tables to view the related records. **Note** In this example (which had two related records in the Genotype table), you could then select the Genotypes table from the Table menu. The related records are displayed, as in this example. 

Genotypes

Sample ID	Category	Lane	Dye	Errs	Checks	Obs	Edit	Allele 1	Allele 2
S001	D12S83	1	B		2 ≠	F		108.61 (4)	100.82 (1)
S001	D13S171	1	G			F		192.57 (4)	182.74 (2)
S001	D1S220	1	Y			F		234.39 (2)	232.49 (1)
S001	D2S391	1	G		2 ≠	F		152.23 (4)	148.24 (2)
S001	D3S1266	1	Y			F		290.92 (2)	289.08 (1)
S001	D7S517	1	B		2 Edit	F		254.88 (5)	254.88 (5)

Count: 6 records of 84

Search Criteria: Related to 1 Sample record

Layout: Summary

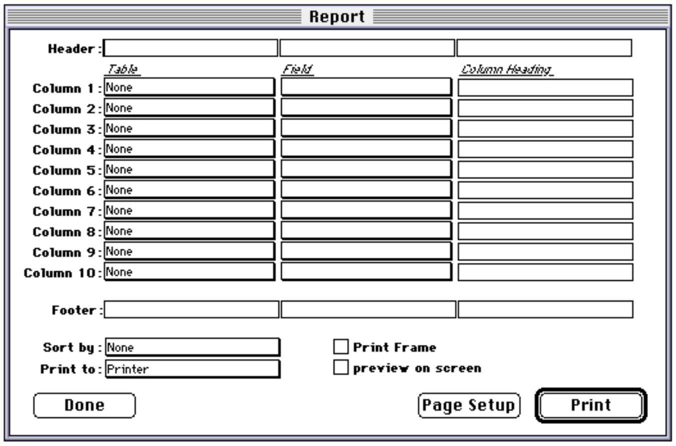
continued on next page

Printing Reports

The Report command under the Record menu provides a way to construct custom reports in a format that you select. You can then print the report, or create a tab delimited text file.

You may then save these report formats for future use.

To print reports:

Step	Action
1	Select in the Table menu the table for which you want to create a report.
2	Select Report (⌘-R) from the Record menu. The following dialog box appears:  Column 1 : None Column 2 : None Column 3 : None Column 4 : None Column 5 : None Column 6 : None Column 7 : None Column 8 : None Column 9 : None Column 10 : None Sort by : None Print to : Printer ☐ Print Frame ☐ preview on screen
3	Enter the header and footer for the printed document.
4	Select the contents for each column using the two pop-up menus. **Note** Your selection will be displayed in the Column Heading field to the right. You may highlight and change the contents of this field if necessary.
5	Use the “Sort by” pop-up menu to select sort criteria, if needed.
6	From the “Print to” pop-up menu, select the Printer to send the document to the default printer, or Disk File to convert the report to tab delimited text and save it as a file.
7	Use the Print Frame checkbox to print a frame around the report.

To print reports: *(continued)*

Step	Action
8	Check “Preview on screen” to see your document before it is sent to the printer. **Note** This is a recommended step that can save you time and paper.
9	Click Print. **Note** If you selected “Preview on screen”, a preview will appear. You can zoom in to particular sections of the document by placing or dragging the zoom box on an area and clicking Zoom. You may either cancel or continue printing from that point.

Preparing the Database

4

Chapter Overview

Introduction Prior to importing data, you should enter information in the database that relates to the records you plan to import from the Genotyper software or the GenoPedigree software. The Genotyper software only imports Categories and Genotypes, and without the related information (individuals, sample, etc.), the GenBase Database application cannot create export files or transfer complete information to the GenoPedigree software.

It is possible to import data without setting up the database, but you must first set up the tables you plan to use after importing data.

Note Most of the procedures in this chapter require Read and Full Write access to the GenBase Database application.

In This Chapter This chapter contains the following topics:

Setting-Up the Database

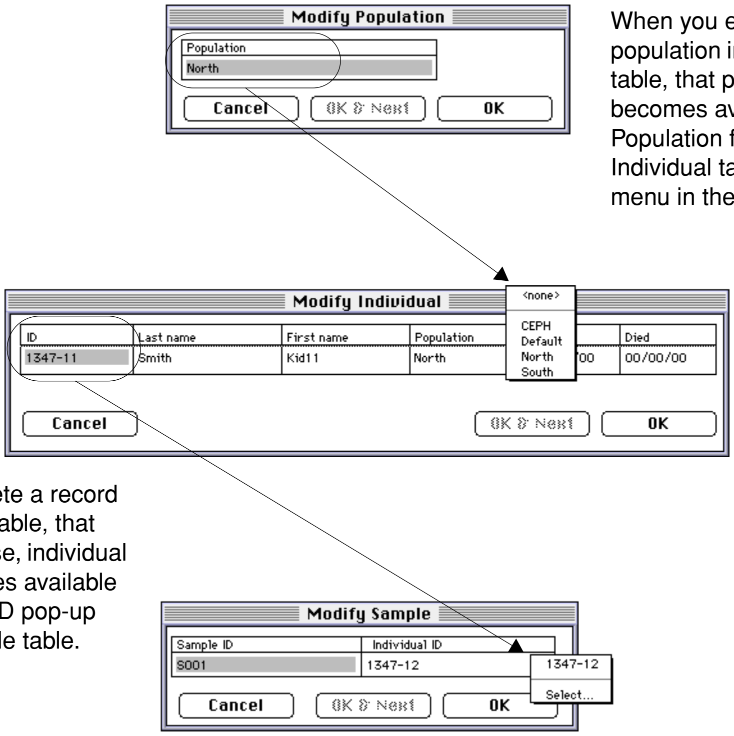
Introduction Setting up the database simply means filling selected fields in each table in a specified order. Start with a new, empty database before entering your own data.

Note The database included in this document is for instructional use only. You should not use this database to store your own data.

Setting up the database requires understanding both how to set up tables and the order in which they must be set up.

Database Information Flow

Tables must be set up in an order dictated by the way information flows from one table to the next. For example, the Individual ID field (a pop-up menu) in the Sample table only accepts information that has already been entered in the Population and Individual table (Figure 4-1).



Population: North

ID: 1347-11
Last name: Smith
First name: Kid11
Population: North
00
Died: 00/00/00

Sample ID: S001
Individual ID: 1347-12

When you enter a population in the Population table, that population becomes available to the Population field in the Individual table (pop-up menu in the figure below).

Once you complete a record in the Individual table, that record (in this case, individual 1347-12) becomes available in the Individual ID pop-up field of the Sample table.

Figure 4-1 Information flow

Set up tables in the order in which data flows to them (Table 4-1).

Table 4-1 Tables and contents

Table	Contents
Traits	Defines traits being studied.
Populations	Lists populations being studied.
Maps	Lists names of maps. (Map locations are recorded in the Markers table.)
Alternate Marker Name Sets	Lists sets of marker-name synonyms. (Alternate marker names are recorded in the Markers table.)
Markers	Defines the markers being studied. You can specify map locations (for a particular Map) and alternate names (for a particular Alternate Marker Name Set).
Alleles	Defines the alleles being studied. You can specify allele frequencies (for a particular Population).
Panels	Lists sets of Genotyper categories.
Categories	Lists categories and associates them with a panel and marker.
Individuals	Defines individuals in study.
Samples	Matches Sample IDs to individuals. (There can be multiple DNA samples for each individual.)
Pedigree Structures	Lists names of hypotheses for Pedigree relationships.
Pedigrees	Lists Pedigree information for an individual that relates to a Pedigree hypothesis (Pedigree Structure).
Liability Classes	Defines liability classes for traits.
Phenotypes	Lists Trait information for individuals.
Penetrances	Lists Penetrance data for trait, liability class, and genotypes.
QTL Variances	Lists a Variance for two quantitative traits or a variance for a single quantitative trait.
External Sources	Lists sources (other than the Genotyper software) of called genotype data.
Called Genotypes	Lists final genotype results from individuals. (May be derived from the Genotyper software or from external sources.)

Table 4-1 Tables and contents *(continued)*

Table	Contents
Category Members	Stores category definitions from the Genotyper software.
Genotypes	Stores genotype results from the Genotyper software.
Genotype Summaries	Same as Genotype table, except data is displayed in a compact format.
Gels	Names of gel files for genotype records.

Designing Your Database

The GenBase Database application is a flexible application that allows you to construct a database to best suit your needs. It is not necessary to use all the tables that are available in GenBase. You may pick and choose the ones that best support your requirements.

When setting up your database, select tables based on how you want to import records. For example, setting up a database for use with the Genotyper software will be different from setting up a database exclusively for use with the GenoPedigree software.

Setting Up a Database For Genotyper Data

To use the GenBase Database application with Genotyper data, set up the following tables:

- Samples
- Populations
- Individuals
- Categories
- Traits
- Phenotypes

When you import data from the Genotyper software, the following tables are filled out automatically:

- Genotypes
- Category Members
- Gels

Setting Up a Database for GenoPedigree Records

If you are using the GenBase Database application purely for phenotype data, fill out the following tables:

- Population
- Pedigree Structure
- Phenotypes
- Traits
- Liability Classes
- QTL Variances

The GenoPedigree software can exchange (write data to or read data from) information with the following tables:

- Phenotypes
- Pedigrees
- Traits
- Markers
- Genotypes

When importing data into the GenBase Database application, the GenoPedigree software may also create related records in the following tables:

- Individuals
- QTL Variances
- Liability Classes
- Alleles
- Penetrances

continued on next page

What Database Information Is Used For

Each table in the GenBase Database application has a specific function, and you might not need to use all of them every time you use GenBase. Before setting up your database, plan how to use the prepared data as described in Table 4-2.

Table 4-2 Database table usage

Table	**Suggested Initial Setup**	**Used by Genotyper**	**Used by GenoPedigree**
Traits	Yes		Yes
Populations	Yes		
Maps			Yes
Alternate Marker Name Sets			Yes
Markers	Yes		Yes
Alleles			Yes
Panels	Yes		
Categories	Yes	Yes	
Individuals	Yes		Yes
Samples	Yes	Yes	
Pedigree Structures		Yes	Yes
Pedigrees	Yes	Yes	Yes
Liability Classes			Yes
Phenotypes			Yes
Penetrances			Yes
QTL Variances			Yes
External Sources			
Called Genotypes			Yes
Category Members		Yes	
Genotypes		Yes	Yes
Genotype Summaries		Yes	Yes
Gels		Yes	

continued on next page

About GenBase Interaction With Genotyper and GenoPedigree

The GenBase Database application can share data with other components of the ABI PRISM Genotyping Software System. In all cases, the exchange of data is conducted through either the Genotyper software or the GenoPedigree software, not the GenBase Database application. GenBase must be open, and links must be established between the programs, but the actual commands to import or export data from the GenBase Database application are used outside this program.

Creating or Reading From Related Fields

When you import data from the Genotyper software, Genotyper can fill in records in the following tables:

- Genotyper
- Categories
- Gels

No other records are created. See Chapter 5, “Importing and Checking Data,” for more information on importing data from the Genotyper software.

When you import data from the GenoPedigree software, records relating to the imported table may be created. For example, importing Phenotypes from the GenoPedigree software may also create records in the Individual, Traits, QTL Variance, and Liability tables. Because these related fields are necessary parts of the Phenotypes table, GenoPedigree does not import records from the Phenotypes table unless these fields are filled out. For more information, see “Interfacing with the GenoPedigree Software” on page 5-34.

Entering Data

Introduction

In general, set up tables in the GenBase Database application prior to importing either Genotyper or GenoPedigree data. The two basic ways to enter data into GenBase tables are:

- ♦ *Manually*–Filling in dialog boxes for each table
- ♦ *Using the Import Command*–Importing data from text files

It is not necessary to complete all the tables before importing data from Genotyper software or the GenoPedigree software. When you do fill out the tables, follow the order described in this chapter.

Manually Entering Data

The advantage of using dialog boxes to enter data is that the GenBase Database application checks each entry against other tables to make sure the data matches related fields. For example, the GenBase Database application allows you to specify the population to which a person belongs. The dialog box for creating an individual record has a pop-up menu that lists only valid population choices for that field.

Manual Data Entry Conventions

To manually enter data in an open table, select New (⌘–N) from the Record menu. A dialog similar to the one in Figure 4-2 opens, and you can enter data into each available field through:

- ♦ *Text fields*–Accept either real numbers or characters
- ♦ *Pop-Up Menus*–Allow you to select from choices dictated by entries in related tables (see below).
- ♦ *Checkboxes*–Used for simple yes/no entries.

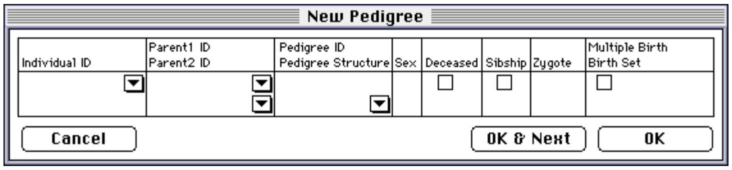


Figure 4-2 New dialog box (Pedigrees table)

Pop-Up Menus

Table fields that are restricted to information already entered in previous tables can be accessed through pop-up menus. These pop-up menus allow you to enter data quickly and eliminate the need to remember previous entries.

Using the Tab Key

When filling in tables, use the Tab key to jump from field to field (non-pop-up fields only).

Closing the New Record Dialog

When you finish entering data in a table, click either:

- *OK* – If you are finished entering data
- *OK & Next* – If you want to add another record
- *Cancel* – If you do not want to implement the change to the previous table

Note If you accidently click OK & Next, click Cancel. If you click OK, you will enter a blank record in your database.

Importing Text Files

Importing data from text files is much quicker than entering data manually. Importing text files may also create records outside the table into which you are importing data. For example, if you are importing data into the Individual table, the GenBase Database application checks the existing populations, and creates records in the Population table for each new population.

The GenBase Database application does not check the validity of the data. If you misspell the name of a population when importing an individual record, the GenBase Database application assumes that you have specified a new population and creates records accordingly.

Importing data from text files is most useful when you already have validated data from another database. Because imported text data can end up in tables other than the table into which you are importing data, and because there is no way to easily reverse imports, you must verify that data is in a format that matches the table’s, and that your data is correct.

For a complete discussion of importing text files, see “Importing External Source Data and Text Files” on page 5-12.

Setting Up GenBase Tables

Introduction

This section includes all the tables you can enter data into when setting up the database. These tables are listed in the order in which data flows from table to table. Entering data is easier if you fill the tables in this order.

You can also use this information to check tables prior to importing text data. By looking at Table 4-3, you can check which fields are shared with other tables in the database. These shared fields may receive data you import into the Traits table.

Creating Records in the Traits Table

A trait must be listed here if you want to keep track of trait information for individuals. You may track more than one trait in the database. Trait types include either Affection Status (an integer records whether or not a person has the trait) or Quantitative (a floating-point number records the value of the trait).

Table 4-3 Traits table

Contents	Fields	Shared Fields
Defines traits being studied	Name (15 characters) Type (boolean) Mutation Rate Male (real number) Mutation Rate Female (real number)	Name

continued on next page

Creating Records in the Populations Table

Depending on requirements, you may need to make distinctions between groups of individuals. The population table allows you to designate an individual's group affiliation.

If there are multiple populations, they can be selected from the Column Options command in the Special menu. Open the Allele table and use this command to view Allele data for different populations (Table 4-4).

Table 4-4 Populations table

Contents	Fields	Shared Fields
List of populations being studied	Population (20 characters)	Population

Creating New Map Names

Different maps may specify different locations for markers. To track more than one map, enter the names of the maps in the Maps table. To change the map displayed in the Marker table, select Column Options from the Special menu and choose a new map.

Table 4-5 Maps table

Contents	Fields	Shared Fields
List of names of maps (Map locations are recorded in the Markers table)	Map Names (15 characters)	Map Names

Creating Alternate Marker Name Sets

This table allows you to specify alternate names for Markers in your database. You may need to have Alt Marker Names if a downstream program has requirements about name length.

Table 4-6 Alternate Marker Name Sets table

Contents	Fields	Shared Fields
List of sets of marker-name synonyms. Alternate marker names are recorded in the Markers table.	Name Set (15 characters)	Name Set

continued on next page

Creating Records in the Markers Table

The markers that are used in your work are listed here inTable 4-7. Optionally, you can supply chromosome number and map location data for each marker. If you use Genotyper category names that are the same as the marker names, use the Call Genotypes command to fill in this table automatically.

If you have multiple Maps or Marker Name Sets, you can change the displayed map or name set by selecting Set Column Options and choosing alternate settings from the pop-up menus.

Table 4-7 Markers table

Contents	Fields	Shared Fields
Defines markers for which you can specify map locations and alternate names.	Name (15 characters) Chromosome (integer) Est. Num. Alleles (integer) Sex Linked (boolean) Mutation Rate Male/Female (real number) Default Map Location (15 characters) Alternate Name (15 characters)	Marker Name

continued on next page

Creating Records in the Alleles Table

The names of the alleles for each marker are listed in the Allele table (Table 4-8). If you are importing data from the Genotyper software, the allele name should be the same as the corresponding category member name in the Genotyper software.

Optionally, you can record the allele frequency for a particular population in this table. If you do not have to record frequencies, use the Call Genotypes command to fill in this table automatically.

Table 4-8 Allele table

Contents	Fields	Shared Fields
Defines alleles, and can be used to specify allele frequency to a particular population.	Marker/Trait Name (pop-up) Allele Name (seven characters) Population Frequency (integer)	Marker/Trait Name Allele Name

Creating Records in the Panels Table

If you want to define collections of Genotyper categories, first define panels in the Panels table (Table 4-9), and associate categories with a particular panel in the Categories table (Table 4-10 on page 4-14).

Note This is an optional field, but if you do not enter Panels, you must leave this field blank when entering Categories.

Table 4-9 Panels table

Contents	Fields	Shared Fields
List of sets of Genotyper categories	Panel name (20 characters)	Panel name

continued on next page

Creating Records in the Categories Table

The Categories table is intended to have one entry for each category. If the category is a member of a panel, the panel name is specified. The panel name is optional, however.

The Categories table also specifies the marker associated with the category. It is possible to have more than one Genotyper category for the same marker if, for example, different primer pairs are used at different times. If the category name is the same as the marker name, use the Call Genotypes command to fill in this table automatically.

