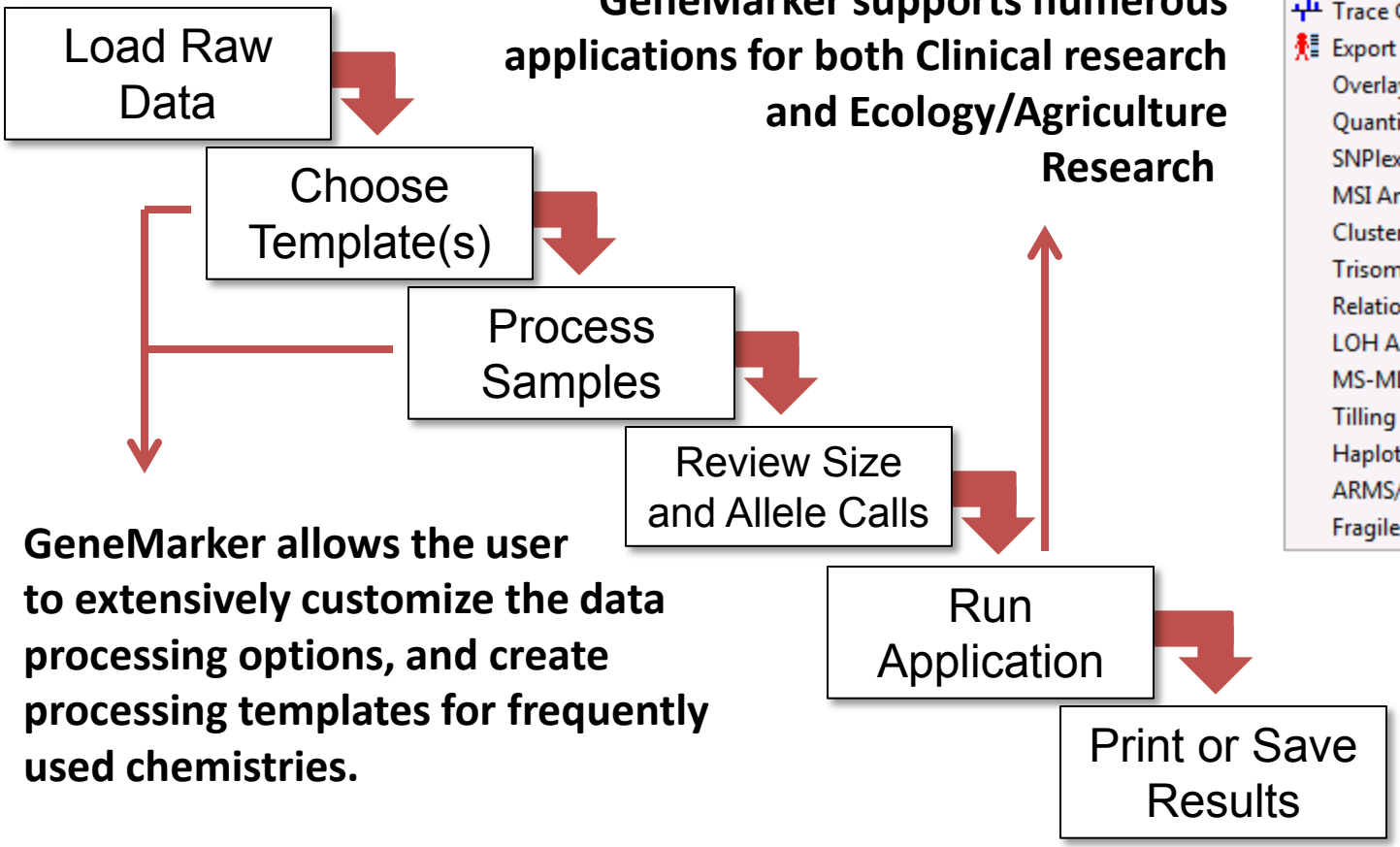
Dotmatics

SoftGenetics

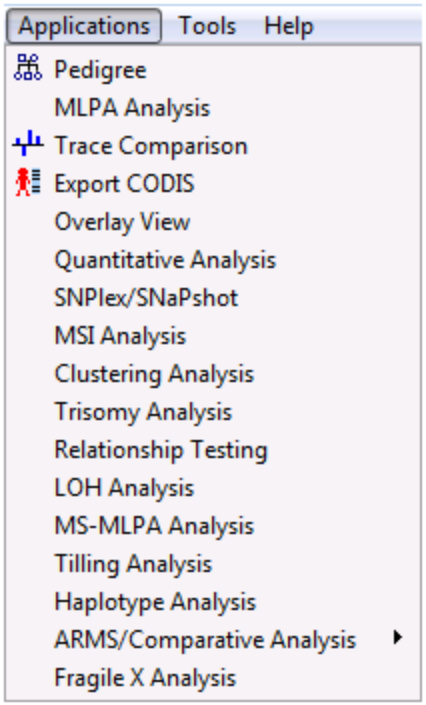
# GeneMarker

## Quick Start Guide

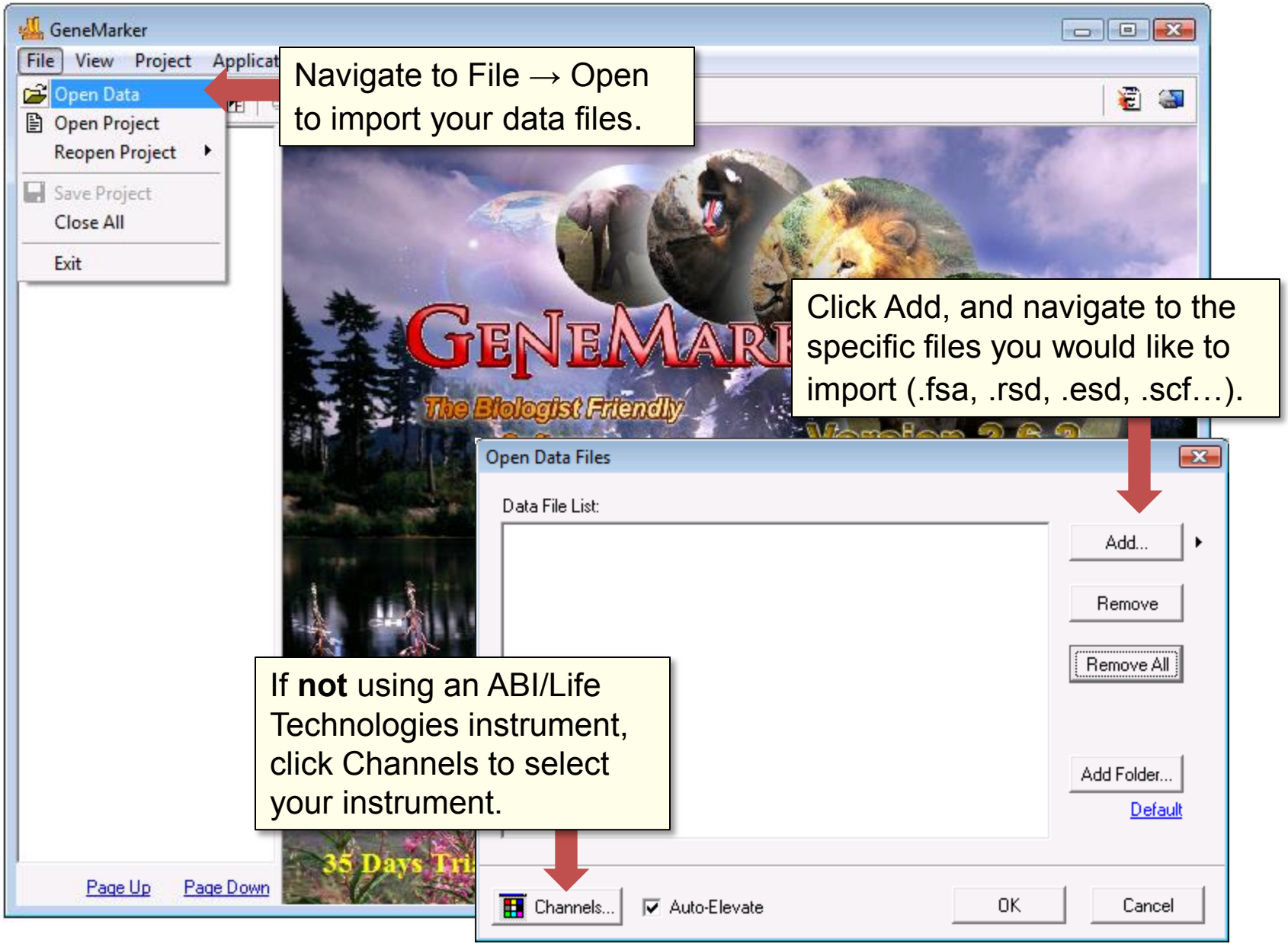