Note Although markers and categories are commonly used interchangeably, the GenBase Database application maintains a distinction between the two. It is possible to have several categories assigned to a particular maker.

Table 4-10 Categories table

Contents	Fields	Shared Fields
Lists categories and associates them with a panel and marker	Category Name (31 characters) Panel Name (pop-up) Marker Name (pop-up)	Category Name

continued on next page

Creating Records in the Individuals Table

The Individual table identifies individuals by individual ID. You can, optionally, supply the name of the individual, along with dates of birth and death, and population (Table 4-11). The individual ID is used in all other tables of the GenBase Database application to identify an individual.

Table 4-11 Individual table

Contents	Fields	Shared Fields
Defines individuals in the study	ID (15 characters) Last name (20 characters) First name (20 characters) Population (pop-up) Born (date) Died (date)	ID

Creating Records in the Samples Table

This table is needed when there is the possibility of multiple samples for one individual. Even if there is only one sample per individual, it may be a good idea to distinguish the two in case a sample is misidentified. If the Sample ID and the Individual ID are distinct, this kind of error can be corrected by modifying a single record in this table instead of modifying multiple records throughout the database. If the individual and sample IDs are identical, use the Call Genotypes command to fill in this table automatically.

Table 4-12 Sample table

Contents	Fields	Shared Fields
Matches Sample IDs to individuals. There can be multiple DNA samples for an individual.	Sample ID (15 characters) Individual ID (15 characters) Entry Data (non-editable) Entry Time (non-editable)	Sample ID

continued on next page

Creating Records in the Pedigree Structure Table

You may want to keep track of different possible pedigrees for the same individuals. For example, you may not be sure of the exact relationships among family members and may have different theories, or you may need to duplicate individuals and break loops for certain analytical tasks. You may also need to disguise pedigrees for privacy reasons prior to publication. In the Pedigree Structures table, you provide names for these variations (Table 4-13).

Note See the *GenoPedigree User's Manual* for information on how to set pedigree IDs.

Table 4-13 Pedigree Structures table

Contents	Fields	Shared Fields
List of names of hypotheses for pedigree relationships	Name (31 characters)	Pedigree Name

continued on next page

Creating Records in the Pedigrees Table

The information in the Pedigrees table is typically provided by interface with the GenoPedigree software (Table 4-14). Instructions for linking and exchanging data with the GenoPedigree software are provided in Chapter 5. If you defined multiple structure names, you can have more than one record for each individual.

Table 4-14 Pedigrees table

Contents	Fields	Shared Fields
Pedigree information for an individual that relates to a pedigree structure	Individual ID (15 characters) Structure name (15 characters) Mother/Father (15 characters) Sex (boolean) Pedigree ID (31 characters)) Pedigree Structure (pop-up) Deceased (boolean) Zygote (integer) Birth Set (integer) Sibship (boolean) Multiple Birth (boolean)	Individual ID Structure name

Creating Records in the Liability Classes Table

This table matches traits with liability classes. This data is used by the GenBase Database application and the GenoPedigree software to create data files for linkage analysis programs.

Table 4-15 Liability Classes table

Contents	Field	Shared Fields
Defines liability classes for traits	Trait Name (pop-up) Liability Class (15 characters) QTL Type (pop-up)	Trait Name Liability Class

continued on next page

Creating Records in the Phenotypes Table

This table matches individuals with particular traits, which the GenoPedigree software can use to display inheritance patterns, and the GenBase Database application and the GenoPedigree software can use to create linkage-compatible data files (Table 4-16). The traits can be either Quantitative or Affection status types.

Table 4-16 Phenotype table

Contents	Fields	Shared Fields
Trait information for specific individuals	Individual ID (15 characters) Trait name (pop-up) Affected (pop-up) QTL Type (pop-up) QTL Value 1 and 2 (real number) Liability Class (pop-up) Exam Date (date)	Individual ID Trait name

Creating Records in the Penetrances Table

This table is used by the GenBase Database application and the GenoPedigree software to prepare input files for linkage analysis programs (Table 4-17).

Table 4-17 Penetrances table

Contents	Fields	Shared Fields
Penetrance data for trait, liability class, and genotypes	Trait name (pop-up) Liability Class (pop-up) Allele (1 and 2) (pop-up) Penetrance (real number) Type (2 characters) Variance (real number)	Trait name Liability Class Allele (1 and 2)

continued on next page

Creating Records in the QTL Variance Table

This table is used by the GenBase Database application and the GenoPedigree software to prepare input files for the linkage analysis program (Table 4-18).

Table 4-18 QTL Variances table

Contents	Fields	Shared Fields
Covariance for two quantitative traits or variance for a single quantitative trait	Trait (pop-up) Trait 1 (pop-up) Trait 2 (pop-up) Variance 1 (real number) Covariance (real number) Variance 2 (real number) Multiplier (real number)	Trait Trait 1, 2

Creating or Modifying User Lists

Overview Once you have set up a database, designate the people who can use the database and what tasks they are allowed to complete by setting up the GenBase User List.

The purpose of the User List table is to create an accountability trail. In large experiments with multiple users and high volumes of data, sorting out who did what and when is vital. The User table allows you to assign names and levels of access to all users of the database, and keep track of their review comments when reviewing Genotyper records. The edit user list dialog contains the following commands:

- *New*–Creates new name
- *Delete*–Deletes selected name
- *Modify*–Modifies selected name
- *Password Reset*–Clears password of selected name

Note System Admin access is required to create or modify user names. You cannot have duplicate user names on a single database.

Levels of Access GenBase user access is designed to let you control the number of people who have access to your data and the extent of their access. Levels of access should match the requirements of each individual, and the role they play during the experiment (Table 4-19). You may select access levels when you create a new name or modify an existing one.

Table 4-19 Access levels and privileges

Level Of Access	User Privileges
System Admin	At least one user must have this privilege. Allows the user to add new names to the user list and modify privileges.
Database Admin	This access allows users to delete files in the Genotypes, Category Members, and Gels tables.
Read Access	All active users should have Read Access enabled. If a user is no longer active, leave the user's name in the database but turn off this privilege. That way, the database can still keep track of the edits and checks made by the user.

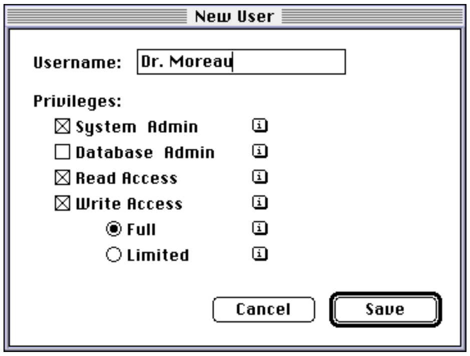
Table 4-19 Access levels and privileges *(continued)*

Level Of Access	User Privileges
Full Write Access	This allows the user to import, create, modify, and delete all records that can normally be edited. For example, this access does not allow a user to modify or delete genotype records.
Limited Write Access	This allows the user to create or edit sample records, check genotype records, set them to be current or obsolete, and import records from the Genotyper software or the GenoPedigree software.

How to Set Up User Lists

Access to the User List requires System Admin access. This is the default access when you first create the database.

To set up the User table:

Step	Action
1	Select Edit User List from the Admin menu.
2	Click New in the dialog box that appears.
3	The New User dialog box appears.  New User Username: Dr. Moreau Privileges: ☒ System Admin ☐ Database Admin ☒ Read Access ☒ Write Access ● Full ○ Limited
4	Enter Username and Privileges. **Note** You may click the “i” button to the right of the privilege to see an explanation of that particular privilege. You cannot have duplicate user names on a single database.
5	Click Save.

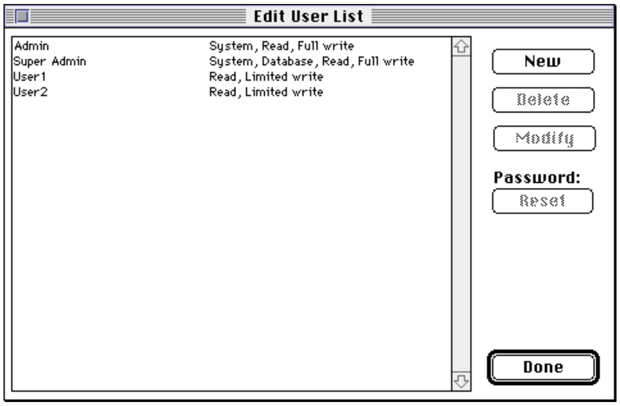
continued on next page

How to Change User Lists

During an experiment, it may be necessary to change or update the users or their access privileges.

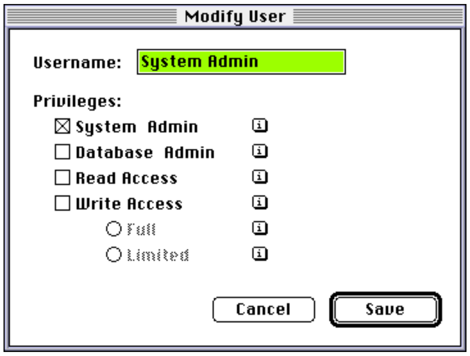
To modify the User list:

<table>
<tr><th>Step</th><th>Action</th></tr>
<tr><td>1</td><td>
Select Edit User List from the Admin menu.The following dialog box appears:



Admin	System, Read, Full write
Super Admin	System, Database, Read, Full write
User1	Read, Limited write
User2	Read, Limited write

</td></tr>
<tr><td>2</td><td>Select the User you want to change.</td></tr>
<tr><td>3</td><td>
Click Modify or double-click the name.The following dialog box appears:



Username: System Admin

Privileges:
☒ System Admin
☐ Database Admin
☐ Read Access
☐ Write Access
○ Full
○ Limited

</td></tr>
<tr><td>4</td><td>
Enter Username or change Privileges and click Save.

Note The “i” buttons to the right of the names provide information regarding each selection.
</td></tr>
</table>

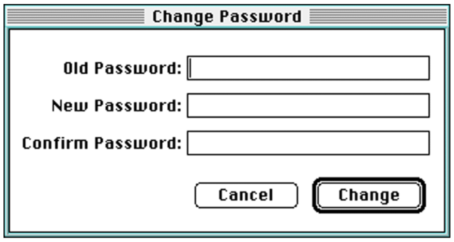
continued on next page

Assigning Passwords

After you have created a new user and assigned an access level, let users pick their own passwords or assign them.

Note If a user forgets his or her password, the system administrator can reset the password in the Edit User dialog box. Because of this feature, it is probably easier to let each user set a password.

To set user passwords:

Step	Action
1	Select Set User from the File menu.
2	Enter User name and password. If there isn't an existing password, simply leave the field blank.
3	Select Change Password from the Admin menu. The following dialog box appears: 
4	Enter the old password (leave blank if entering a password for the first time).
5	Enter the new password.
6	Confirm the password.
7	Click Change to change the password. **Note** If you enter a password incorrectly in one of the fields, the GenBase Database application will display an alert, and you must re-enter the password.

Additional Table Information

Additional Tables

Other tables in the database that are not required for set up are shown in Table 4-20 and Table 4-21.

Table 4-20 Results tables

Table	Description
Called Genotype	Genotype records that have been checked and approved for use in the experiment. These can be created by “Calling” Genotype records, or by importing data from an external source.
External Source	Identification for data brought in from sources other than the Genotyper software

The Genotyper tables contain data imported from the Genotyper software and cannot be edited.

Table 4-21 Genotyper tables

Table	Description
Genotype	Genotyper records that may be reviewed in the GenBase Database application, but must be exported to the Genotyper software for editing.
Category Members	Information relating to categories–Allele names and summaries of their definitions. Note that only the summaries are contained in this table. If you export this table as a text file or to the printer, only the summary information will be saved or printed.
Gels	Information from GeneScan regarding the Gel containing the raw data from which Genotyper records were constructed.

Backing Up Your Data

Overview

Whether you are an advanced or novice user, you can lose data because of software or hardware problems. To prevent data loss, keep a current backup at all times, and establish a regular backup routine. If you backup every day, the most you can lose is one day's work.

Setting a Routine

Assign one individual the responsibility for backing up data at a particular time of the day.

Note Be sure to post the steps included in the backup routine near the computer where the backup is performed.

GenBase File Size

Database files can grow quickly. Small GenBase files are generally larger than 1 MB. Your backup procedure depends on the size of your database files.

How to Back Up Data Files

Follow these or similar procedures to back up data files:

Step	Action
1	Close the GenBase Database application and locate the database file.
2	Create a folder entitled GenBase "date", where "date" is the date of the backup, on the destination drive.
3	Drag and drop file's icon on the destination folder's icon.

Importing and Checking Data

5

Chapter Overview

Introduction

Records exported from the Genotyper software into the GenBase Database application reside in the Genotype table, where you can review them prior to importing them into the Called Genotypes table. Records from other applications, such as the GenoPedigree software, can be imported directly into the Called Genotype table. However, they cannot be reviewed.

Text files imported into the GenBase Database application are placed directly into an open table. Because your text files may or may not be properly formatted for the GenBase tables, carefully check this data when you import it.

You can also establish a connection with the GenoPedigree software and import or export information directly from the Pedigrees, Called Genotypes, Phenotypes, Markers, and Traits tables.

Note These procedures require Read and Limited Write access.

continued on next page

In This Chapter

This chapter contains the following topics:

Importing Genotyper Category Members

Overview

Category members exported from the Genotyper software into the GenBase Database application contain definitions stored in the Category Member table. GenBase checks these definitions when creating called genotype records. You can still create called genotype records without these definitions using the "Create Undefined Records" feature of the Call Genotypes command (see "Looking up Related Records" on page 5-28).

You can also export the categories back to the Genotyper software for editing (see "Exporting Records to Genotyper" on page 5-22).

About Setting Up Categories

The Category Members table defines which categories correspond to which markers. You can use the same name for the category and marker, though one difference between the two in the GenBase Database application is that you can specify to which panel a marker belongs. See "Creating Records in the Panels Table" on page 4-13 and "Creating Records in the Categories Table" on page 4-14.

Note For more information on setting up categories, consult your Genotyper manual.

Setting up Database Categories

To set up and import categories:

Step	Action	See Page
1	Assign Panel Names.	4-13
2	Set up the Categories table.	4-14
3	Link the two programs.	5-4
4	Export categories from the Genotyper software into the GenBase Database application.	5-5

continued on next page

Setting up the Connection from Genotyper to GenBase

This procedure is only required before communication between the GenBase Database application and the Genotyper software is established.

Note Use this same procedure to connect to the GenoPedigree software. Simply substitute GenoPedigree for Genotyper in Step 2 and Step 3.

To set up communications between Genotyper and GenBase:

Step	Action
1	Ensure that both programs are open.
2	In the Genotyper software, click the GenBase linking button. If the programs were linked previously, the GenBase Database application becomes the active program. If not, the Genotyper software asks you to locate the GenBase Database application, and then opens GenBase as the active program.
3	In the GenBase main window, click the Genotyper linking button. If the programs are linked, the Genotyper software becomes the active program. If not, the GenBase Database application asks you to locate the Genotyper software, and then opens Genotyper as the active program.

continued on next page

Importing Category Members

This procedure is executed in the Genotyper software. The only requirement is that the two programs are open and linked.

To export categories from Genotyper into GenBase:

Step	Action
1	In the Genotyper software, mark the categories to be exported to the GenBase Database application and unmark any categories you do not want exported. **Note** Marked categories have a bullet (•) at the beginning of their entries in the category list.
2	Link the two programs by selecting Choose GenBase from the Link submenu of the File menu.
3	From the Category menu, select Export Marked Categories to Database. **Note** Ensure that only the Categories you mean to move are marked.

Importing Genotyper Records to GenBase Database Application

Overview

The only way to enter data in the Genotypes table is to export genotype records from the Genotyper software into the GenBase Database application. This task is completed through the Genotyper software, which must first be set up to communicate with the GenBase Database application. Data is sent from the Genotyper software to the GenBase Database application in a format specified by Genotyper's Set Up Table command.

You can also export the genotypes back to the Genotyper software for editing (see "Exporting Records to Genotyper" on page 5-22).

Importing Genotyper Records

To import genotyper records from the Genotyper software:

Step	Action	See Page
1	Link the Genotyper software with the GenBase Database application.	5-7
2	Check Genotyper table format.	5-7
3	Export Genotyper records to the GenBase Database application.	5-11

continued on next page

Setting up the Connection from Genotyper to GenBase

This procedure is only required before communication between the GenBase Database application and the Genotyper software is established.

Note Use this same procedure to connect to the GenoPedigree software. Simply substitute GenoPedigree for Genotyper in Step 2 and Step 3.

To set up communications between Genotyper and GenBase:

Step	Action
1	Ensure that both programs are open.
2	In the Genotyper software, click the GenBase linking button. If the programs were linked previously, the GenBase Database application becomes the active program. If not, the Genotyper software asks you to locate the GenBase Database application, and then opens GenBase as the active program.
3	In the GenBase main window, click the Genotyper linking button. If the programs were linked previously, the Genotyper software becomes the active program. If not, the GenBase Database application asks you to locate the Genotyper software, and then opens Genotyper as the active program.

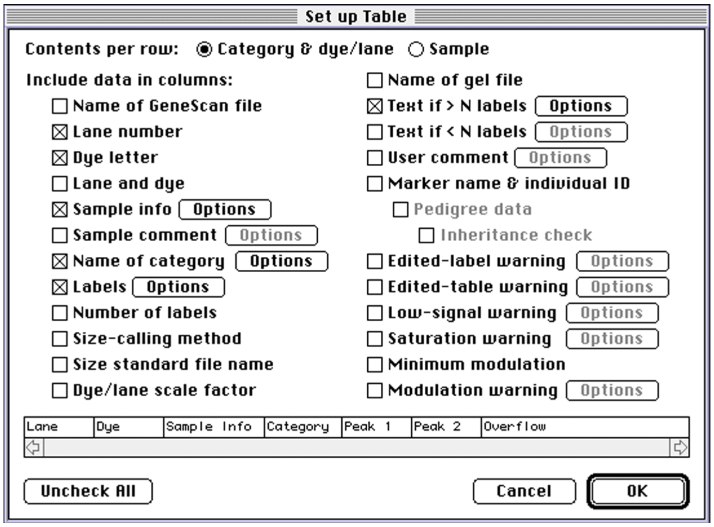
Checking Table Format

You may prepare records for import to the GenBase Database application by using the Set Up Table command in the Genotyper software. Proper set up ensures that the data you want to use in the GenBase Database application is transmitted correctly.