# GeneMarker Workflow

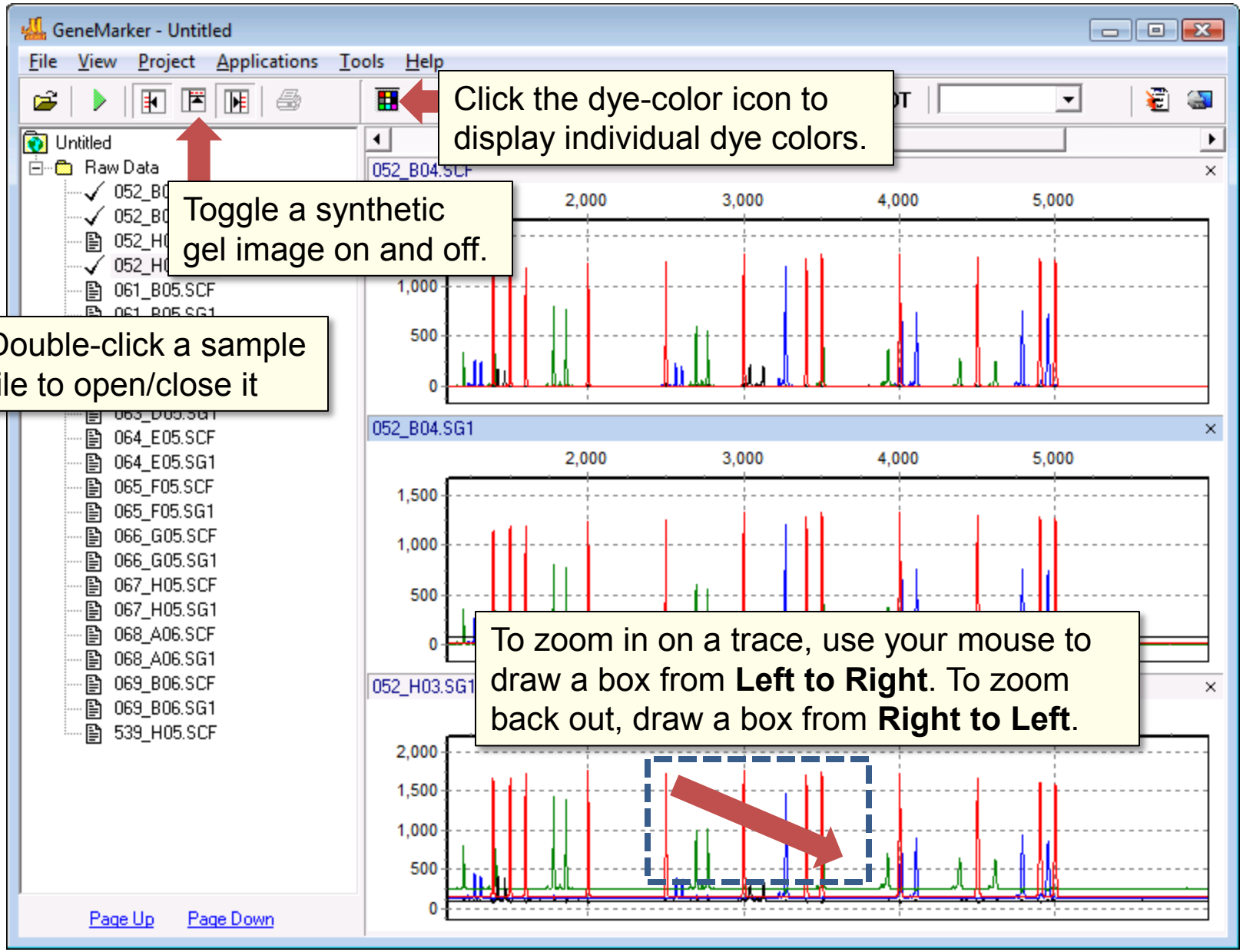


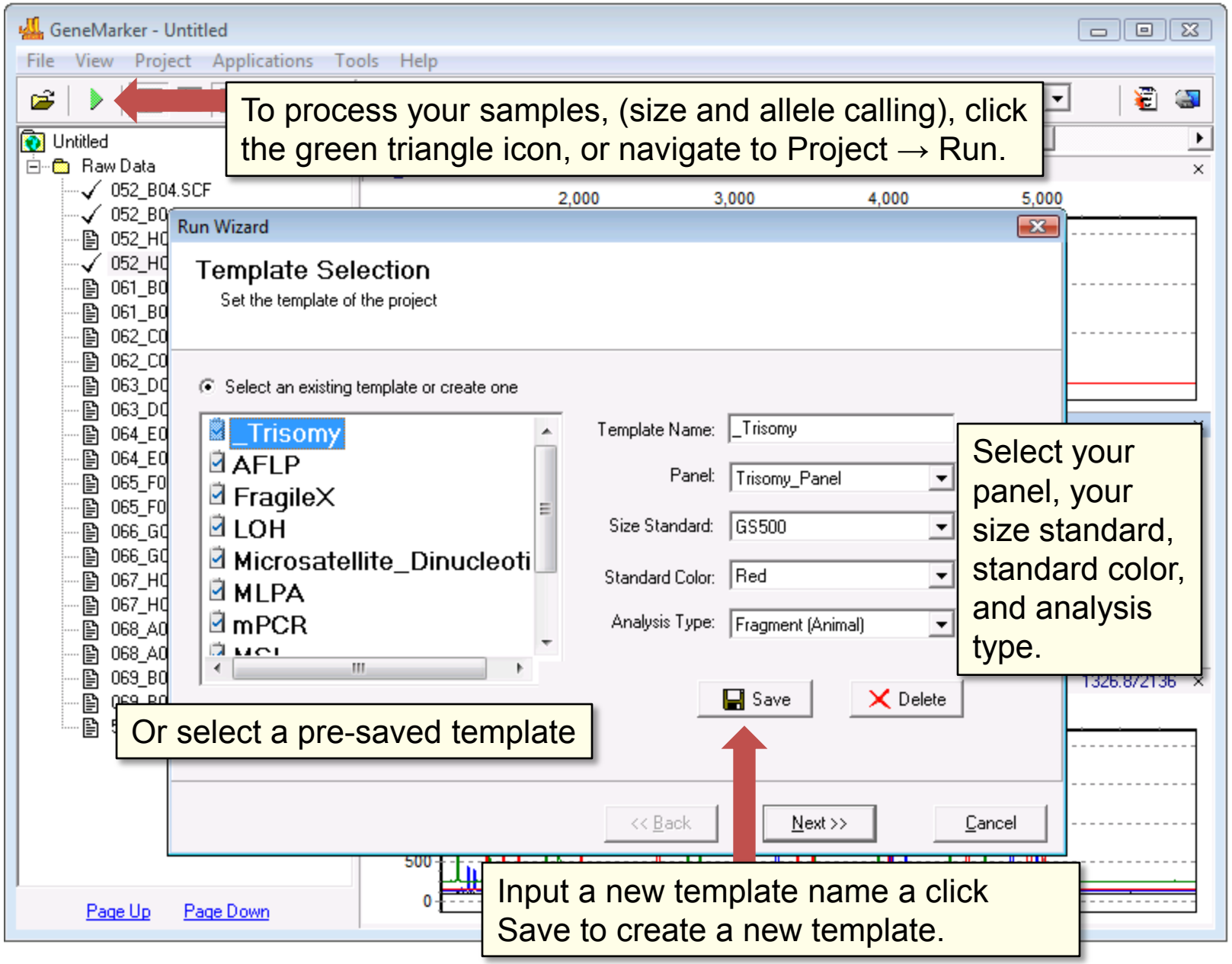

**GeneMarker supports numerous applications for both Clinical research and Ecology/Agriculture Research**

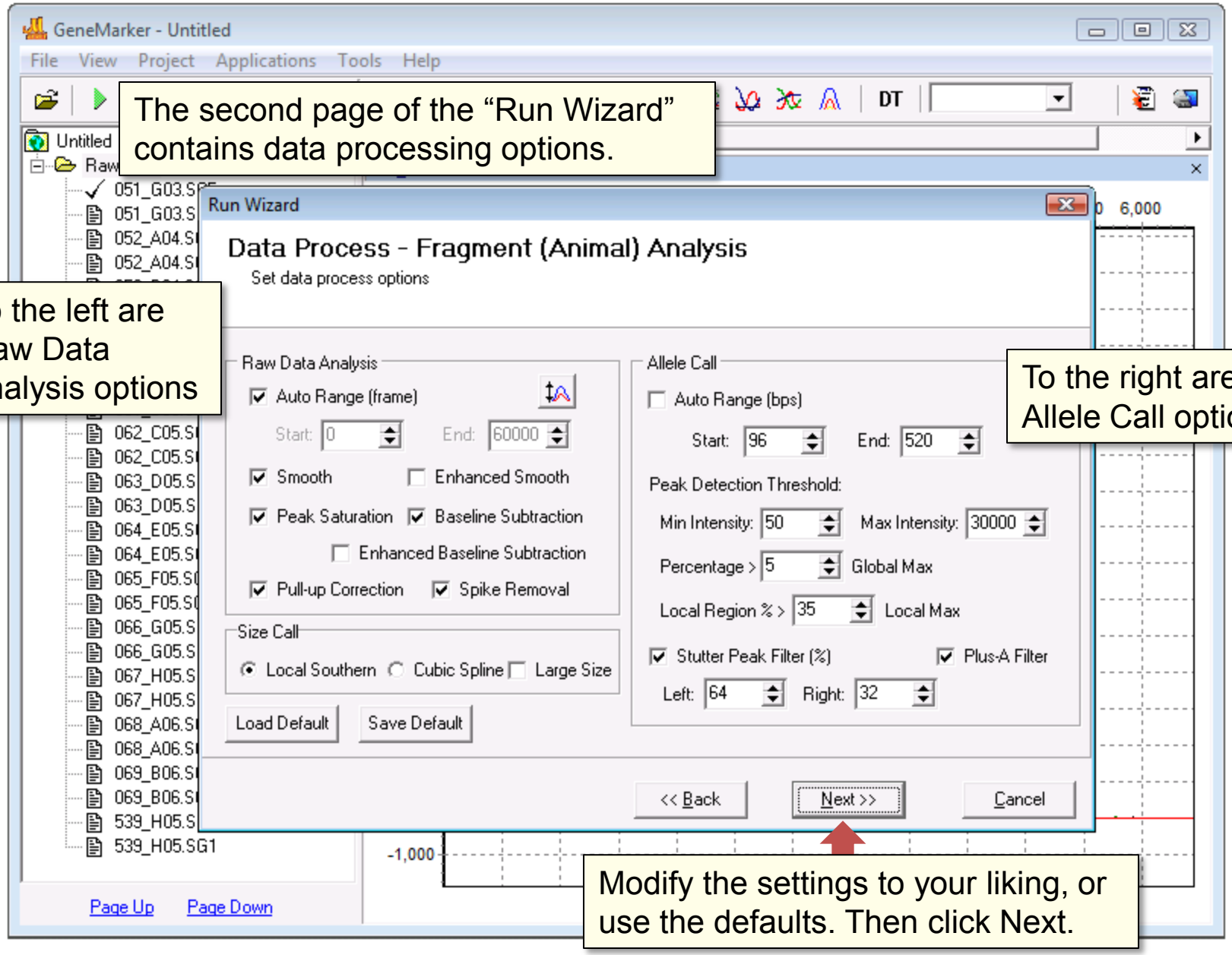