To set up tables in the Genotyper software:

Step	Action
1	Open the Genotyper software and, if necessary, establish a connection to the GenBase Database application (see “Setting up the Connection from Genotyper to GenBase” on page 5-7).
2	Select Clear Table from the Analysis menu.

To set up tables in the Genotyper software: *(continued)*

Step	Action
3	Select Set Up Table from the Table menu, and the following dialog box appears: 

Set up Table

Contents per row: ● Category & dye/lane ○ Sample

Include data in columns:

☐ Name of GeneScan file
☒ Lane number
☒ Dye letter
☐ Lane and dye
☒ Sample info
☐ Sample comment
☒ Name of category
☒ Labels
☐ Number of labels
☐ Size-calling method
☐ Size standard file name
☐ Dye/lane scale factor

☐ Name of gel file
☒ Text if > N labels
☐ Text if < N labels
☐ User comment
☐ Marker name & individual ID
  ☐ Pedigree data
    ☐ Inheritance check
☐ Edited-label warning
☐ Edited-table warning
☐ Low-signal warning
☐ Saturation warning
☐ Minimum modulation
☐ Modulation warning

To set up tables in the Genotyper software: *(continued)*

Step	Action
4	Select the following checkboxes: ♦ Sample Info (the information should include Sample ID) ♦ Name of category ♦ Labels The following fields are optional, and are sent only if checked: ♦ Size-calling method ♦ Text if >N labels ♦ Low-signal warning ♦ Saturation warning ♦ Minimum modulation ♦ Modulation warning The following fields are sent automatically and do not need to be checked: ♦ Name of GeneScan file ♦ Lane number ♦ Dye letter ♦ Name of Gel file ♦ Edited-label warning ♦ Edited-table warning **Note** Other fields are not available for export.

To set up tables in the Genotyper software: *(continued)*

<table>
<tr><th>Step</th><th>Action</th></tr>
<tr><td>5</td><td>
<p>Choose the kind of label data that you want to send to the GenBase Database application.</p>
<table>
<tr><th>If your labeled peaks have...</th><th>Then...</th></tr>
<tr><td>Size labels only</td><td>Choose one label per peak in the labels options of the Set Up Table command.</td></tr>
<tr><td>Sizes <i>and</i> allele names</td><td><p>Choose two labels per peak in the labels options of the Set Up Table command.</p><p>Note Category member names correspond to allele names.</p></td></tr>
</table>
<p>IMPORTANT Do not label peaks with allele names only. The GenBase Database application requires allele sizes to create called genotypes.</p>
<p>Note If you choose sizes and allele names, make sure that every labeled peak has two labels. For a description of how to set up your categories to insure that all peaks are labeled with either the correct allele name or with the name “Unknown” to indicate a new allele has been detected, consult Chapter 6 of the <i>Genotyper User’s Manual.</i></p>
</td></tr>
<tr><td>6</td><td>Click OK.</td></tr>
</table>

continued on next page

Sending Genotyper Records to GenBase

You can send records from the Genotyper software to the GenBase Database application, provided that both programs are open, communicating, and Genotyper is set up properly. Moving records is performed from inside the Genotyper software, and the following procedure assumes that you are familiar with that program. Consult Chapter 10 of the *Genotyper User's Manual* for further information.

When a genotype record is imported from the Genotyper software, existing genotype records with the same sample ID and category name are automatically marked obsolete, and the newly imported records are marked current. After the import operation is finished, the Genotype table shows only current records. To display both current and obsolete records, select Show All from the Record menu.

Note For more information about current and obsolete records, see "Managing Current and Obsolete Records" on page 5-24.

To import genotype records into GenBase:

Step	Action
1	Establish links with the GenBase Database application (see page 5-7).
2	Configure the Set Up Table dialog box (see page 5-7).
3	Select from Dye/lane list the dye/lanes that you want to include in the table.
4	Label peaks and filter labels.
5	Select Append to Table from the Table menu, and the selected records are displayed in the table.
6	Select Export to GenBase from the Table menu.

Importing External Source Data and Text Files

Introduction

The GenBase Database application can also store genotype data that was not created by the Genotyper software. Imported genotypes from the GenoPedigree software or text files must go into the Called Genotype table, not the Genotype table, and are referred to as External Source Data.

The GenBase Database application can also import tab-delimited files directly into tables other than the Called Genotypes table by using the Import command in the Records menu. Because tables share data files, if you import data into one table, shared fields are entered into other tables. Adding to the category table, for example, adds panels and markers to the database. Importing incorrectly formatted data (particularly large files) causes problems. Use caution when using these commands.

Note Appendix A provides a complete list of table fields.

IMPORTANT Unlike Genotyper and GenoPedigree data, there is no guarantee that data from other sources is correctly formatted for the GenBase Database application. See your database administrator prior to importing external source data.

Importing External Source Data

Choosing the Import command from the Called Genotype table allows you to create new Called Genotypes records without calling them from the Genotype table. The records must be in a format that matches the table. Unlike importing Genotyper or GenoPedigree data, data from external sources may not be formatted correctly. See your database administrator if you have any questions.

To import external source data:

Step	Action
1	Close all open tables.
2	Open the Called Genotype table.
3	Select Import from the Record menu.
4	Choose the file you want to import. **Note** The eighth column of the text file is the source name. If this field is not present, the current GenBase user name is used as the external source name.
5	Click Import.

To import external source data: *(continued)*

Step	Action
6	A dialog appears telling you which records have been created.
7	Check each Called Genotype record you've added.

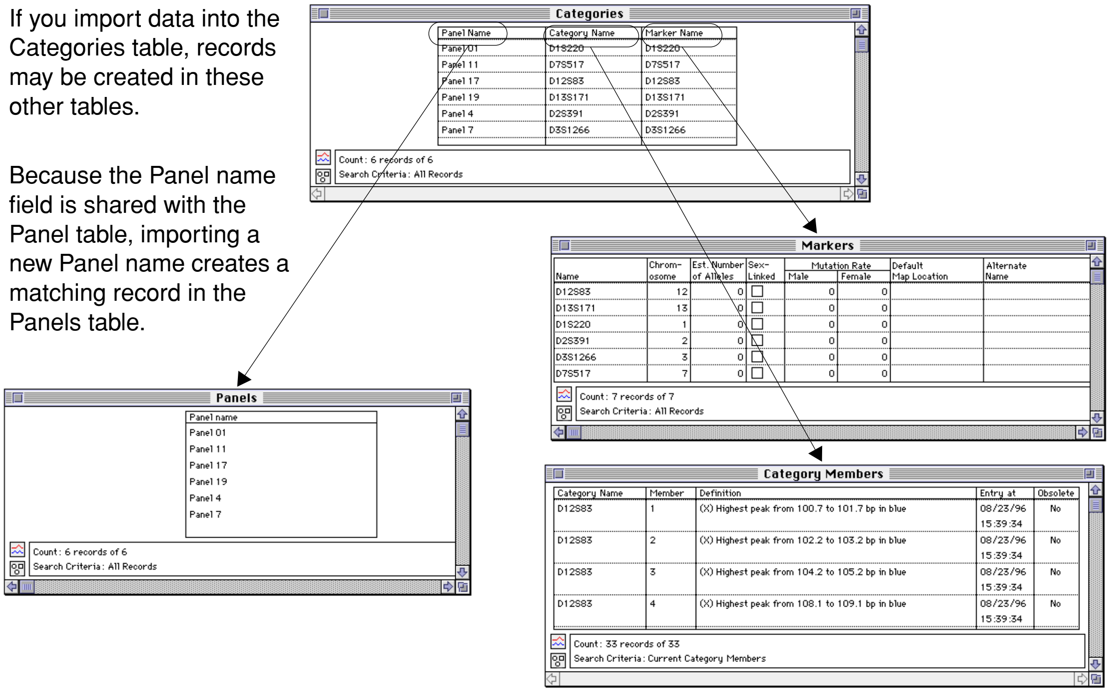
Verifying Data

You may also import tab-delimited text files directly into a selected table. As with importing external data into the Called Genotype table, you must verify that the data is in the correct format.

To verify GenBase table format:

Step	Action
1	Open the table in the GenBase Database application into which you want to import data.
2	Create several records by selecting New (⌘-N) from the Records menu.
3	Choose the Export command to create tab-delimited text files of these records.
4	Open this text file in the MicroSoft® Excel™ and check the format against the data you plan to import. **Note** Chapter 3 contains a list of the fields for each table, as well as the format of each field. Appendix A contains more detailed information relating to database tables. **IMPORTANT** Always back up your database data file before importing records.

It's also important that you verify that the data is correct. The GenBase Database application creates records in other tables which share fields with the open tables (Figure 5-1). If the data is incorrect, you may end up creating numerous false records.



If you import data into the Categories table, records may be created in these other tables.

Because the Panel name field is shared with the Panel table, importing a new Panel name creates a matching record in the Panels table.

Categories

Panel Name	Category Name	Marker Name
Panel 01	D1S220	D1S220
Panel 11	D7S517	D7S517
Panel 17	D12S83	D12S83
Panel 19	D13S171	D13S171
Panel 4	D2S391	D2S391
Panel 7	D3S1266	D3S1266

Count: 6 records of 6
Search Criteria: All Records

Panels

Panel name
Panel 01
Panel 11
Panel 17
Panel 19
Panel 4
Panel 7

Count: 6 records of 6
Search Criteria: All Records

Markers

Name	Chrom-osome	Est. Number of Alleles	Sex-Linked	Mutation Rate		Default Map Location	Alternate Name
				Male	Female		
D12S83	12	0	☐	0	0		
D13S171	13	0	☐	0	0		
D1S220	1	0	☐	0	0		
D2S391	2	0	☐	0	0		
D3S1266	3	0	☐	0	0		
D7S517	7	0	☐	0	0		

Count: 7 records of 7
Search Criteria: All Records

Category Members

Category Name	Member	Definition	Entry at	Obsolete
D12S83	1	(X) Highest peak from 100.7 to 101.7 bp in blue	08/23/96 15:39:34	No
D12S83	2	(X) Highest peak from 102.2 to 103.2 bp in blue	08/23/96 15:39:34	No
D12S83	3	(X) Highest peak from 104.2 to 105.2 bp in blue	08/23/96 15:39:34	No
D12S83	4	(X) Highest peak from 108.1 to 109.1 bp in blue	08/23/96 15:39:34	No

Count: 33 records of 33
Search Criteria: Current Category Members

Because the Category Name and Marker Name fields are shared with the Markers and Category Members tables, records are created in those tables as well. Unlike the Panels table, these tables have multiple fields for each record. These additional fields remain empty because there is no data to fill them.

Figure 5-1 Importing text files into shared fields

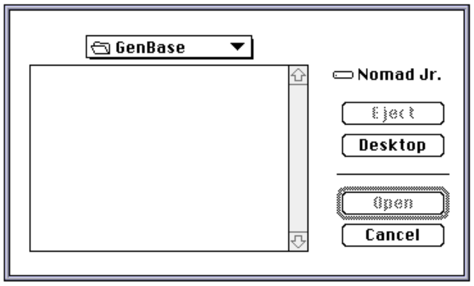
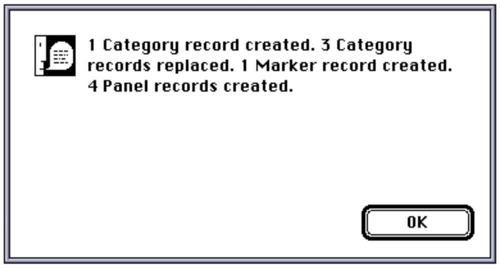
Obviously, the GenBase Database application does not create new records if there are existing records with identical information. However, misspelling an entry can create a new record based on that misspelling. If you are importing data into the Individual table and wish to assign them to the default population, the entry for the Population field should be “default.” If this entry is misspelled, for example “defalt,” the GenBase Database application creates a new record in the population table for the new population group “defalt.”

continued on next page

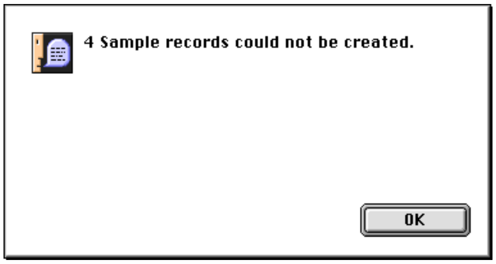
Importing Text Files into Tables

Simple mistakes can quickly become big problems using the Import command. For that reason, always back up your database before importing data.

To import text files into GenBase tables:

Step	Action
1	**IMPORTANT** Back up your database.
2	Open only the table into which you want to import data. **Note** All other tables must be closed.
3	Select Import from the Record menu. The following dialog box appears: 
4	Select the document you want to bring into your database, and click import.
5	You are advised how many fields in the table were filled. 1 Category record created. 3 Category records replaced. 1 Marker record created. 4 Panel records created.

To import text files into GenBase tables: *(continued)*

Step	Action
6	Check the data you imported, both in the open table, and in the related tables containing imported data. For example, if you imported data into the Category table, you also bring data into the Panels and Markers tables. **IMPORTANT** Check each field in all related tables to make sure data was imported correctly. If the data was in the wrong format (for example, the table fields require integers and you imported characters) you may see the following dialog.  4 Sample records could not be created. Re-verify your data and start again at step 3.

Checking Imported Genotype Records

Overview

After importing data, you or any database user with Limited Write access may review the individual genotype records for errors. User comments are attached to the record by name and become part of the database, allowing you to sort selected records by reviewer recommendations.

Data-Checking Options

It is important to keep in mind that checking data in the GenBase Database application, while convenient, is not absolutely necessary. You may review or not review records as you choose, based on your knowledge of the accuracy of the imported records.

Blind Review of Records

The GenBase Database application allows you to conduct a review of genotype records without being influenced by previous reviewers. Their comments, while still stored by the database, are hidden from the person conducting the review.

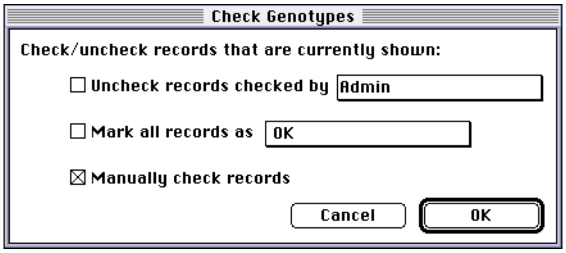
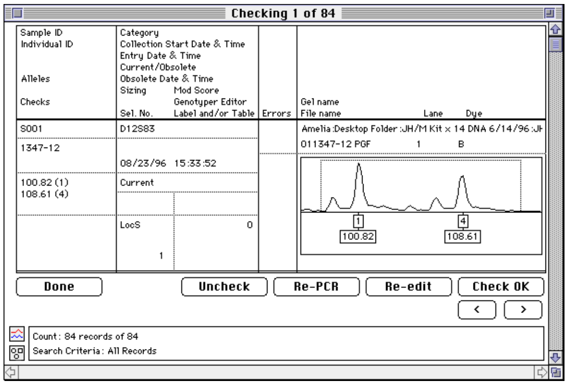
Reviewing Records

Whether you choose to have multiple users checking all imported records or to have one user spot-check records, the basic procedure is the same.

To review genotype records:

Step	Action
1	Show genotype records by selecting Genotypes from the Table menu.
2	Select the genotype records to be reviewed. **Note** Use the Find Genotypes command from the Special menu or select records manually by highlighting them and selecting the Show Selected command from the Record menu.

To review genotype records: *(continued)*

<table>
<tr><th>Step</th><th>Action</th></tr>
<tr><td>3</td><td>Select Check Genotype records from the User menu, and the following dialog box appears:

Check Genotypes
Check/uncheck records that are currently shown:
☐ Uncheck records checked by Admin
☐ Mark all records as OK
☒ Manually check records
</td></tr>
<tr><td>4</td><td>To clear records already reviewed, check the first box, and select your name.
To mark all records shown, uncheck “Manually check records,” check “Mark all records as,” and select the status to be applied to the records from the pop-up menu.</td></tr>
<tr><td>5</td><td>The Genotype Record window appears with the review selection buttons available.

Checking 1 of 84
Sample ID: S001
Individual ID: 1347-12
Alleles: 100.82 (1)
108.61 (4)
Category: D12S83
Entry Date & Time: 08/23/96 15:33:52
Current/Obsolete: Current
Sizing: LocS
Mod Score: 0
Sel. No.: 1
Gel name: Amelia :Desktop Folder :JH/M Kit x 14 DNA 6/14/96 :JH
File name: 011347-12 PGF Lane: 1 Dye: B
Count : 84 records of 84
Search Criteria : All Records

The review buttons are:
♦ *Done* – Exits review function
♦ *Uncheck* – Undoes your review recommendations
♦ *Re-PCR* – Sample must be PCR-amplified again
♦ *Re-edit* – Record is to be exported to the Genotyper software for editing
♦ *Check OK* – Record is suitable for the database</td></tr>
</table>

To review genotype records: *(continued)*

Step	Action
6	For each record, after review, select either Re-PCR, Re-Edit, or Check OK. Your name and recommendation appear in the genotype records. **Note** If a Genotyper record was imported with an edited table or label alert, that alert is displayed in red on the Genotyper record. You can move among the records by clicking the arrow buttons.
7	Click Done when you have completed your review.

Reviewing the Genotype Table

Overview

After Genotyper records are imported and checked by reviewers, you may search the database using the reviewer's recommendations, and either send the records back for review or move them to the Called Genotype table. Both categories and genotype records can be sent back to the Genotyper software for editing.

Note Reviewing records is ultimately your choice. You may or may not review records based on your knowledge of the accuracy of the records.

Review Comments and Error Messages

Review comments are displayed in the Genotype table. The display includes the reviewers names, and their evaluations of the record. The options available to a reviewer are

- Re-edit
- Re-PCR
- Check OK

Warning messages are information imported with the record from the Genotyper software and are displayed in genotype records. These messages are shown in Table 5-1.