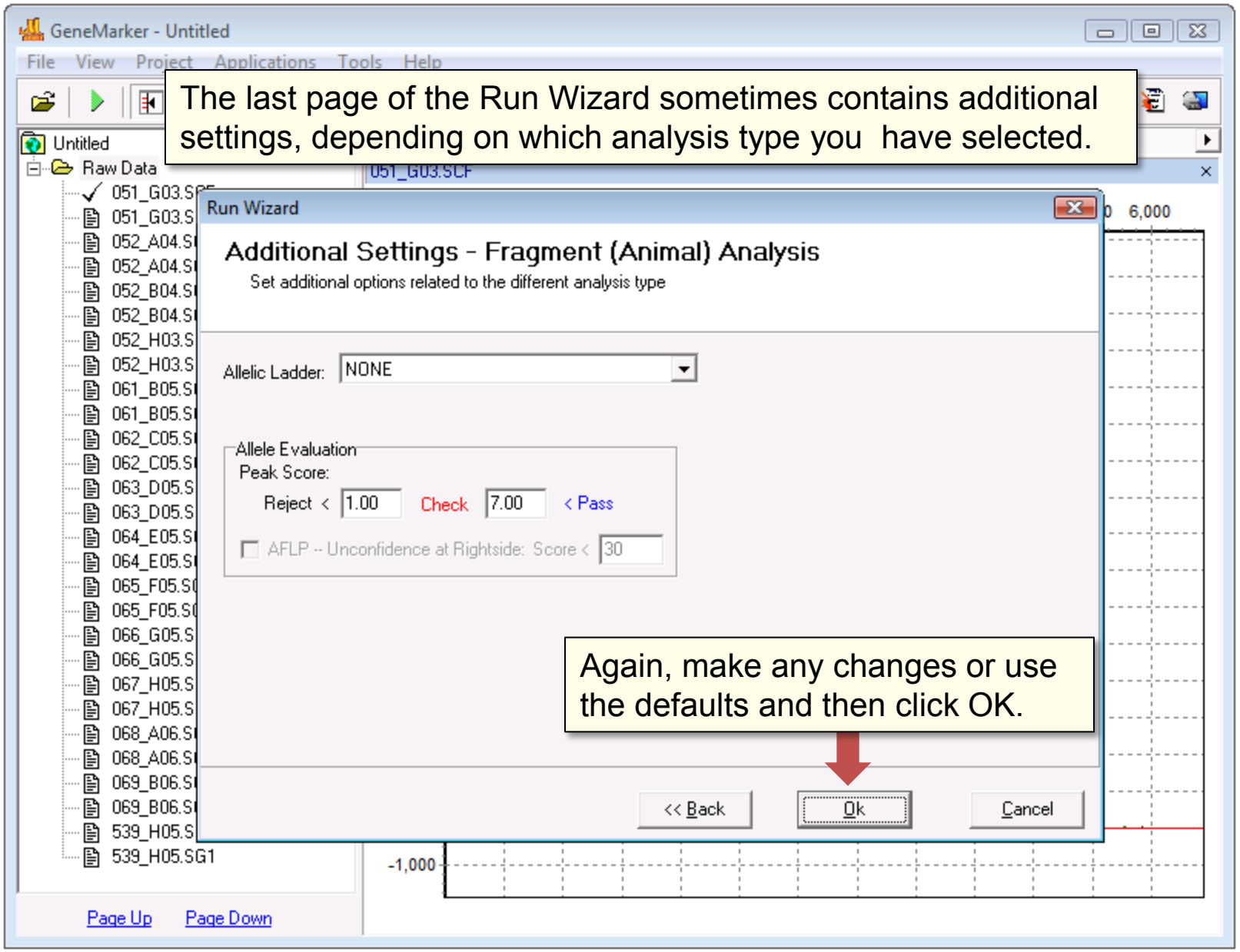

**GeneMarker allows the user to extensively customize the data processing options, and create processing templates for frequently used chemistries.**