Table 5-1 Warning messages

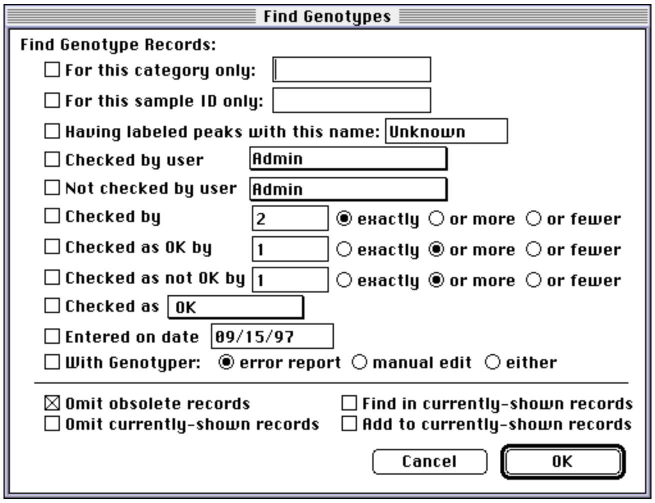
Error Message	Definition
O/F	Overflow (too many labeled peaks)
SAT	Saturated signal
LOW	Signal low
MOD	Modulation low
Table	Record manually edited in the Genotyper software
Label	Record manually edited in the Genotyper software

continued on next page

Searching Genotype Records by Reviewers

The Find Genotype Records command in the Special menu allows you to conduct searches based on review comments and to quickly verify the reviewer's recommendations.

To search reviewer records:

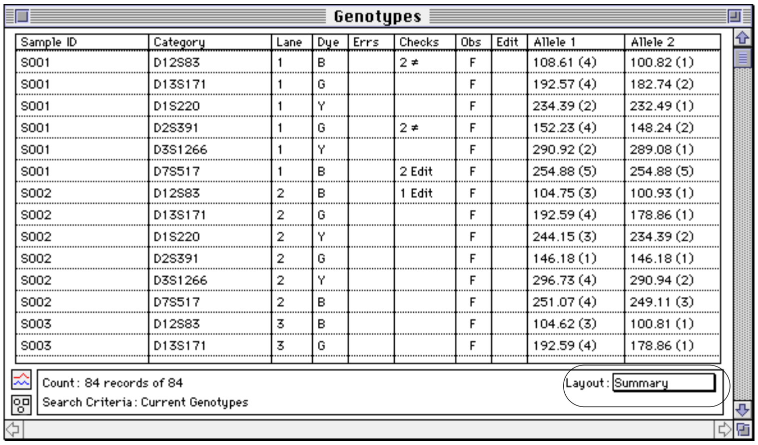
Step	Action
1	Open the Genotype table by selecting it in the Table menu.
2	Select Find Genotype Records from the Special menu. The following window appears:  Find Genotypes Find Genotype Records: ☐ For this category only: ☐ For this sample ID only: ☐ Having labeled peaks with this name: Unknown ☐ Checked by user Admin ☐ Not checked by user Admin ☐ Checked by 2 ● exactly ○ or more ○ or fewer ☐ Checked as OK by 1 ○ exactly ● or more ○ or fewer ☐ Checked as not OK by 1 ○ exactly ● or more ○ or fewer ☐ Checked as OK ☐ Entered on date 09/15/97 ☐ With Genotyper: ● error report ○ manual edit ○ either ☒ Omit obsolete records ☐ Find in currently-shown records ☐ Omit currently-shown records ☐ Add to currently-shown records Cancel OK
3	Use the check-boxes and pull down menus to configure your search. To find records reviewed by the user "Admin," check the Checked by user box, and Select Admin from the pop-up menu.
4	Click OK.
5	Located records are displayed as genotype records.

continued on next page

Using the Genotype Summary View

You can use the Genotype Summary view to quickly review all reviewer comments made to Genotype records. The Genotype Summary view Checks column shows the number of reviewers who checked a particular record, and if they agreed, it also shows their recommendation.

If multiple reviewers checked the record with different recommendations, the not-equals symbol (≠) is displayed (Figure 5-2).



Sample ID	Category	Lane	Dye	Errs	Checks	Obs	Edit	Allele 1	Allele 2
S001	D12S83	1	B		2 ≠	F		108.61 (4)	100.82 (1)
S001	D13S171	1	G			F		192.57 (4)	182.74 (2)
S001	D1S220	1	Y			F		234.39 (2)	232.49 (1)
S001	D2S391	1	G		2 ≠	F		152.23 (4)	148.24 (2)
S001	D3S1266	1	Y			F		290.92 (2)	289.08 (1)
S001	D7S517	1	B		2 Edit	F		254.88 (5)	254.88 (5)
S002	D12S83	2	B		1 Edit	F		104.75 (3)	100.93 (1)
S002	D13S171	2	G			F		192.59 (4)	178.86 (1)
S002	D1S220	2	Y			F		244.15 (3)	234.39 (2)
S002	D2S391	2	G			F		146.18 (1)	146.18 (1)
S002	D3S1266	2	Y			F		296.73 (4)	290.94 (2)
S002	D7S517	2	B			F		251.07 (4)	249.11 (3)
S003	D12S83	3	B			F		104.62 (3)	100.81 (1)
S003	D13S171	3	G			F		192.59 (4)	178.86 (1)

Count : 84 records of 84

Search Criteria : Current Genotypes

Layout : Summary

Figure 5-2 Genotype Summary view

Access the Summary view through the Layout pop-up menu in the lower right corner of the Genotype table (circled in the figure above).

Exporting Records to Genotyper

After verifying the reviewer comments, you may have to export records to the Genotyper software for editing. Both category and genotype records can be exported. In both cases, the procedure is conducted from inside the Genotyper software, where you can edit and resend records or categories to the GenBase Database application.

To export records to the Genotyper software:

Step	Action
1	Select the Genotypes to edit using the Find Genotypes dialog.
2	Select Find Related Records to display and select related records in the Category Members table.
3	In the Genotyper software, open a new, untitled document.

To export records to the Genotyper software: *(continued)*

Step	Action
4	Set up tables in the same format you used to import the records into the GenBase Database application (see “Importing Genotyper Records” on page 5-6).
5	Choose Append from the GenBase Database application from the Table menu, and click OK.
6	Choose Import Categories from Database from the Category menu.
7	Select row(s) in the table.
8	Select Re-Import Dye/Lane From File to bring in GeneScan data. **Note** This step only works if the GeneScan sample files have not been moved from their original folder.
9	Make changes to the Category definitions as necessary.
10	Review and edit labels or table cells as necessary.
11	Update table to apply edits.
12	Select Update to GenBase from Table to update the Genotypes table with your edits. If you have redefined or modified categories, select Export Modified Categories to Database in the Category menu.

Managing Current and Obsolete Records

Compare Genotypes Command

It is important to keep track of genotyper records when you have multiple copies of identical records and multiple reviewers. It is quite possible that the same genotype record was imported to the GenBase Database application several times.

Genotype information is considered current until a new record for the same sample and category is imported. If this happens, the GenBase Database application marks the older record “obsolete.” To resolve conflicts between current and obsolete records, use the Compare Genotypes feature.

Comparing Double-Blind Allele Calls

In Linkage Analysis, double-blind allele calls may be made by two different people, and a third person compares the two calls to see if they agree with each other.

The following table describes what actions to take when comparing calls.

If the calls...	Then
match,	the allele calls are considered final.
do not match.	the reviewer needs to answer the following questions: ♦ Which of the two calls are correct? ♦ If one of the calls is not satisfactory, should it be edited using the Genotyper software? ♦ If both calls are wrong, should both calls be edited using the Genotyper software, or should the PCR be repeated for either one or both?

continued on next page

Comparing Duplicate Genotypes

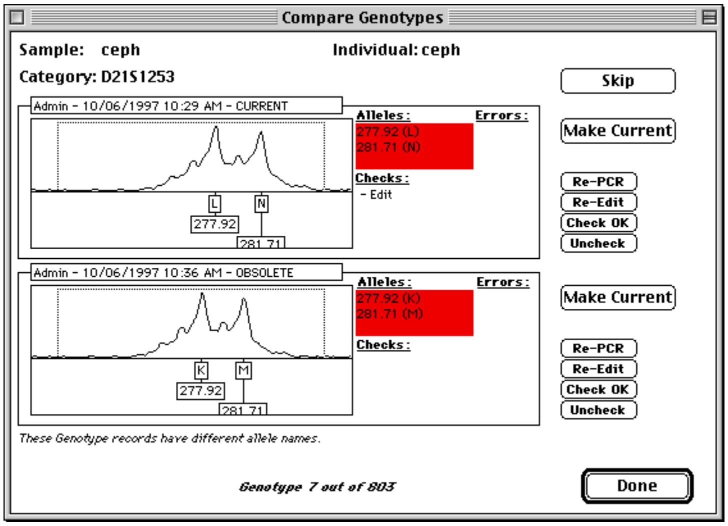
Comparisons also need to be made is in the laboratory where duplicate genotypes are generated. These comparisons are done by actually loading the duplicate samples in different lanes of a gel or on different gels on the ABI PRISM 373 or the ABI PRISM 377 or in different capillary runs on the ABI PRISM 310.

Once the reviewer makes a decision about the duplicate calls, those genotypes can be put into the Called Genotypes table that contains the final allele calls.

How Compare Genotypes Works

The Compare Genotypes feature compares allele calls (allele names only, not size) between duplicate Genotype records for the same Sample ID and Category.

Figure 5-3 shows the Compare Genotypes dialog box. To access this dialog box, select Compare Genotypes from the Special menu.



Compare Genotypes

Sample: ceph

Individual: ceph

Category: D21S1253

Admin - 10/06/1997 10:29 AM - CURRENT

Alleles:
277.92 (L)
281.71 (N)

Errors:

Checks:
- Edit

Admin - 10/06/1997 10:36 AM - OBSOLETE

Alleles:
277.92 (K)
281.71 (M)

Errors:

Checks:

These Genotype records have different allele names.

Genotype 7 out of 803

Figure 5-3 Compare Genotypes dialog

continued on next page

Using Compare Genotypes

The following table explains how to use Compare Genotypes.

<table>
<tr><th>Step</th><th>Action</th></tr>
<tr><td>1</td><td>Select Compare Genotypes from the Special menu to compare all Genotype records, or to speed up processing time, open the Genotypes table and select specific Genotypes to compare.
The GenBase Database application uses the following algorithm to select a set of Genotype records:
<table>
<tr><th>If the Genotypes table is...</th><th>Then the software uses...</th></tr>
<tr><td>not open,</td><td>all of the Genotype records in the database (long process).</td></tr>
<tr><td>open but no records are highlighted (selected),</td><td>all of the records currently displayed in the Genotypes window (moderate process).</td></tr>
<tr><td>open and some records are highlighted,</td><td>only those highlighted records (shortest process).</td></tr>
</table>
</td></tr>
<tr><td>2</td><td>From this set of Genotype records, the GenBase Database application determines all of the unique combinations of Sample ID and Category. These combinations are the distinct Genotypes the software analyzes.</td></tr>
<tr><td>3</td><td>Starting at the first Genotype record in the set, which is sorted alphabetically by Sample ID and Category, the GenBase Database application looks for the first Genotype record with differences.
The software uses the following algorithm to determine a difference:
<table>
<tr><th>If GenBase...</th><th>Then...</th></tr>
<tr><td>can find two Genotype records for the Genotype,</td><td>a current record and the most recently entered obsolete record are used.</td></tr>
<tr><td>can find no current records,</td><td>the two most recently entered obsolete records are used.</td></tr>
<tr><td>cannot find two Genotype records,</td><td>this Genotype is skipped.</td></tr>
</table>
</td></tr>
</table>

<table>
<tr><th>Step</th><th>Action</th></tr>
<tr><td>4</td><td>The GenBase Database application compares the allele peak names (not sizes) and the errors of the two Genotype records.
<table>
<tr><th>If both of these values are...</th><th>Then...</th></tr>
<tr><td>identical,</td><td>these records are considered to be similar, and this Genotype is skipped.</td></tr>
<tr><td>not identical,</td><td>the two Genotype records are displayed in the Compare Genotypes window.</td></tr>
</table></td></tr>
<tr><td>5</td><td>You can see the two Genotype records being analyzed for this Genotype and do some checking, such as mark either record for re pcr, re-edit, ok, or uncheck by clicking those buttons on the Compare Genotypes dialog box.
<table>
<tr><th>If you click...</th><th>Then...</th></tr>
<tr><td>the Skip button,</td><td>the Genotype software starts looking for the next Genotype with differences, using the same algorithm described in step 3.</td></tr>
<tr><td>either of the Make Current buttons next to each of the displayed Genotype records,</td><td>that record is marked as the current record, and the GenBase Database application looks for the next Genotype with differences using the same algorithm described in step 3.</td></tr>
</table></td></tr>
<tr><td>6</td><td>After you have processed all of the Genotypes with differences, the GenBase Database application displays a message that you are done and closes the window.</td></tr>
</table>

Creating the Called Genotype Table

The Called Genotype Table

The Called Genotype table is the result of data entry and is created by reading Genotype records and creating (or editing) a corresponding Called Genotype record. The table should only contain records that you have reviewed and feel are accurate and complete.

Looking up Related Records

When you transfer records from the Genotype table to the Called Genotype table, the GenBase Database application must look up certain information in the other tables of the database. A Genotype record refers to a particular sample ID, category and allele name(s), all of which must exist in their respective tables. Alleles in genotype records imported to the Called Genotypes table must have corresponding records in the Allele table.

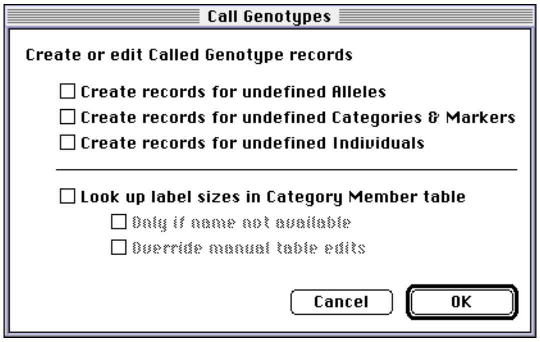
If the information cannot be found in the related tables, the GenBase Database application reports an error, and does not create or edit a Called Genotype record for that particular Genotype record. However, the checkboxes in the Call Genotypes dialog box have options that let you create missing records as described in the following section.

Note The Call Genotypes command only works with current genotype records.

The Call Genotypes Dialog Box

It is possible to send genotype records to the GenBase Database application from the Genotyper software without having first set up all the related tables (Sample, Marker, Allele, Individual). You may choose to have the GenBase Database application automatically create the related records that are necessary to create a Called Genotype record.

To create related records, use the checkboxes in the Call Genotypes dialog (see Figure 5-4).


Call Genotypes

Create or edit Called Genotype records

☐ Create records for undefined Alleles
☐ Create records for undefined Categories & Markers
☐ Create records for undefined Individuals

☐ Look up label sizes in Category Member table
  ☐ Only if name not available
  ☐ Override manual table edits

Figure 5-4 Call Genotypes dialog box

Creating Undefined Alleles

The "Create records for undefined Alleles" checkbox is selected by default, and creates records in the Allele table for all new or undefined alleles in the genotype records being called.

Creating Undefined Categories and Markers

The "Create records for undefined Categories and Markers" checkbox is selected by default, and creates records in the Categories table and the Markers table for all new or undefined categories or markers in the genotype records being called.

Creating Undefined Individuals

The "Create records for undefined Individuals" checkbox creates records in the Individuals table for all new or undefined individuals in the genotype records being called. If there are no records in the sample table that correspond to the individual, the GenBase Database application creates a Sample ID record in the Samples table, using the Individual ID as the Sample ID.

Note Using this command with undefined Individuals may create records in the Samples table that use the Individual ID in the Sample ID field. You can correct this by changing the Individual IDs in the Individual table. The IDs are automatically updated in the Individual ID field of the Sample table.

continued on next page

Looking Up Label Sizes in the Category Member Table

Check the “Look up label sizes in Category Member table” to have the GenBase Database application look up the allele peak size (bin) in the Category Member table to call the genotype. Select this checkbox if you know there are discrepancies in the allele names in the Genotype records and you want to ignore them and use the names in the Category Members table instead.

If Allele Names are Unavailable

If you also select the “Only if name not available” checkbox, the GenBase Database application only looks up the allele name when the peaks are unnamed. If you only imported peak sizes from the Genotyper software and the Category Member tables contains the allele names, the GenBase Database application assigns the allele names to the peaks using definitions in the Category Members table.

Note You can define all possible alleles (category members) for any marker in the Genotyper software, export the categories to the GenBase Database application and store them. Then in the Genotyper software, you can build a table that contains only peak sizes, export the table to the GenBase Database application, and call the alleles based on entries in the Category Members table.

IMPORTANT Allele names created in this manner appear only in the Called Genotypes table and are not appended to the peaks in the Genotypes table. If you import genotypes from the GenBase Database application to the Genotyper software for editing, the names are not sent with the genotype data.

Overriding Manual Edits

Use the “Override manual table edits” to have the GenBase Database application look up label sizes for Genotyper records with manual table edits. If the checkbox is not selected, the GenBase Database application ignores manual edits to the sizes/allele names and uses the original labels from the Genotyper software.

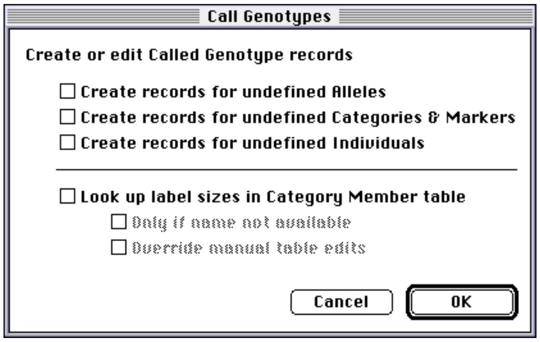
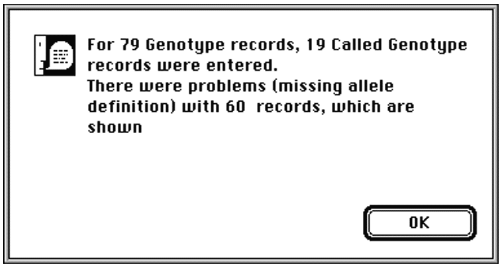
Use the checkboxes to create records in the tables related to the Called Genotypes table, or to have the GenBase Database application look up label sizes in the Category Member table.

continued on next page

Creating Called Genotype Records

If you are missing related records and do not select the checkboxes that assign the GenBase Database application to create these records, GenBase displays an error message.

To create Called Genotype records:

Step	Action
1	Select Genotypes from the Table menu. **Note** Ensure that the Genotype table is showing only the records you want to move to the Called Genotype table. Use the Find Genotypes command to abridge the table.
2	Select Call Genotypes from the Special menu, and the following dialog box appears:  Call Genotypes Create or edit Called Genotype records ☐ Create records for undefined Alleles ☐ Create records for undefined Categories & Markers ☐ Create records for undefined Individuals ☐ Look up label sizes in Category Member table ☐ Only if name not available ☐ Override manual table edits
3	Check the appropriate boxes to create Called Genotypes records. See the explanations starting with "The Call Genotypes Dialog Box" on page 5-28 in this section for a complete description of these options.
4	Click OK. The GenBase Database application searches for genotype records and displays the following dialog box:  For 79 Genotype records, 19 Called Genotype records were entered. There were problems (missing allele definition) with 60 records, which are shown
5	If there were errors, as there were in this example, you may correct them and redo the procedure.

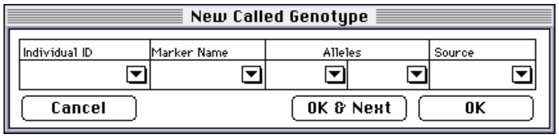
To create Called Genotype records: *(continued)*

Step	Action
6	You may view your new called genotypes in the Called Genotype table.

Adding or Changing Called Genotypes

You can also manually change or add records in the Called Genotype table. If all the genotype records have been added to the called genotype table, you cannot add new called genotypes. The GenBase Database application only allows you to enter fields that correspond to the remainder of the database. You cannot enter an Individual ID, Marker, or Allele Name that is not entered in the database.

To manually add new Called Genotypes:

Step	Action
1	Select Called Genotypes from the Table menu.
2	Select New (⌘-N) from the Records menu. The following dialog appears: 
3	Type or select from the pop-up menu an individual ID (must already be in the database). **Note** Each individual can have only one called genotype record for that marker.
4	Select Marker Name.
5	Select Alleles.
6	Select Source.
7	Click OK when complete or OK & Next to enter more records. If you tried to create a record for an individual who already has a called genotype record for that marker, an alert dialog appears, and the record is not entered in the called genotype table.

continued on next page

Importing Records to the Called Genotype Table

You can import called Genotypes in the same manner that you import text files to tables. Ensure that you verify the data and the format of that data before importing it to the Called Genotype table. Records in other tables are created when you import data.

The GenBase Database application assigns the external source field your user name and creates a record with your name in the External Source field if you import records with no entry in that field position.

Interfacing with the GenoPedigree Software

Introduction

The GenoPedigree software can use the GenBase Database application to construct pedigree diagrams. These diagrams can be printed or exported to LINKAGE, GAS, or other linkage analysis and mapping programs. The GenoPedigree software accesses information in the GenBase Database application using the GenoPedigree interface.

The GenoPedigree software also creates pedigree and locus data and exports it to the GenBase Database application.

Creating Linkage and Gas Files

LINKAGE, GAS, and CRI-MAP files require different information, but the GenBase Database application supports information for creating files in all of these applications. LINKAGE and CRI-MAP files can be created directly from the GenBase Database application. Which fields you fill out in GenBase tables depends on the application for which you are constructing files.

Importing Tables to GenBase

Pedigrees imported from the GenoPedigree software must have Structure names assigned before they enter the database. The GenoPedigree software directly reads from and writes data to the Pedigree, Phenotype, Called Genotype, Marker, and Trait tables (Figure 5-5). GenoPedigree, in the process of writing data to these tables, may create records in tables with fields related to these tables in the same way as importing text files creates related records.

continued on next page

GenBase Locus and Trait Fields

Markers and Trait data in the GenBase Database application is for linkage applications. Markers are encoded as “Named Alleles” type data, and traits are encoded as “Affection Statues” or Quantitative Trait” data for linkage applications.

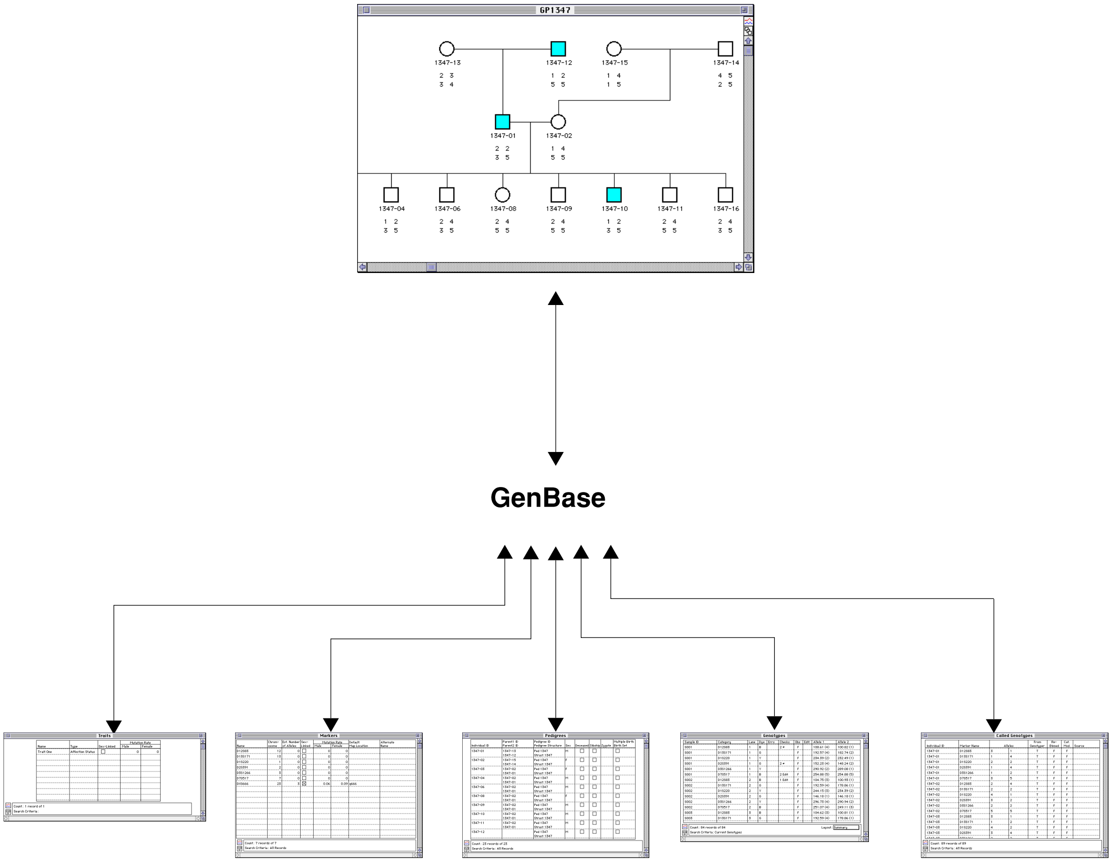


Figure 5-5 GenBase Database application and GenoPedigree software interaction

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Linking GenoPedigree with GenBase

All commands for interactions between the GenoPedigree software and GenBase Database application are executed in GenoPedigree. These commands are located in GenoPedigree's GenBase menu, and are not available unless the two programs are linked.

To link GenoPedigree and GenBase:

Step	Action
1	Ensure that both programs are open.
2	In the GenoPedigree software, click the GenBase linking button. If the programs are linked, the GenBase Database application becomes the active program. If not, the GenoPedigree software asks you to locate the GenBase Database application, and opens GenBase as the active program.
3	In the GenBase main window, click the GenoPedigree linking button. If the programs are linked, the GenoPedigree software becomes the active program. If not, the GenBase Database application asks you to locate the GenoPedigree software, and opens GenoPedigree as the active program.

Tables Used By GenoPedigree

The GenoPedigree software can read and write data from the following GenBase tables:

- Pedigrees
- Phenotypes
- Traits
- Markers
- Called Genotypes

When writing data to GenBase tables, the GenoPedigree software may also create records in the following tables:

- Pedigree Structure
- Individual

- Allele
- QT Variance
- Penetrances
- Liability Classes

The GenoPedigree software can also read but not write to the following tables:

- Maps
- Populations

Which tables and fields you fill out in the GenBase Database application depends on what data you want to store in GenBase and if you want to construct LINKAGE or GAS files using the GenoPedigree software.

Creating LINKAGE or GAS Files in GenoPedigree

Table 5-2 lists the tables and fields required by the GenoPedigree software to produce LINKAGE or GAS files. Because the requirements of each file format are different, fill out database information only as necessary for the particular type of file to create.

Table 5-2 Tables and fields used to construct LINKAGE or GAS files

Table	Field	Used By LINKAGE	Used By GAS
Pedigrees	Individual ID	Yes	Yes
	Parent IDs	Yes	Yes
	Pedigree ID Pedigree Structure	No No	No No
	Sex	Yes	Yes
	Deceased	No	No
	Sibship	No	No
	Multiple Birth Set	No	No
Penetrances	Trait Name	Yes	Yes
	Liability Class	Yes	Yes
	Alleles (two fields)	Yes	Yes
	Penetrance	Yes	Yes
	Type	No	Yes
	Variance	No	Yes

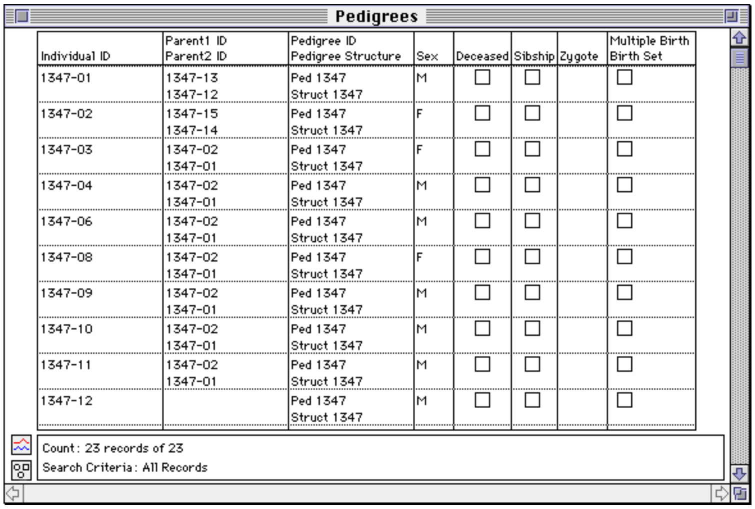
Table 5-2 Tables and fields used to construct LINKAGE or GAS

Table	Field	Used By LINKAGE	Used By GAS
QTL Variances	Trait	Yes	No
	Trait 1 (liability class)	Yes	No
	Trait 2 (liability class)	Yes	No
	Variance 1	Yes	No
	Covariance	Yes	No
	Variance 2	Yes	No
	Multiplier	Yes	No
Phenotypes	Individual ID	Yes	Yes
	Trait Name	Yes	Yes
	Affected	Yes	Yes
	QTL Type	Yes	Yes
	QTL Value 1	Yes	Yes
	QTL Value 2	Yes	No
	Liability Class	No	Yes
Traits	Name	Yes	Yes
	Type	Yes	Yes
	Sex-Linked	Yes	Yes
	Mutation Rate	Yes	No
Liability Classes	Trait Name	Yes	Yes
	Liability Class	Yes	Yes
	QTL Type	Yes	Yes

continued on next page

Importing Pedigrees

The GenBase Database application can use multiple pedigree structures imported from the GenoPedigree software. This data is stored in the Pedigree table. The names of alternate Pedigree structures are stored in the Pedigree Structure table (Figure 5-6 and Table 5-3)



Individual ID	Parent1 ID Parent2 ID	Pedigree ID Pedigree Structure	Sex	Deceased	Sibship	Zygote	Multiple Birth Birth Set
1347-01	1347-13 1347-12	Ped 1347 Struct 1347	M	☐	☐		☐
1347-02	1347-15 1347-14	Ped 1347 Struct 1347	F	☐	☐		☐
1347-03	1347-02 1347-01	Ped 1347 Struct 1347	F	☐	☐		☐
1347-04	1347-02 1347-01	Ped 1347 Struct 1347	M	☐	☐		☐
1347-06	1347-02 1347-01	Ped 1347 Struct 1347	M	☐	☐		☐
1347-08	1347-02 1347-01	Ped 1347 Struct 1347	F	☐	☐		☐
1347-09	1347-02 1347-01	Ped 1347 Struct 1347	M	☐	☐		☐
1347-10	1347-02 1347-01	Ped 1347 Struct 1347	M	☐	☐		☐
1347-11	1347-02 1347-01	Ped 1347 Struct 1347	M	☐	☐		☐
1347-12		Ped 1347 Struct 1347	M	☐	☐		☐

Count: 23 records of 23

Search Criteria: All Records

Figure 5-6 Pedigree table

Table 5-3 Pedigree table fields

Field	Description
Individual ID	As defined in Individual table.
Mother/Father	Parents as assigned by the GenoPedigree software.
Pedigree ID	Particular Family Pedigree as defined by the GenoPedigree software.
Pedigree Structure	As defined in the Pedigree Structure table. **Note** "Structure name" refers to a theory covering all individuals in the database, while "Pedigree ID" might only refer to particular families. See "Structure Names" on page 5-40.
Sex	As defined in Individual table.
Deceased	Whether or not the individual is deceased.

Field	Description
Sibship	A checkmark indicates place-holder parents (for example, parents who are not real individuals) for a sibship that can be diagrammed in the GenoPedigree software with no visible parents.
Zygote	As defined by the GenoPedigree software.
Multiple Birth Birthset	Whether or not the individual was part of a multiple birth.

Note Fields defined by the GenoPedigree software can be manually edited or entered through the Modify Record Command in the Report menu.

When you import pedigree information from the GenoPedigree software, each pedigree structure is identified in the GenBase Database application by a unique Structure Name. These names are stored in the Pedigree Structure table.

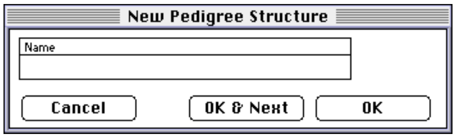
Structure Names

Each Structure Name represents a theory of the overall lineage structure. It's possible to have several pedigree structures for each database.

Note The "Pedigree ID" refers to specific lineage diagrams, not to an overall structure theory, and is assigned in the GenoPedigree software. There is no need to modify the Pedigree ID from inside the GenBase Database application.

Normally, Structure Names are assigned in the GenoPedigree software when you use the Export Pedigree command, and you can add or modify structure names in the GenBase Database application.

To create new pedigree structures:

Step	Action
1	Select Pedigree Structures from the Table menu.
2	Select New from the Record menu, and the following window appears: 
3	Enter ID number and description.

Step	Action
4	Click OK if you are finished, or OK & Next if you have more structures to create.

Writing to the Pedigrees Table

Transferring data to or from the GenoPedigree software is done from inside the GenoPedigree software. Consult the *GenoPedigree User's Guide* for specific directions.

After conducting this procedure, check the Pedigree table in the GenBase Database application to verify that pedigree data successfully imported.

Related Tables

Importing a pedigree diagram into the GenBase Database application also imports the associated individuals into the Individual table.

Importing or Exporting Data From Other GenBase Tables

The GenoPedigree software can read or write data directly to the following GenBase tables in addition to Pedigrees:

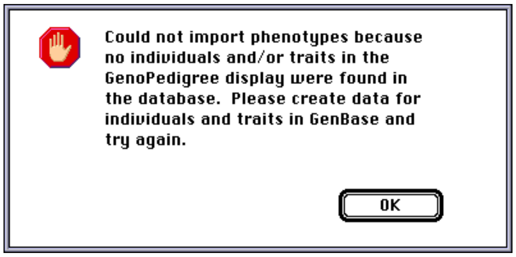
- Phenotypes
- Traits
- Markers
- Called Genotypes

Importing or Exporting to the Phenotypes Table

When the GenoPedigree software writes data into the Phenotypes table, records may also be created in the following tables:

- Individuals
- Traits
- Liability Classes

If you import the Phenotypes table from the GenBase Database application into the GenoPedigree software, an error message may appear if related records do not exist in the Individuals and Traits tables (Figure 5-7).



Could not import phenotypes because no individuals and/or traits in the GenoPedigree display were found in the database. Please create data for individuals and traits in GenBase and try again.

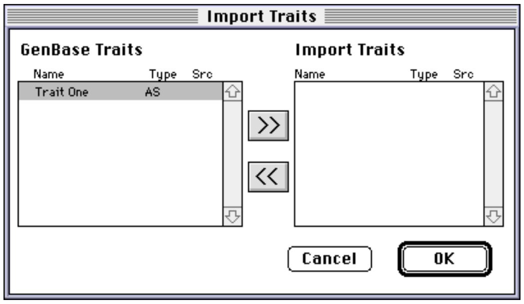
Figure 5-7 GenoPedigree error message

Importing or Exporting Data to the Traits Table

When you write data from the GenoPedigree software to GenBase's Traits table, you may also create related records in the following tables:

- Alleles
- Penetrances
- Liability Classes
- QTL Variances

It is not necessary to have data in records related to the Traits table for the GenoPedigree software to read data from this table. When you do, the GenoPedigree software uses the following dialog to let you choose the traits to import:



Import Traits

GenBase Traits

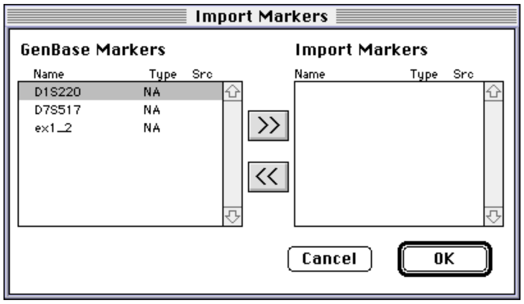
Name	Type	Src
Trait One	AS	

Import Traits

Name	Type	Src

Importing or Exporting Data From the Markers Table

Importing GenoPedigree data into the GenBase Markers table may also create records in the Alleles table. The GenoPedigree software uses the following dialog to let you choose the markers to import:



Name	Type	Src
D1S220	NA	
D7S517	NA	
ex1_2	NA	

Importing or Exporting Data From the Called Genotypes Table

When you import Genotype data from the GenBase Database application into the GenoPedigree software, you must have all related records in the Individual, Markers, and Alleles tables. If you do not, the GenoPedigree software displays an error (Figure 5-8).