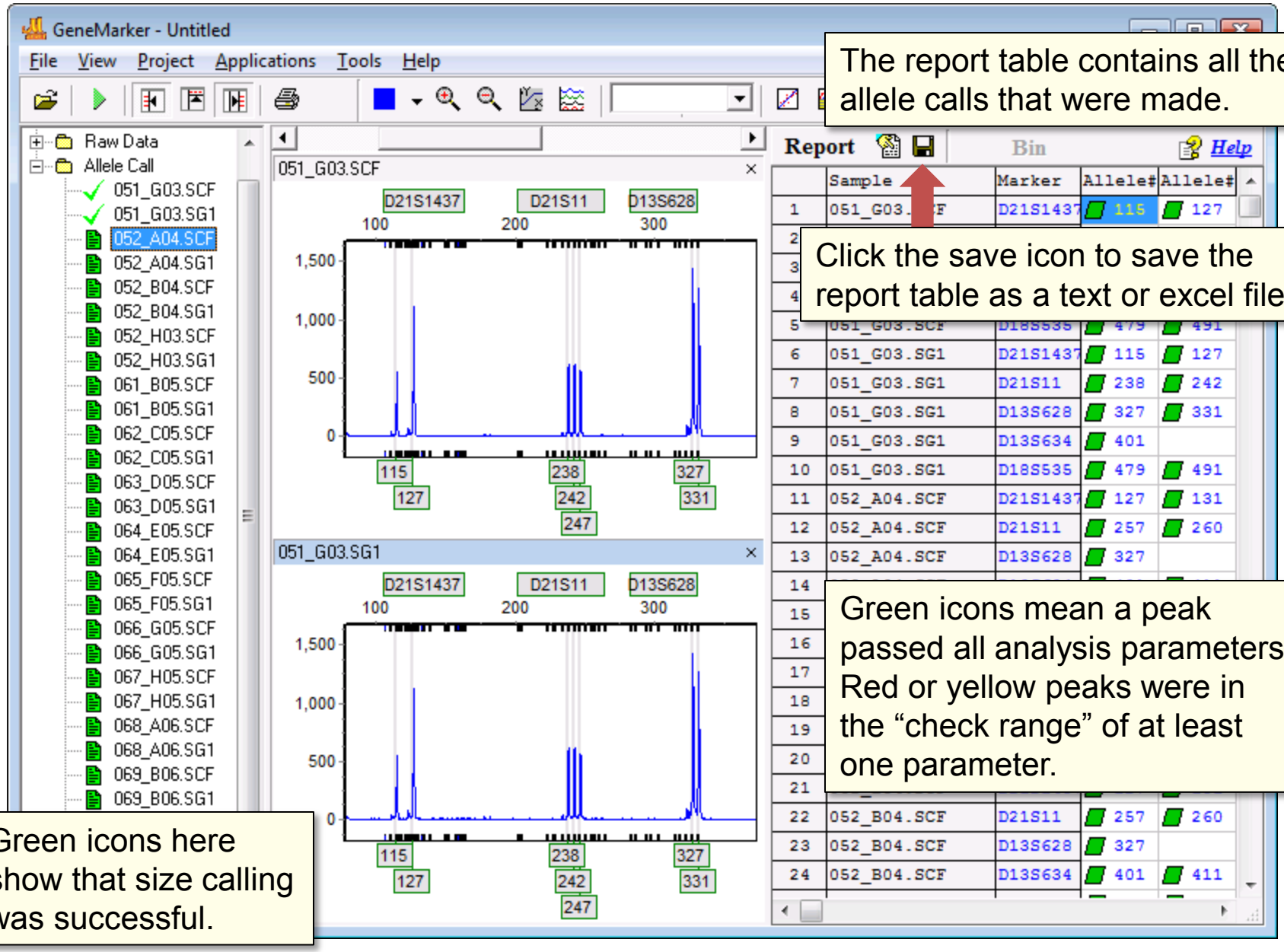

GeneMarker
File View Project Applicat
Open Data
Open Project
Reopen Project
Save Project
Close All
Exit
Navigate to File → Open to import your data files.
The Biologist Friendly
Click Add, and navigate to the specific files you would like to import (.fsa, .rsd, .esd, .scf…).
Open Data Files
Data File List:
Add...
Remove
Remove All
Add Folder...
Default
If **not** using an ABI/Life Technologies instrument, click Channels to select your instrument.
Page Up
Page Down
Channels...
Auto-Elevate
OK
Cancel


GeneMarker - Untitled
File View Project Applications Tools Help
Click the dye-color icon to display individual dye colors.
Toggle a synthetic gel image on and off.
Double-click a sample file to open/close it
To zoom in on a trace, use your mouse to draw a box from **Left to Right**. To zoom back out, draw a box from **Right to Left**.
Untitled
Raw Data
061_B05.SCF
064_E05.SCF
064_E05.SG1
065_F05.SCF
065_F05.SG1
066_G05.SCF
066_G05.SG1
067_H05.SCF
067_H05.SG1
068_A06.SCF
068_A06.SG1
069_B06.SCF
069_B06.SG1
539_H05.SCF
Page Up
Page Down
052_B04.SCF
052_B04.SG1
052_H03.SG1
2,000
3,000
4,000
5,000
1,500
1,000
500
0


GeneMarker - Untitled
File View Project Applications Tools Help
To process your samples, (size and allele calling), click the green triangle icon, or navigate to Project → Run.
Untitled
Raw Data
052_B04.SCF
2,000
3,000
4,000
5,000
Run Wizard
Template Selection
Set the template of the project
Select an existing template or create one
_Trisomy
AFLP
FragileX
LOH
Microsatellite_Dinucleoti
MLPA
mPCR
Template Name: _Trisomy
Panel: Trisomy_Panel
Size Standard: GS500
Standard Color: Red
Analysis Type: Fragment (Animal)
Select your panel, your size standard, standard color, and analysis type.
Save
Delete
Or select a pre-saved template
<< Back
Next >>
Cancel
Input a new template name a click Save to create a new template.
Page Up
Page Down