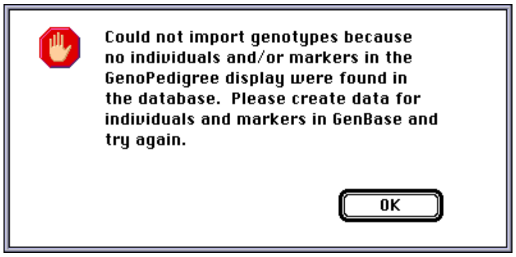

Could not import genotypes because no individuals and/or markers in the GenoPedigree display were found in the database. Please create data for individuals and markers in GenBase and try again.

Figure 5-8 Import error

Using the Database

6

Chapter Overview

Introduction This chapter discusses the following GenBase Database application tools:

- *Queries*–Commands that allow you to search individual database tables based on specific criteria. You can also query the entire database for records related to a selected record. The GenBase Database application includes query dialogs specifically designed for the Genotype and Called Genotypes tables.
- *Reports*–In addition to printing the contents of individual tables, the GenBase Database application provides a tool for building custom reports that can be printed or saved as text files.
- *Exports*–Create text files from tables.

Note Advanced search and sort functions are located in Appendix A.

In this Chapter This chapter contains the following topics:

Using the Find Command

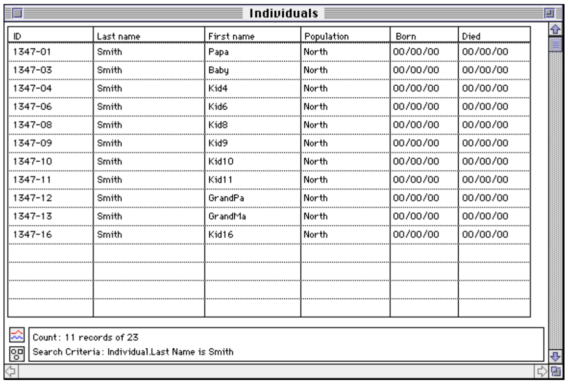
Introduction The Find command is a query that allows you to locate records using criteria relevant to the active table. The dialog contains pop-up menus from which you can select a single field and how that field compares to a particular value.

Using the Find Command Before you begin the search, use the radio buttons to select whether to clear the list before a search, to search within an already refined list, or add the results of your search to an already created list.

To use the Find command:

Step	Action
1	Open the table in which you want to find records. **Note** You can only search one table at a time.
2	Select Find (⌘-F) from the Record menu. The Find dialog box appears.  Find Individuals where: Individual Last Name is Smith List: ● Create New ○ Search Within ○ Append
3	Select the primary field to be searched from the first pop-up menu. **Note** Only fields available to the open table can be searched.
4	Select the secondary field to be searched from the second pop-up menu. **Note** These are fields that are available in the table you selected from the first pop-up menu.
5	Select the search operator. The values in this pop-up menu vary depending on the type of field you selected in the second pop-up menu.
6	Enter the value for which you want to search.

To use the Find command: *(continued)*

<table>
<tr><th>Step</th><th>Action</th></tr>
<tr><td>7</td><td>Using the radio buttons, select whether to
♦ <i>Clear list, then search</i> – Searches the entire table
♦ <i>Search within list</i> – Searches within the current list of records
♦ <i>Append to list</i> – Searches the entire table, then appends the records to the current list of records</td></tr>
<tr><td>8</td><td>Click Search.</td></tr>
<tr><td>9</td><td>The records that match the search criteria are shown.
Note The search criteria used is listed at the bottom of the screen.


<table>
<tr><th>ID</th><th>Last name</th><th>First name</th><th>Population</th><th>Born</th><th>Died</th></tr>
<tr><td>1347-01</td><td>Smith</td><td>Papa</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-03</td><td>Smith</td><td>Baby</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-04</td><td>Smith</td><td>Kid4</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-06</td><td>Smith</td><td>Kid6</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-08</td><td>Smith</td><td>Kid8</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-09</td><td>Smith</td><td>Kid9</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-10</td><td>Smith</td><td>Kid10</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-11</td><td>Smith</td><td>Kid11</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-12</td><td>Smith</td><td>GrandPa</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-13</td><td>Smith</td><td>GrandMa</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
<tr><td>1347-16</td><td>Smith</td><td>Kid16</td><td>North</td><td>00/00/00</td><td>00/00/00</td></tr>
</table>
Count: 11 records of 23
Search Criteria: Individual.Last Name is Smith</td></tr>
<tr><td>10</td><td>The table remains in this configuration as long as the program is open. If you want to show all records, select Show All (⌘-J) from the Record menu.</td></tr>
</table>

Using the Sort Command

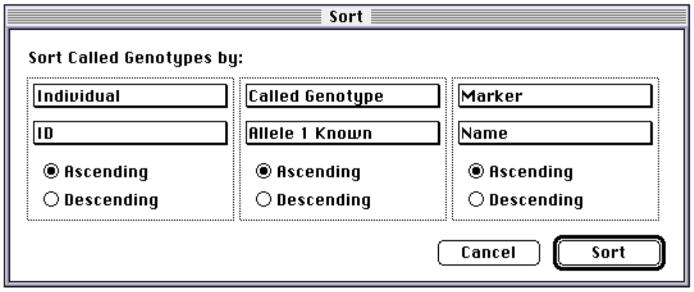
Overview

You can use the Sort command to arrange records in an open table based on up to three fields at one time.

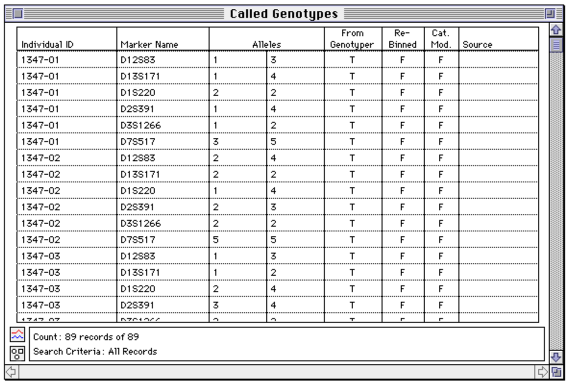
Using the Sort Function

You can have multiple tables open when you select the Sort command, but only the active table is effected.

To sort tables:

Step	Action
1	Select the table you want to sort from the Table menu.
2	Select Sort (⌘-S) from the Record menu. The following dialog box appears:  Sort Called Genotypes by: Individual ID ● Ascending ○ Descending Called Genotype Allele 1 Known ● Ascending ○ Descending Marker Name ● Ascending ○ Descending
3	Select sort criteria. Each set of criteria is comprised of a single field, which is selected through pop-up menus. All of the pop-up menus refer to fields in the open table, but use different field combinations.

To sort tables: *(continued)*

Step	Action
4	Select Sort. The sorted table is displayed. 

Called Genotypes

Individual ID	Marker Name	Alleles		From Genotyper	Re-Binned	Cat. Mod.	Source
1347-01	D12S83	1	3	T	F	F	
1347-01	D13S171	1	4	T	F	F	
1347-01	D1S220	2	2	T	F	F	
1347-01	D2S391	1	4	T	F	F	
1347-01	D3S1266	1	2	T	F	F	
1347-01	D7S517	3	5	T	F	F	
1347-02	D12S83	2	4	T	F	F	
1347-02	D13S171	2	2	T	F	F	
1347-02	D1S220	1	4	T	F	F	
1347-02	D2S391	2	3	T	F	F	
1347-02	D3S1266	2	2	T	F	F	
1347-02	D7S517	5	5	T	F	F	
1347-03	D12S83	1	3	T	F	F	
1347-03	D13S171	1	2	T	F	F	
1347-03	D1S220	2	4	T	F	F	
1347-03	D2S391	3	4	T	F	F	

Count : 89 records of 89
Search Criteria : All Records

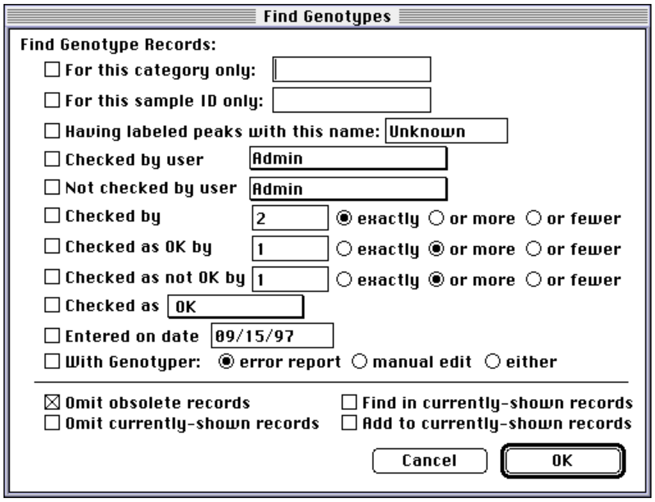
Searching Genotype Records

Overview

Search through genotype records with the Find Genotype Records command located in the Special menu. The window contains multiple search options selected through checkboxes. The Status field shows how many records were selected out of the total number of records.

Using the Find Genotypes Command

The Find Genotype Records command is in the Special menu. The checkboxes and pop-up menus allow you to find particular records using a wide variety of criteria.



Find Genotypes

Find Genotype Records:

☐ For this category only:

☐ For this sample ID only:

☐ Having labeled peaks with this name: Unknown

☐ Checked by user Admin

☐ Not checked by user Admin

☐ Checked by 2 ● exactly ○ or more ○ or fewer

☐ Checked as OK by 1 ○ exactly ● or more ○ or fewer

☐ Checked as not OK by 1 ○ exactly ● or more ○ or fewer

☐ Checked as OK

☐ Entered on date 09/15/97

☐ With Genotyper: ● error report ○ manual edit ○ either

☒ Omit obsolete records

☐ Omit currently-shown records

☐ Find in currently-shown records

☐ Add to currently-shown records

Browsing Reviewer Comments

The Find Genotype Records command allows you to quickly view records by the comments of particular reviewers. For example, you can quickly find all records checked by a particular reviewer or all records marked with a particular reviewer comment.

These search options are useful when you have multiple users importing and reviewing genotype records. Using this command with the Compare Genotypes command allows you to coordinate multiple copies of a particular record and multiple reviews of those records.

continued on next page

Example Search Procedure

For example, say that you wanted to find records with the following criteria

- Records checked by Lawrence.
- Records that at least one reviewer marked for re-edit.
- Records that contain a Genotyper manual edit.
- Records that reside in the genotype records you have currently displayed.

To conduct the Genotype record search.

Step	Action
1	Select the Find Genotype Records in the User menu.
2	Check the Checked by User Box and select Lawrence from the pop-up menu.
3	Check the Checked As box and select Re-Edit from the pop-up menu.
4	Check the With Genotyper box and click the “manual edit” radio button.
5	In the second section, Check the “Find in currently-shown records” checkbox.
6	Click OK. The records matching the search criteria are displayed in the Genotypes table.

Searching Called Genotypes

Introduction

Search Called Genotype records using the Find Called Genotype command in the User menu. This command works the same as the Find Genotype Records command but uses different search criteria.

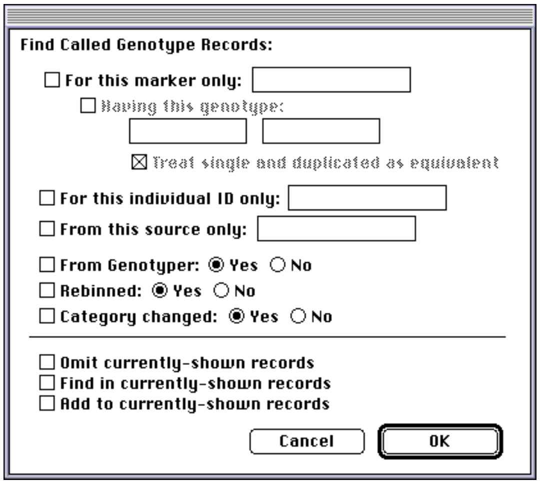
Using the Find Called Genotype Command

The Find Called Genotype dialog includes the following options for searching the Called Genotype table:

- For this marker only–Find Called Genotypes records with a particular maker
- Having this genotype–Specify a particular genotype for which to search
- Treat single and duplicated as equivalent–Prevent the search from displaying multiple copies of a single record.
- For this Individual/Source only–Narrow your search to a particular individual and or source (source name field in the Called Genotype table and External Source table).
- From the Genotyper software–Narrow the search to records called from the Genotypes table.
- Rebinned–Narrow the search to records with a check in the Rebinned field.
- Category Changed–Find records whose categories were changed.

The final three checkboxes allow you to either exclude records already selected from your search, search only the selected records, or add the new records to the list of records you currently have selected.

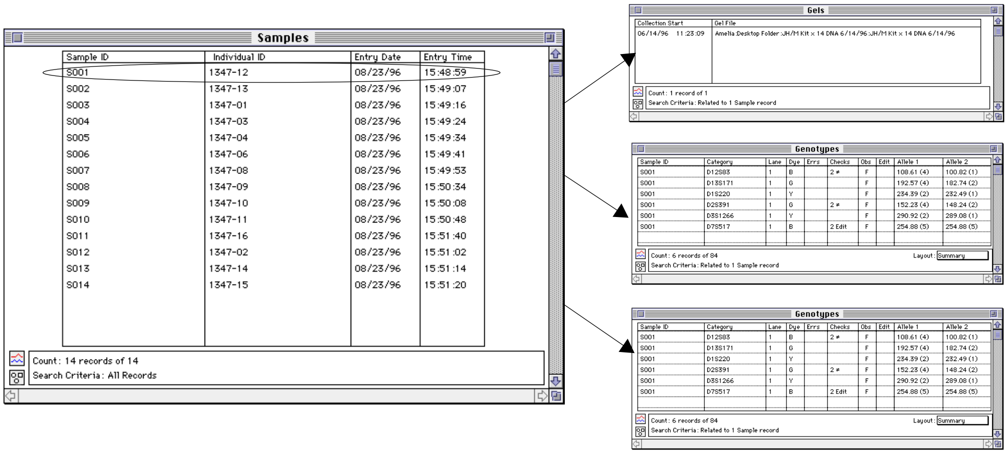
To find called genotypes:

Step	Action
1	Open the Called Genotype table by selecting Called Genotype from the Table menu.
2	Select Find Called Genotype from the User menu. The following window appears:  Find Called Genotype Records: ☐ For this marker only: ☐ Having this genotype: ☒ Treat single and duplicated as equivalent ☐ For this individual ID only: ☐ From this source only: ☐ From Genotyper: ● Yes ○ No ☐ Rebinned: ● Yes ○ No ☐ Category changed: ● Yes ○ No ☐ Omit currently-shown records ☐ Find in currently-shown records ☐ Add to currently-shown records
3	Check boxes as required to set up your search.
4	When the search is complete, the selected records are shown in the Called Genotype table.

Finding Related Records

Overview This command allows searches through the database for records related to ones you have highlighted. The related records are displayed when you open their respective tables.

Finding Related Records The Find Related Records command searches the entire database for records that are directly related to the unique identifier of the selected record. Figure 6-1 shows records related to the Sample S001. (This example is from the Example Database.)



Sample ID	Individual ID	Entry Date	Entry Time
S001	1347-12	08/23/96	15:48:59
S002	1347-13	08/23/96	15:49:07
S003	1347-01	08/23/96	15:49:16
S004	1347-03	08/23/96	15:49:24
S005	1347-04	08/23/96	15:49:34
S006	1347-06	08/23/96	15:49:41
S007	1347-08	08/23/96	15:49:53
S008	1347-09	08/23/96	15:50:34
S009	1347-10	08/23/96	15:50:08
S010	1347-11	08/23/96	15:50:48
S011	1347-16	08/23/96	15:51:40
S012	1347-02	08/23/96	15:51:02
S013	1347-14	08/23/96	15:51:14
S014	1347-15	08/23/96	15:51:20

Count: 14 records of 14
Search Criteria: All Records

Collection Start	Gel File
06/14/96 11:23:09	Amelia:Desktop Folder:JH/M Kit x 14 DNA 6/14/96:JH/M Kit x 14 DNA 6/14/96

Count: 1 record of 1
Search Criteria: Related to 1 Sample record

Sample ID	Category	Lane	Dye	Errs	Checks	Obs	Edit	Allele 1	Allele 2
S001	D12S83	1	B		2 ≠	F		108.61 (4)	100.82 (1)
S001	D13S171	1	G			F		192.57 (4)	182.74 (2)
S001	D1S220	1	Y			F		234.39 (2)	232.49 (1)
S001	D2S391	1	G		2 ≠	F		152.23 (4)	148.24 (2)
S001	D3S1266	1	Y			F		290.92 (2)	289.08 (1)
S001	D7S517	1	B		2 Edit	F		254.88 (5)	254.88 (5)

Count: 6 records of 84
Layout: Summary
Search Criteria: Related to 1 Sample record

Sample ID	Category	Lane	Dye	Errs	Checks	Obs	Edit	Allele 1	Allele 2
S001	D12S83	1	B		2 ≠	F		108.61 (4)	100.82 (1)
S001	D13S171	1	G			F		192.57 (4)	182.74 (2)
S001	D1S220	1	Y			F		234.39 (2)	232.49 (1)
S001	D2S391	1	G		2 ≠	F		152.23 (4)	148.24 (2)
S001	D3S1266	1	Y			F		290.92 (2)	289.08 (1)
S001	D7S517	1	B		2 Edit	F		254.88 (5)	254.88 (5)

Count: 6 records of 84
Layout: Summary
Search Criteria: Related to 1 Sample record

Figure 6-1 Related records

These tables do not automatically open when you select Find Related Records. You must open the relevant table manually. After you conduct a search, a dialog appears indicating the records the GenBase Database application found. The table displays only the records related to your search.

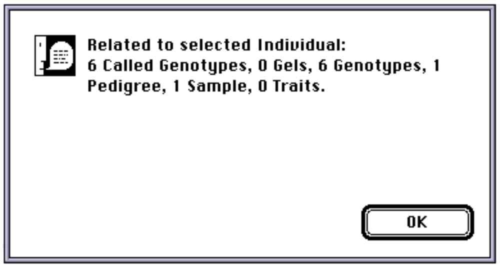
Note The number of displayed records and the search criteria are displayed at the bottom of the table.

This command applies to the following tables:

- Sample
- Genotype
- Pedigree
- Called Genotype

- Individual Trait
- Individual
- Genotype Summary
- Category
- Category Member
- Panels
- Markers
- Gels

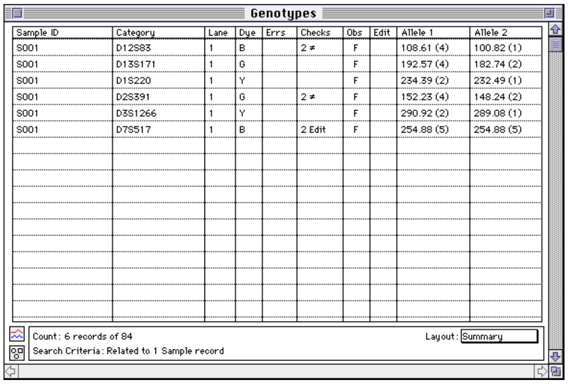
To find related records:

Step	Action
1	Select the record or records you want to find related records for. **Note** This can be single or multiple genotype records or entries from a table.
2	Select Find Related Record from the Records menu.
3	A summary of the related records is displayed in a dialog box:  Related to selected Individual: 6 Called Genotypes, 0 Gels, 6 Genotypes, 1 Pedigree, 1 Sample, 0 Traits.

To find related records: *(continued)*

<table>
<tr><th>Step</th><th>Action</th></tr>
<tr><td>4</td><td>Click OK, and open the designated tables to view the related records.

Note In this example, you could select the Genotypes table from the Table menu, and the related records are displayed.



<table>
<tr><th>Sample ID</th><th>Category</th><th>Lane</th><th>Dye</th><th>Errs</th><th>Checks</th><th>Obs</th><th>Edit</th><th>Allele 1</th><th>Allele 2</th></tr>
<tr><td>S001</td><td>D12S83</td><td>1</td><td>B</td><td></td><td>2 ≠</td><td>F</td><td></td><td>108.61 (4)</td><td>100.82 (1)</td></tr>
<tr><td>S001</td><td>D13S171</td><td>1</td><td>G</td><td></td><td></td><td>F</td><td></td><td>192.57 (4)</td><td>182.74 (2)</td></tr>
<tr><td>S001</td><td>D1S220</td><td>1</td><td>Y</td><td></td><td></td><td>F</td><td></td><td>234.39 (2)</td><td>232.49 (1)</td></tr>
<tr><td>S001</td><td>D2S391</td><td>1</td><td>G</td><td></td><td>2 ≠</td><td>F</td><td></td><td>152.23 (4)</td><td>148.24 (2)</td></tr>
<tr><td>S001</td><td>D3S1266</td><td>1</td><td>Y</td><td></td><td></td><td>F</td><td></td><td>290.92 (2)</td><td>289.08 (1)</td></tr>
<tr><td>S001</td><td>D7S517</td><td>1</td><td>B</td><td></td><td>2 Edit</td><td>F</td><td></td><td>254.88 (5)</td><td>254.88 (5)</td></tr>
</table>
Count : 6 records of 84
Layout : Summary
Search Criteria : Related to 1 Sample record
</td></tr>
</table>

Using Alternate Maps, Markers, or Populations

Introduction

If necessary, maintain alternate maps, markers, or populations to suit your data and requirements for creating exportable files. The following tables support alternate structures:

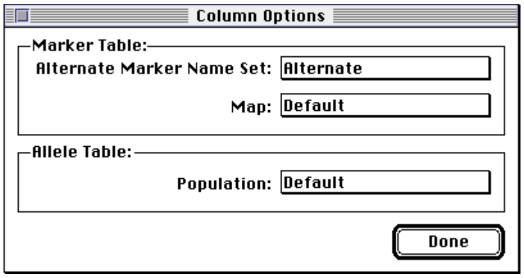
- *Markers*–Assign alternate marker name sets from the Alternate Marker Name Sets table or alternate maps from the Maps table.
- *Alleles*–Assign alleles to populations in the Populations table.

Setting Column Options

The Column Options command allows you to alternate the data displayed in the Alleles or Markers tables. The following options are available in the Column Options dialog box:

- *Alternate Marker Name Set*–Displays markers assigned to alternate name sets. The name sets are designated in the Alternate Marker Name Set table. Choices in the pop-up menu are limited to existing entries in this table. Choosing a name set displays only records with that name in the Marker Name field.
- *Map*–Displays only markers assigned to a particular map. The maps are designated in the Maps table. Only records with that Map name in the Map field are displayed.
- *Population*–Displays the frequency with which an allele occurs in a specific population. Populations are designated in the Population table, and the frequencies displayed are for that population only.

To set column options:

Step	Action
1	Open the table for which you want to set column options. This step is not required.
2	Select Column Options from the Special menu. The following dialog appears:  Marker Table: Alternate Marker Name Set: Alternate Map: Default Allele Table: Population: Default
3	Use the pop-up menus to select new column options. Your choice remains in effect until you change it in the Column Options dialog box.

Alternate Marker Name Sets

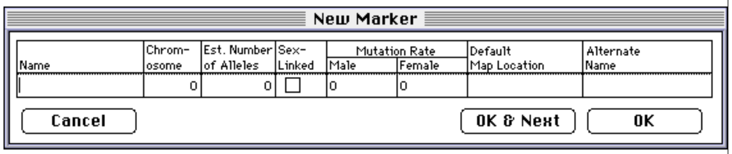
The GenBase Database application allows you to create alternate names for Markers. You may need alternate Marker names because certain downstream applications are selective about the number of characters used to designate Marker names.

Note Before you assign new marker names, you must have created names in the Alternate Marker Names table.

To assign alternate marker names:

Step	Action
1	Select Column Options from the Special menu.
2	Select a marker name from the pop-up menu.
3	Open the Markers table.

To assign alternate marker names: *(continued)*

Step	Action
4	To modify an existing record, double-click the record or highlight it, and select Modify (⌘–M) from the Record menu. To create a new record, select New (⌘–N) from the Record menu. The following dialog appears:  **Note** The name you selected in the Column Options dialog is displayed in the last field (Alternate name).
5	Enter a new name for the marker in the Name field.

New Marker

Name	Chrom-osome	Est. Number of Alleles	Sex-Linked	Mutation Rate Male	Mutation Rate Female	Default Map Location	Alternate Name
	0	0	☐	0	0		

Map Names

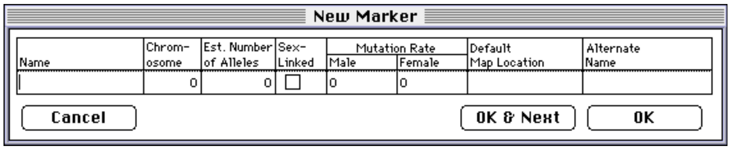
The GenBase Database application allows you to maintain multiple maps for the markers in your database. Multiple maps may be used to account for different measurements or male/female differences.

Note Before you assign new map names, you must have created names in the Maps table.

To assign alternate marker names:

Step	Action
1	Select Column Options from the Special Menu.
2	Select a map from the pop-up menu.
3	Open the Markers table.

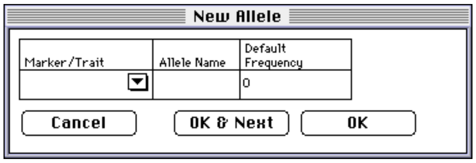
To assign alternate marker names: *(continued)*

Step	Action
4	To modify an existing record, double-click the record or highlight it, and select Modify (⌘–M) from the Record menu. To create a new record, select New (⌘–N) from the Record menu. The following dialog appears:  **Note** The map you selected in the Column Options dialog is displayed in the heading of the Map Location field (Default).
5	Enter the marker location in the new map.

Population Frequencies

The GenBase Database application allows you to specify different allele frequencies to different populations. You may have specified database populations while setting up the Individuals table. If not, create new record(s) in the Populations table before assigning alternate allele frequencies to these populations.

To set allele population frequencies:

Step	Action
1	Open the Allele table.
2	Select Column Options from the Special menu, and select a Population from the pop-up menu.
3	To modify an existing record, double-click the record or highlight it and select Modify (⌘–M) from the Record menu. To create a new record, select New (⌘–N) from the Record menu. The following dialog appears:  **Note** The population for which the frequency applies is specified in the last field (Default).

To set allele population frequencies: *(continued)*

Step	**Action**
4	Enter a new frequency for the specified population.

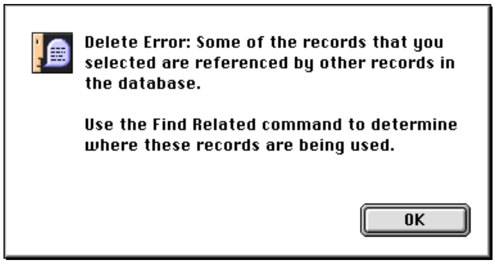
Deleting Records

Maintaining Related Records

Most records share information with several different tables, not just the open one. Because of these interrelations, the GenBase Database application does not allow you to delete records when doing so would create broken cross-references. Deleting an Individual may leave samples assigned to individuals who no longer exist.

Note Three tables, Genotypes, Gels, and Category Members, cannot be edited, and you can only delete records from these tables with the Database Admin access level.

To delete a record:

Step	Action
1	Select the record or records you want to delete.
2	Select Delete from the Record menu. The GenBase Database application conducts a search for related records. If the GenBase Database application determines that deleting the record will cause broken cross-references in the database, the following dialog box appears:  Delete Error: Some of the records that you selected are referenced by other records in the database. Use the Find Related command to determine where these records are being used. Use the Find Related Records command to find and change all related records. If you want to delete an individual from the Individual table, first delete or change records in the Samples table that reference the individual to delete.

To delete a record: *(continued)*

Step	**Action**
3	If the GenBase Database application cannot find records related to the record you want to delete, the following dialog appears:  You selected 1 Map record to delete. Click Delete to proceed. WARNING: You cannot undo this command.
4	Click Delete.

Printing and Exporting Records and Tables

Introduction

The GenBase Database application provides two ways to print documents:

- *Print Record* – Provides a easy way to print selected records or tables.
- *Print Report* – Allows you to construct more detailed reports from data in the open table. The command presents a way of selecting the number of columns of data and what will be in those columns.

You can also choose to export the active table as a text file by using one of two commands:

- *Export* – Saves the active table as a tab-delimited text file formatted as the table itself is.
- *Print Report* – Saves the data in the report to a tab-delimited text file formatted as selected in the report window (this is the same command for printing reports, but uses a different saving option).

Printing Records

This command allows you to quickly print selected records or tables. The printed format matches what you see on the screen.

To create printed records:

Step	Action
1	Select Genotype records or a particular table that you want to print.
2	Select the Print Record command from the File menu.
3	The Page Setup window appears. Configure the window to meet your printing needs, then click OK.

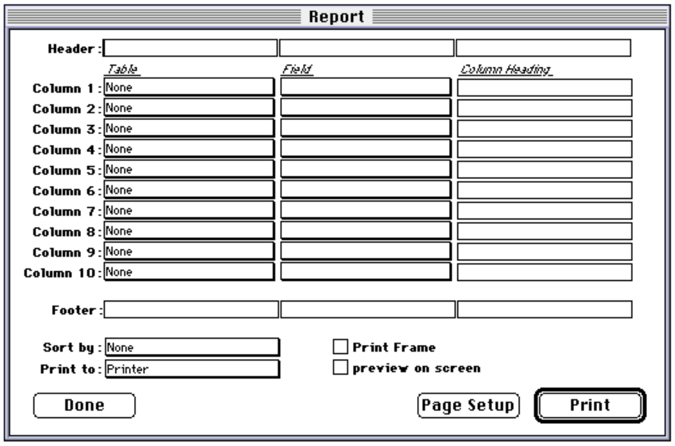
continued on next page

GenBase Reports

The Print Report command under the Record menu provides a way to construct custom reports and save them.

Building Reports

The Report dialog box contains the following sections (Figure 6-2):



Column 1 : None
Column 2 : None
Column 3 : None
Column 4 : None
Column 5 : None
Column 6 : None
Column 7 : None
Column 8 : None
Column 9 : None
Column 10 : None
Sort by : None
Print to : Printer
☐ Print Frame
☐ preview on screen

Figure 6-2 Report dialog box

- *Header*–Three text fields for building a header for your report. The three fields appear in the left corner, middle, and right corner of the page. The header appears on each page of your report.
- *Column 1 through 10*–From pop-up menus, select the Table, Field, and heading for each column.
  - *Table*–Select from the tables that are currently open
  - *Field*–Select fields from the table you selected.
  - *Column Heading*–Create a heading for each column. The GenBase Database application automatically creates a heading based on the field entry that you can modify or delete.
- *Footer*–Three text fields for building a footer for your report. The three fields appear in the left corner, middle, and right corner of the page. The footer appears on each page of your report.
- *Sort by*–Choose one of the field entries by which to sort the report.
- *Print to*–Choose to send the report to the default printer, or create a tab-delimited text file.

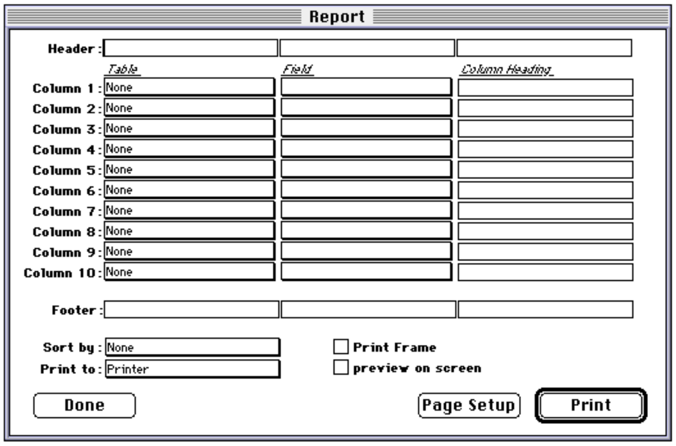
- ♦ *Print Frame*–Choose this option to create a frame around your printed report.
- ♦ *Preview on screen*–Select this checkbox and click Print to see what your report looks like before sending it to the printer.

Printing Reports

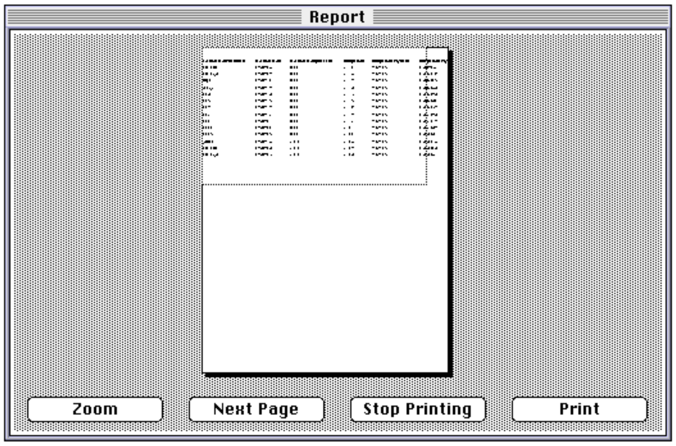
When you have completed selecting the data you want to include in the report, send it to the default printer or create a text file.

Note In most cases, the text file is not in a format that the GenBase Database application can import into a table. To create text files that the GenBase Database application can read, use the Export command from the table menu (see “Creating Text Files” on page 6-24).

To print reports:

Step	Action
1	Select the table for which you want to create a report. You can have multiple tables open, and print only specific records from each.
2	Select Report (⌘-R) from the Record menu. The following dialog box appears:  Column 1 : None Column 2 : None Column 3 : None Column 4 : None Column 5 : None Column 6 : None Column 7 : None Column 8 : None Column 9 : None Column 10 : None Sort by : None Print to : Printer ☐ Print Frame ☐ preview on screen
3	Enter the header and footer for the printed document.
4	Select the contents for each column using the two pop-up menus (Your selection is displayed in the text box to the right. You can edit the text.)
5	Use the “Sort by” pop-up menu to select sort criteria, if needed.

To print reports: *(continued)*

Step	Action
6	From the “Print to” pop-up menu, select the Printer to send the document to or Disk File to convert the report to tab-delimited text and save it as a file.
7	Use the Print Frame checkbox to print a frame around the report.
8	Check “Preview on screen” to see your document before it is sent to the printer. **Note** Do this to save time and paper.
9	Click Print. If you selected “Preview on screen”, a preview appears. Zoom in to particular sections of the preview document, and cancel or continue printing from that point. 

continued on next page

Creating Text Files

Although there are two ways to create text files in the GenBase Database application, the preferred method is by using the Export command in the Records menu. Exported text files contain all the table data in the table's usual format.

All generated text files are tab-delimited and can be read by most word processing or spreadsheet applications.

To create text files using the Export command:

Step	Action
1	From the active table, select Export from the Records menu.
2	From the file dialog that appears, name the file and choose a destination folder.
3	Click Save.

To create text files using the Report command, see "GenBase Reports" on page 6-21, and select Disk File instead of Printer.

Note Keep in mind that text files saved from the Report function are not in a format that the GenBase Database application can use.

Concluding the Experiment

7

Chapter Overview

Introduction This chapter discusses what to do with a completed database regarding downstream applications and long-term storage. The procedures in this chapter primarily require Full Write access.

In This Chapter This chapter includes the following topics:

Exporting Data to Downstream Applications

Discussion The GenBase Database application is designed to be used with the GenoPedigree software to provide data to LINKAGE or other linkage analysis applications (Figure 7-1). The GenBase Database application can also export linkage analysis files using the Linkage Analysis Exports command in the Special menu.