GeneMarker - Untitled
File View Project Applications Tools Help
DT
Untitled
Raw
051_G03.S
051_G03.S
052_A04.S
052_A04.S
062_C05.S
062_C05.S
063_D05.S
063_D05.S
064_E05.S
064_E05.S
065_F05.S
065_F05.S
066_G05.S
066_G05.S
067_H05.S
067_H05.S
068_A06.S
068_A06.S
069_B06.S
069_B06.S
539_H05.S
539_H05.SG1
Page Up
Page Down
6,000
-1,000
The second page of the “Run Wizard” contains data processing options.
To the left are Raw Data analysis options
To the right are Allele Call options
Modify the settings to your liking, or use the defaults. Then click Next.
Run Wizard
Data Process - Fragment (Animal) Analysis
Set data process options
Raw Data Analysis
Auto Range (frame)
Start: 0
End: 60000
Smooth
Enhanced Smooth
Peak Saturation
Baseline Subtraction
Enhanced Baseline Subtraction
Pull-up Correction
Spike Removal
Size Call
Local Southern
Cubic Spline
Large Size
Load Default
Save Default
Allele Call
Auto Range (bps)
Start: 96
End: 520
Peak Detection Threshold:
Min Intensity: 50
Max Intensity: 30000
Percentage > 5
Global Max
Local Region % > 35
Local Max
Stutter Peak Filter (%)
Plus-A Filter
Left: 64
Right: 32
<< Back
Next >>
Cancel


GeneMarker - Untitled
File View Project Applications Tools Help
The last page of the Run Wizard sometimes contains additional settings, depending on which analysis type you have selected.
Untitled
Raw Data
051_G03.SCF
Run Wizard
Additional Settings - Fragment (Animal) Analysis
Set additional options related to the different analysis type
Allelic Ladder: NONE
Allele Evaluation
Peak Score:
Reject < 1.00 Check 7.00 < Pass
AFLP -- Unconfidence at Rightside: Score < 30
Again, make any changes or use the defaults and then click OK.
<< Back
Ok
Cancel
6,000
-1,000
Page Up
Page Down