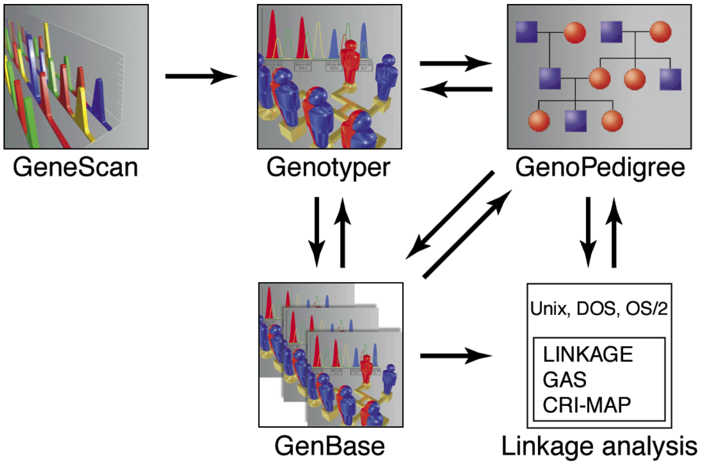


Figure 7-1 Genotyper software interaction

continued on next page

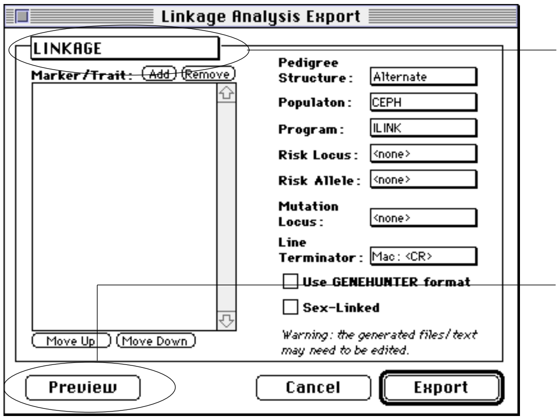
Exporting Directly to Other Applications

You can use the GenBase Database application to create the following types of export files for use with applications other than the GenoPedigree software:

- LINKAGE and GENEHUNTER PRE and DAT files
- CRI-MAP gen files (also can be used by MultiMap)

Note GENEHUNTER and LINKAGE PRE files are equivalent, as are their DAT files with exception of one extra option for GENEHUNTER DAT files.

The GenBase Database application ensures that the data is in the proper format. You select which data to export (Figure 7-2), and the GenBase Database application creates a correctly formatted file for the application you choose.



LINKAGE

Pedigree Structure: Alternate
Populaton: CEPH
Program: ILINK
Risk Locus: <none>
Risk Allele: <none>
Mutation Locus: <none>
Line Terminator: Mac: <CR>
☐ Use GENEHUNTER format
☐ Sex-Linked

Warning: the generated files/text may need to be edited.

Select LINKAGE or CRI-MAP export files

Use the Preview function to check and edit the file before saving

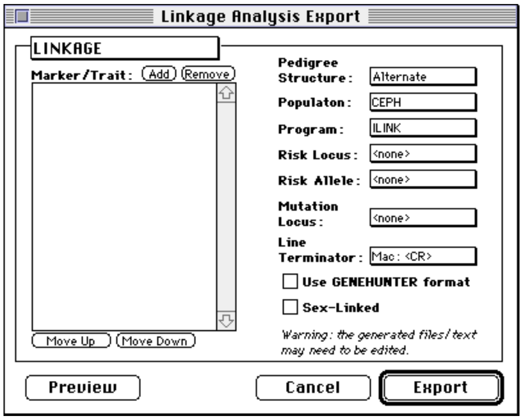
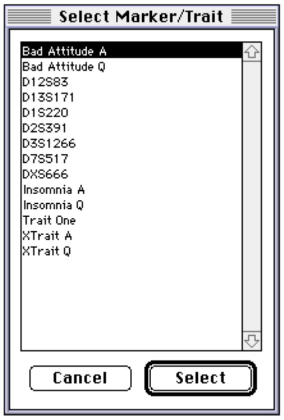
Figure 7-2 Linkage Analysis Export window

LINKAGE Files

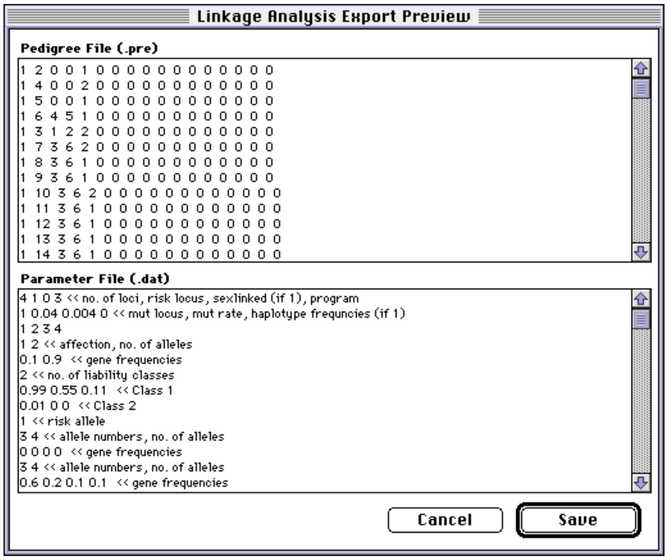
LINKAGE files can be created in the following formats:

- ILINK
- MLINK
- LINKMAP

To create a LINKAGE PRE and/or DAT file:

Step	Action
1	Select Linkage Analysis Exports from the Special menu. The following dialog box appears:  Pedigree Structure: Alternate Populaton: CEPH Program: ILINK Risk Locus: <none> Risk Allele: <none> Mutation Locus: <none> Line Terminator: Mac: <CR> ☐ Use GENEHUNTER format ☐ Sex-Linked *Warning: the generated files/text may need to be edited.*
2	Click Add (located above the Marker/Trait pane). The following window appears:  Bad Attitude A Bad Attitude Q D12S83 D13S171 D1S220 D2S391 D3S1266 D7S517 DXS666 Insomnia A Insomnia Q Trait One XTrait A XTrait Q Highlight the Marker/Trait(s) you want to add and click Select.
3	Select a Pedigree Structure and Population from the pop-up menus. The available selections are taken from the Pedigree Structure and Population tables.

To create a LINKAGE PRE and/or DAT file: *(continued)*

Step	**Action**
4	Select a Program for which to export the file. The available choices are shown in the figure above.
5	Select an allele frequency and population from the pop-up menus.
6	Select Risk Locus and Allele, if any, from the pop-up menus.
7	Select a mutation locus, if any, from the pop-up menus.
8	Select a Line Terminator from the pop-up menu. Make this selection based on the platform the LINKAGE application is to run on.
9	Click Preview. The following window appears: 

Pedigree File (.pre)

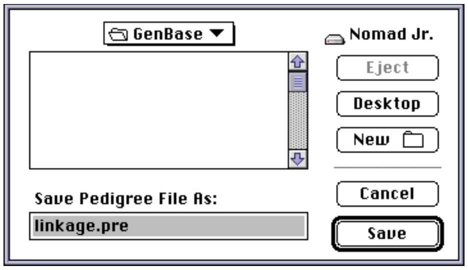
```
1  2  0  0  1  0  0  0  0  0  0  0  0  0  0  0  0  0
1  4  0  0  2  0  0  0  0  0  0  0  0  0  0  0  0  0
1  5  0  0  1  0  0  0  0  0  0  0  0  0  0  0  0  0
1  6  4  5  1  0  0  0  0  0  0  0  0  0  0  0  0  0
1  3  1  2  2  0  0  0  0  0  0  0  0  0  0  0  0  0
1  7  3  6  2  0  0  0  0  0  0  0  0  0  0  0  0  0
1  8  3  6  1  0  0  0  0  0  0  0  0  0  0  0  0  0
1  9  3  6  1  0  0  0  0  0  0  0  0  0  0  0  0  0
1  10  3  6  2  0  0  0  0  0  0  0  0  0  0  0  0  0
1  11  3  6  1  0  0  0  0  0  0  0  0  0  0  0  0  0
1  12  3  6  1  0  0  0  0  0  0  0  0  0  0  0  0  0
1  13  3  6  1  0  0  0  0  0  0  0  0  0  0  0  0  0
1  14  3  6  1  0  0  0  0  0  0  0  0  0  0  0  0  0
```

Parameter File (.dat)

```
4 1 0 3 << no. of loci, risk locus, sexlinked (if 1), program
1 0.04 0.004 0 << mut locus, mut rate, haplotype frequncies (if 1)
1 2 3 4
1 2 << affection, no. of alleles
0.1 0.9  << gene frequencies
2 << no. of liability classes
0.99 0.55 0.11  << Class 1
0.01 0 0  << Class 2
1 << risk allele
3 4 << allele numbers, no. of alleles
0 0 0 0  << gene frequencies
3 4 << allele numbers, no. of alleles
0.6 0.2 0.1 0.1  << gene frequencies
```

10	From the Preview window you can review and edit the output file. If you are creating a LINKAGE DAT file, you must edit the final lines to provide correct recombination fractions or distances. or Click Cancel to return to the Linkage Analysis Exports window.

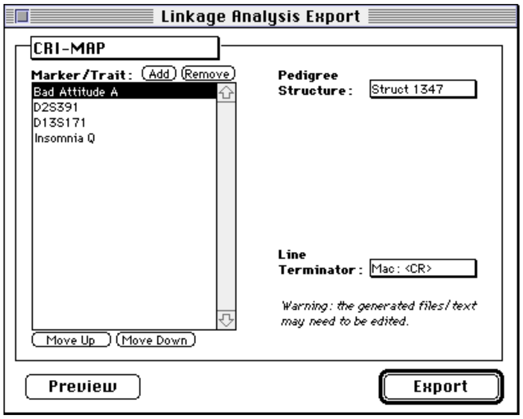
To create a LINKAGE PRE and/or DAT file: *(continued)*

Step	Action
11	Click Export. A save dialog appears:  Save Pedigree File As: linkage.pre
12	Select a destination folder, and click Save.

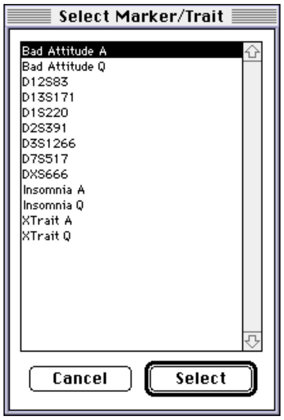
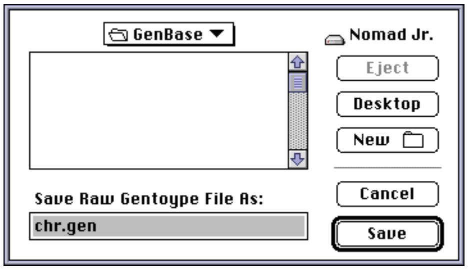
CRI-Map Files

You can also create formatted data files (gen files) for CRI-MAP applications.

To create a CRI-MAP export file:

Step	Action
1	Select Linkage Analysis Exports from the Special menu and select CRI-MAP from the format pop-up menu (see figure below).  Marker/Trait: Bad Attitude A D2S391 D13S171 Insomnia Q Pedigree Structure: Struct 1347 Line Terminator: Mac: <CR> *Warning: the generated files/text may need to be edited.*
2	Select a Pedigree Structure from the pop-up menu. The available selections are taken from the Pedigree Structure table.

To create a CRI-MAP export file: *(continued)*

Step	Action
3	Select a Line Terminator from the pop-up menu.Make this selection based on the platform on which you will be running the CRI-MAP application.
4	Click “Add”. The following window appears:  Highlight the Marker/Trait(s) you want to add to the file and click Select.
5	Click Preview to display the Preview widow.
6	From the Preview window, review and edit the output file. Reprocess the file based on your edits. or Click Cancel to return to the Linkage Analysis Exports window.
7	Click Export. A save dialog appears:  Save Raw Gentoype File As: chr.gen
8	Select a destination folder, and click Save.

continued on next page

GenBase to GenoPedigree

The GenoPedigree software reads data from the GenBase Database application to construct graphical pedigree diagrams, and for further export to linkage analysis and mapping applications. Data must be properly set up in the GenBase Database application prior to constructing pedigree diagrams in the GenoPedigree software. The GenoPedigree software reads information from the following tables:

- Called Genotype
- Liability Classes
- Pedigree
- Pedigree Structure
- Traits
- Phenotypes
- Penetrances
- Individuals
- Markers
- Alleles
- Maps
- Populations

Consult the *GenoPedigree User's Manual* for specific directions on using GenBase data.

The GenoPedigree software can also be used create data files for applications like LINKAGE and GAS. The GenoPedigree software uses data from the tables listed above to export pedigree and locus data files.

Archiving your Database

Archiving Your DataBase

You may choose to continue using your database after completing the experiment, and should archive the data for later use. There are several options including saving a copy i on your hard disk.

To archive your database:

Step	Action
1	Copy your database to a tape backup, magneto-optical (M/O) drive, or hard disk by dragging the database icon onto the drive's icon.
2	Discard all copies of the database on the active hard disk by dragging them into the Trash.
3	Restart the GenBase Database application and select a new, empty database.

GenBase Table Fields

A

Appendix Overview

Introduction This appendix provides a complete list of the fields and their types inside GenBase tables. This information is provided primarily for experienced 4D users who intend to import text files into the database.

In This Appendix This appendix contains the following topics.

Topic	See Page
Table Fields and Types	A-2

Table Fields and Types

Database Fields

This table contains GenBase tables and their respective fields.

Table A-1 GenBase table fields

Table	Field	Type	Unique
Alleles	Marker/Trait Name	pop-up	yes
Alleles	Allele Name	text (7)	yes
Alleles	Population Frequency	real number	
Alt. Marker Name Set	Set	text (15)	yes
Called Genotypes	Individual ID	text (15)	yes
Called Genotypes	Marker Name	pop-up (checkbox for unknown)	yes
Called Genotypes	Allele 1	pop-up (checkbox for unknown)	
Called Genotypes	Allele 2	pop-up	
Called Genotypes	From the Genotyper software	not editable	
Called Genotypes	Rebinned	not editable	
Called Genotypes	Cat. Mod.	not editable	
Called Genotypes	Source	pop-up	
Categories	Panel Name	pop-up	
Categories	Category Name	text (31)	yes
Categories	Marker Name	pop-up	
Category Members	Category Name	not editable	
Category Members	Member	not editable	
Category Members	Definition	not editable	
Category Members	Obsolete	not editable	

Table A-1 GenBase table fields *(continued)*

Table	Field	Type	Unique
External Sources	Source Name	text (31)	yes
Gels	Collection Start	not editable	
Gels	Gel File	not editable	

Table A-1 GenBase table fields *(continued)*

Table	Field	Type	Unique
Genotypes	Sample ID	not editable	
Genotypes	Individual ID	not editable	
Genotypes	Alleles	not editable	
Genotypes	Checks	not editable	
Genotypes	Category	not editable	
Genotypes	Collection Start Date & Time	not editable	
Genotypes	Entry Date & Time	not editable	
Genotypes	Current/Obsolete	not editable	
Genotypes	Obsolete Date & Time	not editable	
Genotypes	Sizing	not editable	
Genotypes	Selection No.	not editable	
Genotypes	Mod score	not editable	
Genotypes	Genotyper Editor	not editable	
Genotypes	Label and/or Table	not editable	
Genotypes	Error O/F	not editable	
Genotypes	Error MOD	not editable	
Genotypes	Error SAT	not editable	
Genotypes	Error LOW	not editable	
Genotypes	Gel name	not editable	
Genotypes	File name	not editable	
Genotypes	Lane	not editable	
Genotypes	Dye	not editable	
Individuals	ID	text (15)	yes
Individuals	Last Name	text (20)	
Individuals	First Name	text (20)	
Individuals	Population	pop-up	
Individuals	Born	date	
Individuals	Died	date	
Liability Classes	Trait Name	pop-up	yes
Liability Classes	Liability Class	text (15)	yes

Table A-1 GenBase table fields *(continued)*

Table	Field	Type	Unique
Liability Classes	Type	pop-up	
Maps	Map Name	text (15)	yes
Markers	Name	text (15)	yes
Markers	Chromosome	integer	
Markers	Est. Num. Alleles	integer	
Markers	Sex Linked	checkbox	
Markers	Mutation Rate Male	real number	
Markers	Mutation Rate Female	real number	
Markers	Default Map Location	text (15)	
Markers	Alternate Name	text (15)	
Panels	Panel Name	text (20)	yes
Pedigrees	Individual ID	text (15)	yes
Pedigrees	Father	text (15)	
Pedigrees	Mother	text (15)	
Pedigrees	Sex	pop-up	
Pedigrees	Pedigree ID	text (31)	
Pedigrees	Pedigree Structure	pop-up	yes
Pedigrees	Multiple Birth	boolean	
Pedigrees	Deceased	boolean	
Pedigrees	Zygote	integer	
Pedigrees	Birth Set	integer	
Pedigrees	Sibship	boolean	

Table A-1 GenBase table fields *(continued)*

Table	Field	Type	Unique
Pedigree Structures	Name	text (31)	yes
Penetrances	Trait Name	pop-up	yes
Penetrances	Liability Class	pop-up	yes
Penetrances	Allele 1	pop-up	yes
Penetrances	Allele 2	pop-up	yes
Penetrances	Penetrance	real number	
Penetrances	Type	text (2)	
Penetrances	Variance	real number	

Table A-1 GenBase table fields *(continued)*

Table	Field	Type	Unique
Phenotypes	Individual ID	pop-up	yes
Phenotypes	Trait Name	pop-up	yes
Phenotypes	Liability Class	pop-up	
Phenotypes	QTL Type	pop-up	
Phenotypes	QTL Value 1	real number (checkbox for unknown)	
Phenotypes	QTL Value 2	real number (checkbox for unknown)	
Populations	Population	text (20)	yes
QTL Variances	Trait	pop-up	
QTL Variances	Trait 1	pop-up	yes
QTL Variances	Trait 2	pop-up	yes
QTL Variances	Variance 1	real number	
QTL Variances	Covariance	real number	
QTL Variances	Variance 2	real number	
QTL Variances	Multiplier	real number	
Samples	Sample ID	text (15)	yes
Samples	Individual ID	text (15)	
Samples	Entry Date	not editable	
Samples	Entry Time	not editable	
Traits	Name	text (15)	yes
Traits	Type	pop-up	
Traits	Sex-Linked	checkbox	
Traits	Mutation Rate Male	real number	
Traits	Mutation Rate Female	real number	

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Headquarters
850 Lincoln Centre Drive
Foster City, CA 94404 USA
Phone: +1 650.638.5800
Toll Free: +1 800.345.5224
Fax: +1 650.638.5884

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