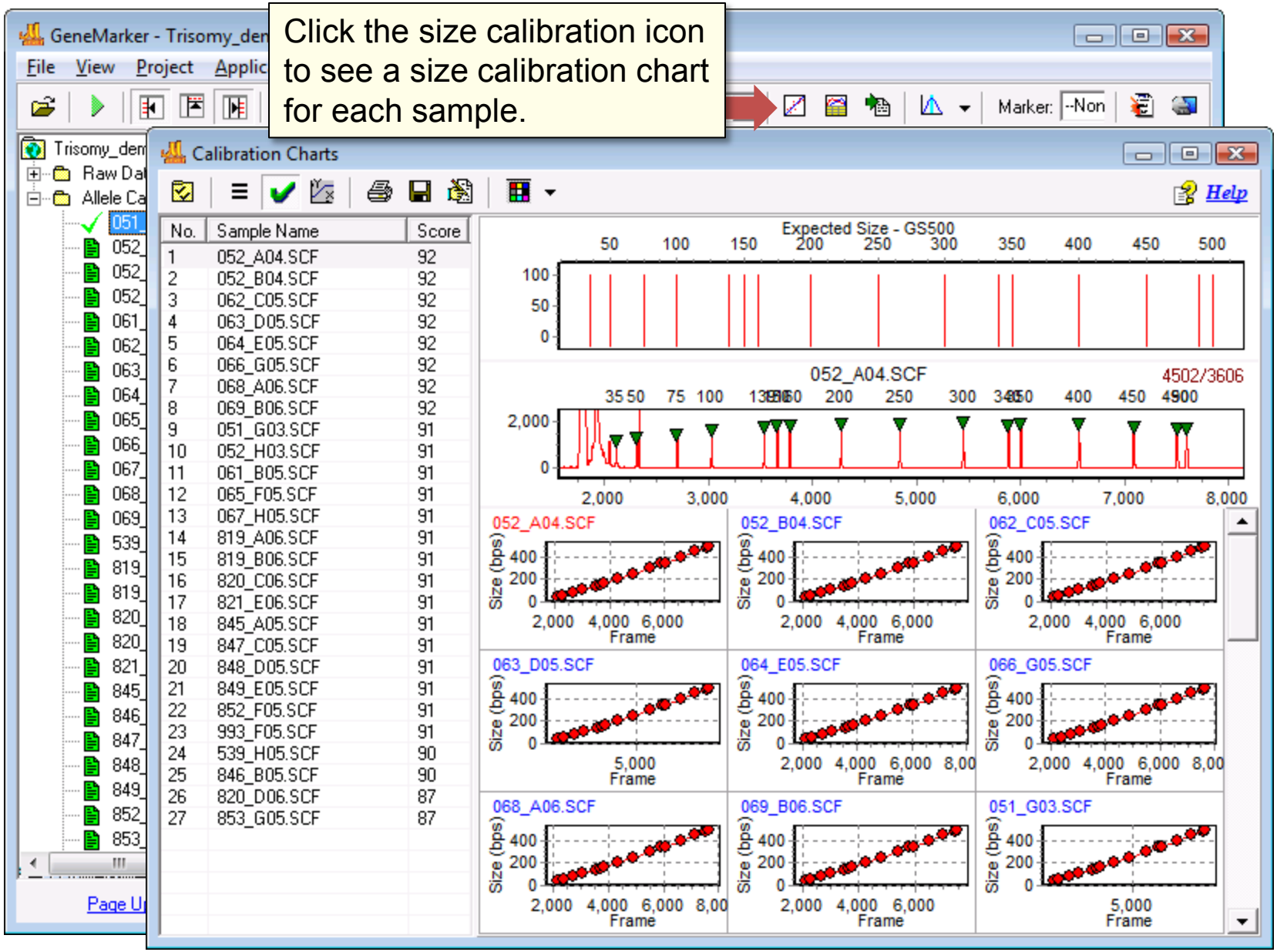


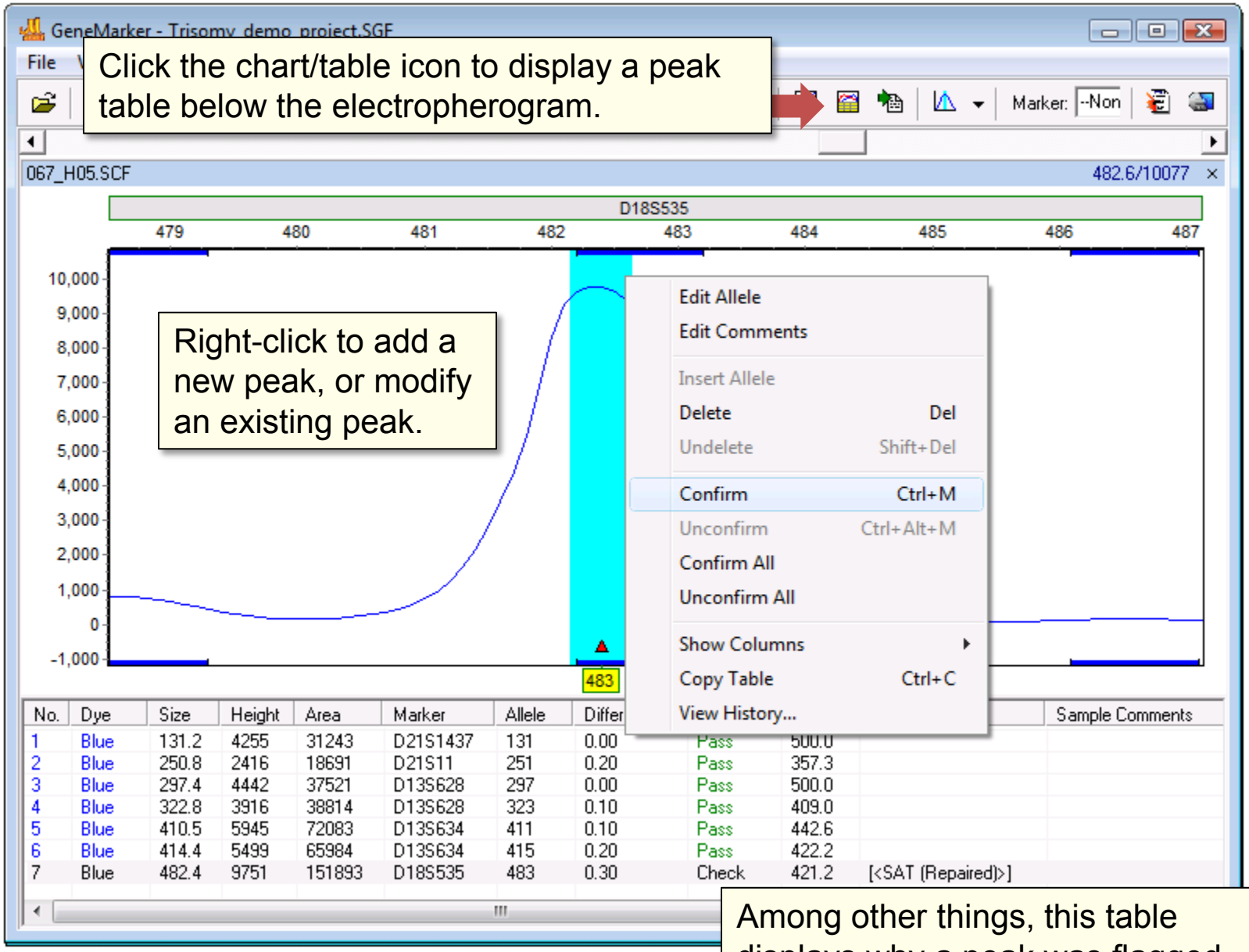

The report table contains all the allele calls that were made.

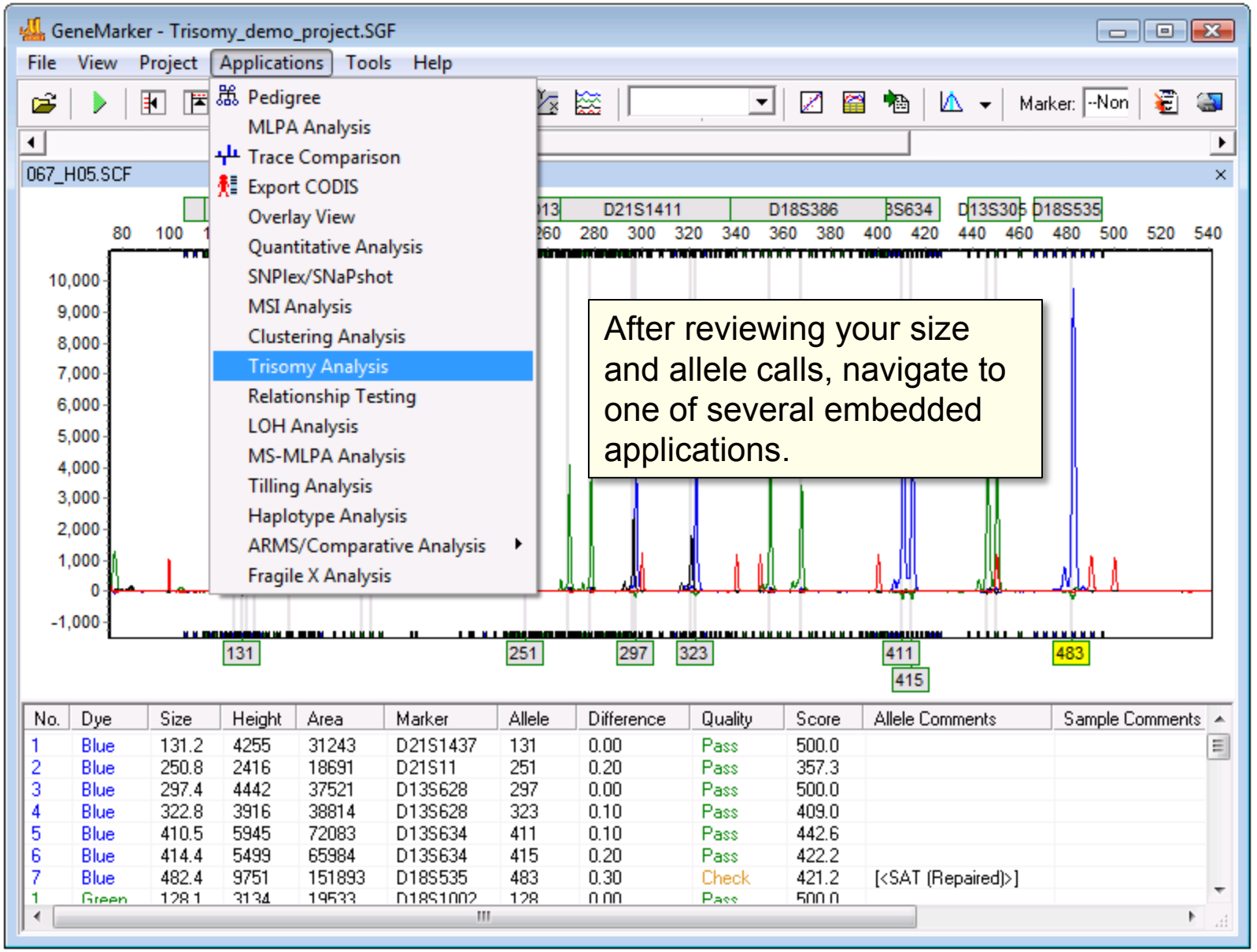
Click the save icon to save the report table as a text or excel file.

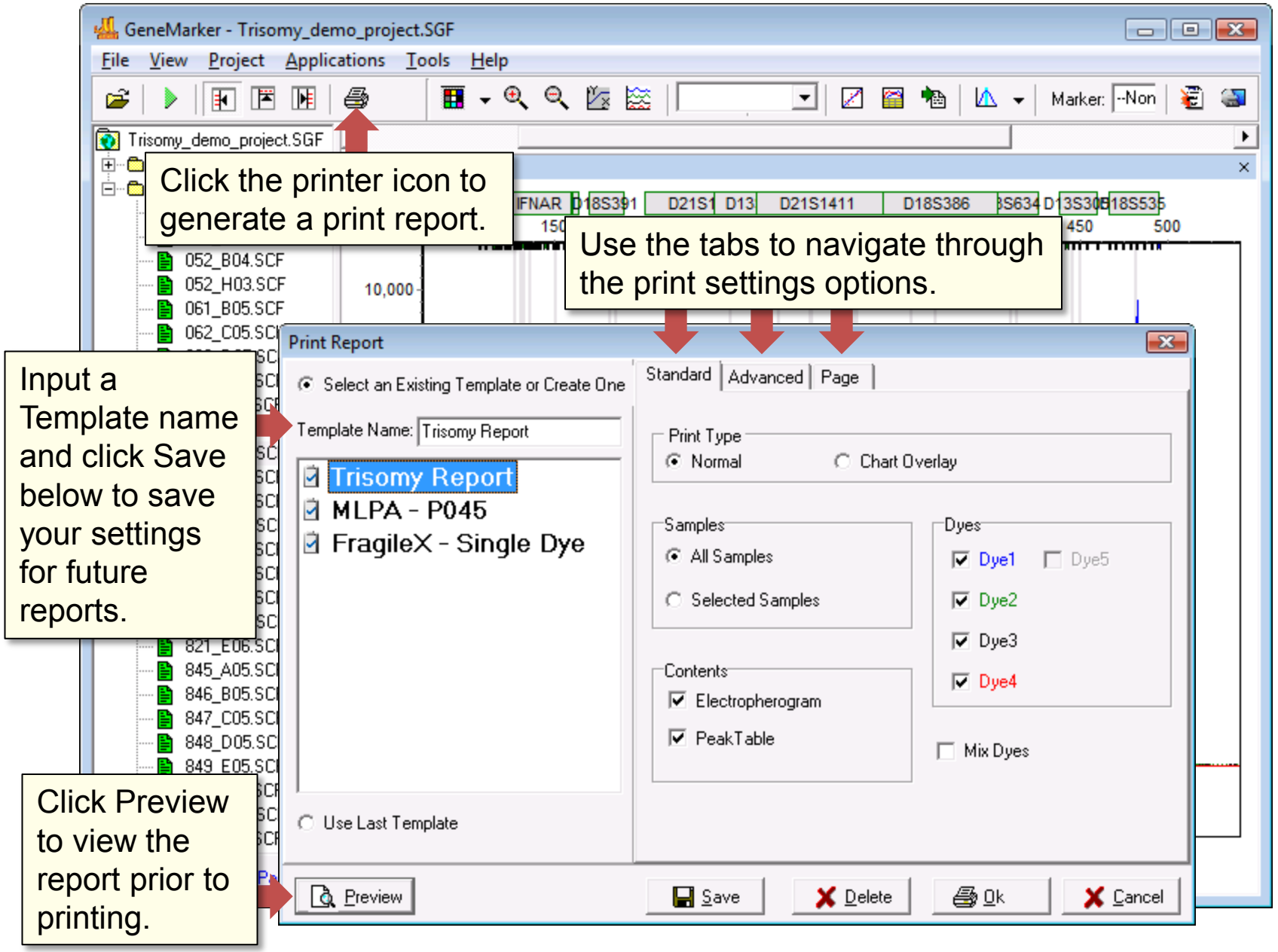
Green icons mean a peak passed all analysis parameters. Red or yellow peaks were in the “check range” of at least one parameter.

Green icons here show that size calling was successful.


Click the size calibration icon to see a size calibration chart for each sample.
GeneMarker - Trisomy_den
File View Project Applic
Marker: --Non
Trisomy_den
Raw Da
Allele Ca
Page U
Calibration Charts
Help
No. Sample Name Score
1 052_A04.SCF 92
2 052_B04.SCF 92
3 062_C05.SCF 92
4 063_D05.SCF 92
5 064_E05.SCF 92
6 066_G05.SCF 92
7 068_A06.SCF 92
8 069_B06.SCF 92
9 051_G03.SCF 91
10 052_H03.SCF 91
11 061_B05.SCF 91
12 065_F05.SCF 91
13 067_H05.SCF 91
14 819_A06.SCF 91
15 819_B06.SCF 91
16 820_C06.SCF 91
17 821_E06.SCF 91
18 845_A05.SCF 91
19 847_C05.SCF 91
20 848_D05.SCF 91
21 849_E05.SCF 91
22 852_F05.SCF 91
23 993_F05.SCF 91
24 539_H05.SCF 90
25 846_B05.SCF 90
26 820_D06.SCF 87
27 853_G05.SCF 87
Expected Size - GS500
052_A04.SCF
4502/3606
Size (bps)
Frame
052_A04.SCF
052_B04.SCF
062_C05.SCF
063_D05.SCF
064_E05.SCF
066_G05.SCF
068_A06.SCF
069_B06.SCF
051_G03.SCF

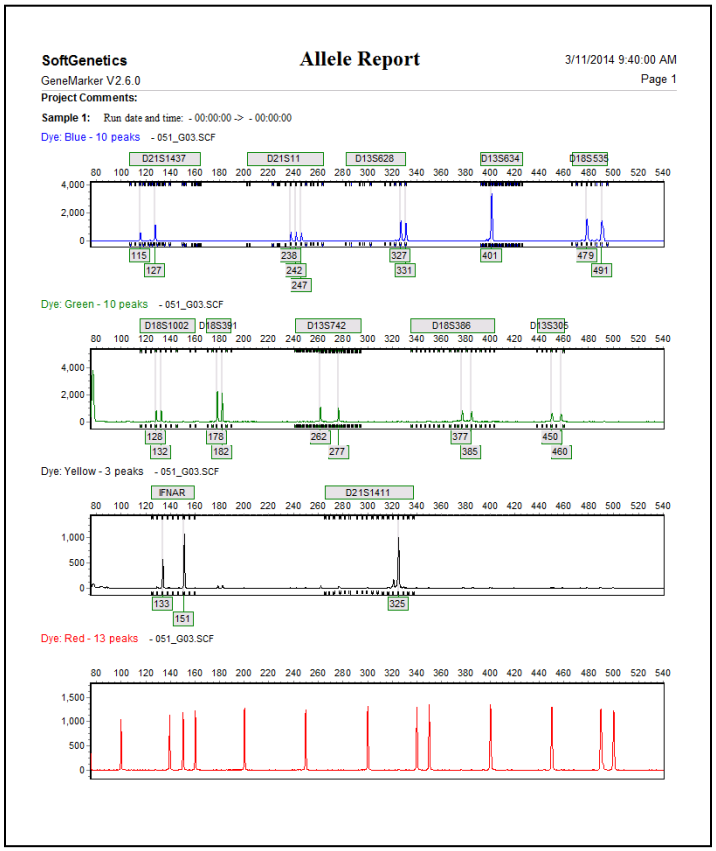

GeneMarker - Trisomy_demo_project.SGF
File
Click the chart/table icon to display a peak table below the electropherogram.
Marker: --Non
067_H05.SCF
482.6/10077
D18S535
479 480 481 482 483 484 485 486 487
10,000 9,000 8,000 7,000 6,000 5,000 4,000 3,000 2,000 1,000 0 -1,000
Right-click to add a new peak, or modify an existing peak.
483
Edit Allele
Edit Comments
Insert Allele
Delete Del
Undelete Shift+Del
Confirm Ctrl+M
Unconfirm Ctrl+Alt+M
Confirm All
Unconfirm All
Show Columns
Copy Table Ctrl+C
View History...
No. Dye Size Height Area Marker Allele Differ Sample Comments
1 Blue 131.2 4255 31243 D21S1437 131 0.00 Pass 500.0
2 Blue 250.8 2416 18691 D21S11 251 0.20 Pass 357.3
3 Blue 297.4 4442 37521 D13S628 297 0.00 Pass 500.0
4 Blue 322.8 3916 38814 D13S628 323 0.10 Pass 409.0
5 Blue 410.5 5945 72083 D13S634 411 0.10 Pass 442.6
6 Blue 414.4 5499 65984 D13S634 415 0.20 Pass 422.2
7 Blue 482.4 9751 151893 D18S535 483 0.30 Check 421.2 [<SAT (Repaired)>]
Among other things, this table displays why a peak was flagged.


GeneMarker - Trisomy_demo_project.SGF
File View Project Applications Tools Help
Pedigree
MLPA Analysis
Trace Comparison
Export CODIS
Overlay View
Quantitative Analysis
SNPlex/SNaPshot
MSI Analysis
Clustering Analysis
Trisomy Analysis
Relationship Testing
LOH Analysis
MS-MLPA Analysis
Tilling Analysis
Haplotype Analysis
ARMS/Comparative Analysis
Fragile X Analysis
Marker: --Non
067_H05.SCF
D21S1411
D18S386
D18S535
After reviewing your size and allele calls, navigate to one of several embedded applications.
No. Dye Size Height Area Marker Allele Difference Quality Score Allele Comments Sample Comments
1 Blue 131.2 4255 31243 D21S1437 131 0.00 Pass 500.0
2 Blue 250.8 2416 18691 D21S11 251 0.20 Pass 357.3
3 Blue 297.4 4442 37521 D13S628 297 0.00 Pass 500.0
4 Blue 322.8 3916 38814 D13S628 323 0.10 Pass 409.0
5 Blue 410.5 5945 72083 D13S634 411 0.10 Pass 442.6
6 Blue 414.4 5499 65984 D13S634 415 0.20 Pass 422.2
7 Blue 482.4 9751 151893 D18S535 483 0.30 Check 421.2 [<SAT (Repaired)>]

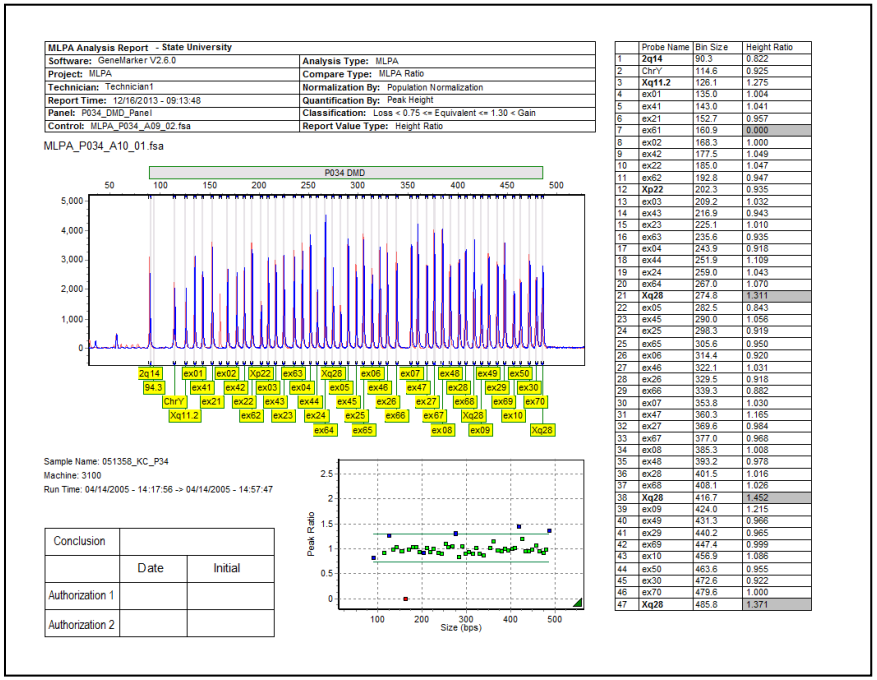

GeneMarker - Trisomy_demo_project.SGF
File View Project Applications Tools Help
Marker: --Non
Trisomy_demo_project.SGF
Click the printer icon to generate a print report.
Use the tabs to navigate through the print settings options.
Input a Template name and click Save below to save your settings for future reports.
Click Preview to view the report prior to printing.
Print Report
Select an Existing Template or Create One
Template Name: Trisomy Report
Trisomy Report
MLPA - P045
FragileX - Single Dye
Use Last Template
Standard
Advanced
Page
Print Type
Normal
Chart Overlay
Samples
All Samples
Selected Samples
Dyes
Dye1
Dye5
Dye2
Dye3
Dye4
Contents
Electropherogram
PeakTable
Mix Dyes
Preview
Save
Delete
Ok
Cancel

# Gene Marker Supports Numerous Reporting Options

Below are two example reports that can be generated in GeneMarker. Each post-genotyping application has its own unique report format that can be extensively customized. Reports can be printed directly or saved as PNG and JPG image files.



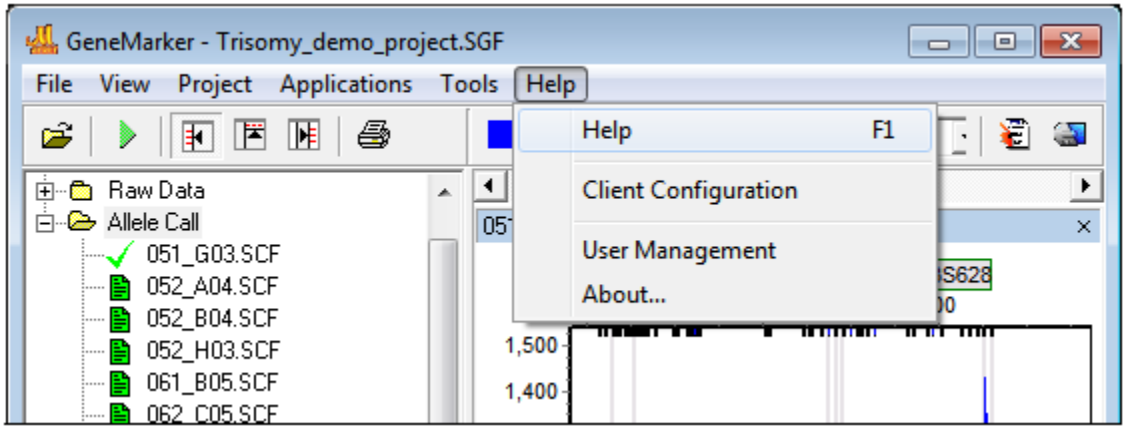

A typical print report from one of the many post-genotyping applications (MLPA):




| | Probe Name | Bin Size | Height Ratio |
|---|---|---|---|
| 1 | 2q14 | 90.3 | 0.822 |
| 2 | ChrY | 114.6 | 0.925 |
| 3 | Xq11.2 | 126.1 | 1.275 |
| 4 | ex01 | 135.0 | 1.004 |
| 5 | ex41 | 143.0 | 1.041 |
| 6 | ex21 | 152.7 | 0.957 |
| 7 | ex61 | 160.9 | 0.000 |
| 8 | ex02 | 168.3 | 1.000 |
| 9 | ex42 | 177.5 | 1.049 |
| 10 | ex22 | 185.0 | 1.047 |
| 11 | ex62 | 192.8 | 0.947 |
| 12 | Xp22 | 202.3 | 0.935 |
| 13 | ex03 | 209.2 | 1.032 |
| 14 | ex43 | 216.9 | 0.943 |
| 15 | ex23 | 225.1 | 1.010 |
| 16 | ex63 | 235.6 | 0.935 |
| 17 | ex04 | 243.9 | 0.918 |
| 18 | ex44 | 251.9 | 1.109 |
| 19 | ex24 | 259.0 | 1.043 |
| 20 | ex64 | 267.0 | 1.070 |
| 21 | Xq28 | 274.8 | 1.311 |
| 22 | ex05 | 282.5 | 0.843 |
| 23 | ex45 | 290.0 | 1.056 |
| 24 | ex25 | 298.3 | 0.919 |
| 25 | ex65 | 305.6 | 0.950 |
| 26 | ex06 | 314.4 | 0.920 |
| 27 | ex46 | 322.1 | 1.031 |
| 28 | ex26 | 329.5 | 0.918 |
| 29 | ex66 | 339.3 | 0.882 |
| 30 | ex07 | 353.8 | 1.030 |
| 31 | ex47 | 360.3 | 1.165 |
| 32 | ex27 | 369.6 | 0.984 |
| 33 | ex67 | 377.0 | 0.968 |
| 34 | ex08 | 385.3 | 1.008 |
| 35 | ex48 | 393.2 | 0.978 |
| 36 | ex28 | 401.5 | 1.016 |
| 37 | ex68 | 408.1 | 1.026 |
| 38 | Xq28 | 416.7 | 1.452 |
| 39 | ex09 | 424.0 | 1.215 |
| 40 | ex49 | 431.3 | 0.966 |
| 41 | ex29 | 440.2 | 0.965 |
| 42 | ex69 | 447.4 | 0.999 |
| 43 | ex10 | 456.9 | 1.086 |
| 44 | ex50 | 463.6 | 0.955 |
| 45 | ex30 | 472.6 | 0.922 |
| 46 | ex70 | 479.6 | 1.000 |
| 47 | Xq28 | 485.8 | 1.371 |

# Need Assistance?

**Access the User Manual anytime by navigating to Help > Help**




**You can view webinars on GeneMarker here:**
**http://softgenetics.com/GeneMarker_reference.html**

**Or, Please Email:** tech_support@softgenetics.com

**Or Please Call:** 814-237-9340

# GeneMarker

*The Biologist Friendly Software*