HUMAN HEALTH

ENVIRONMENTAL HEALTH

# PERKINELMER ARRAY SOFTWARE SOLUTIONS

## GENOGLYPHIX SOFTWARE

PerkinElmer Array Software Solutions

Not for distribution in the USA

PerkinElmer®
*For the Better*

*Genoglyphix software is for Research Use Only.*
*Not for use in diagnostic procedures.*

# GENOGLYPHIX: TURNING 50,000 SAMPLES INTO MEANING

Genoglyphix® is a powerful data visualization software and database based on the analysis of over 50 000 validated cytogenetic samples.

Developed at Signature Genomics by Cytogeneticists for Cytogeneticists, Genoglyphix offers a proven analysis solution with a complete workflow including sample tracking, aberration categorization, final report creation functionality, and data sharing options with other centres. It also offers the possibility to create user-specific database tracks. Genoglyphix provides intuitive data visualization and annotation features for streamlined, rapid and clear interpretation of CGX data.

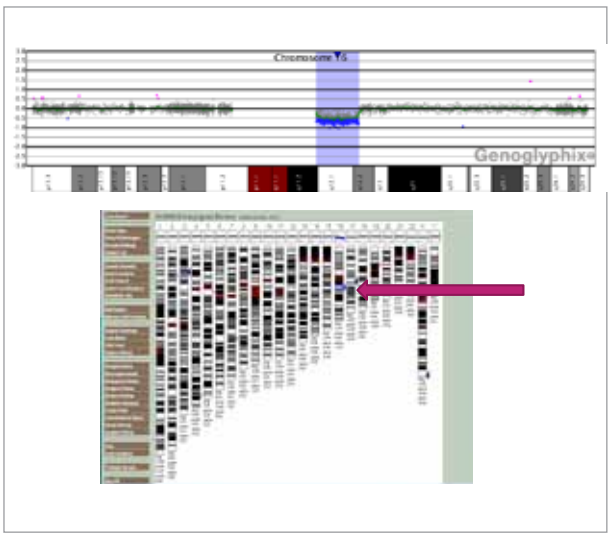
## Genoglyphix software – bringing multiple genetic databases into one tool

- **Access to Signature's database** containing over 12,000 validated genetic alterations identified in over 50,000 samples
- **Direct links to other relevant resources** such as the DGV, OMIM, PubMed, UCSC and Ensembl databases for easy data interpretation
- **Secure web based (128-bit encryption)** access to Genoglyphix database and software

## Genoglyphix guides cytogeneticists through the analysis with intuitive and easy-to use tools

- Generation of user-defined databases and custom tracks displaying abnormal results, copy number variants and analysis notes
- Report creation functionality
- Optional sharing of data with other Genoglyphix users
- Availability of FISH probes for result confirmation

**Figure 1: Case Review - Karyogram View**




Data visualization in Genoglyphix in Chromosome and Karyogram View

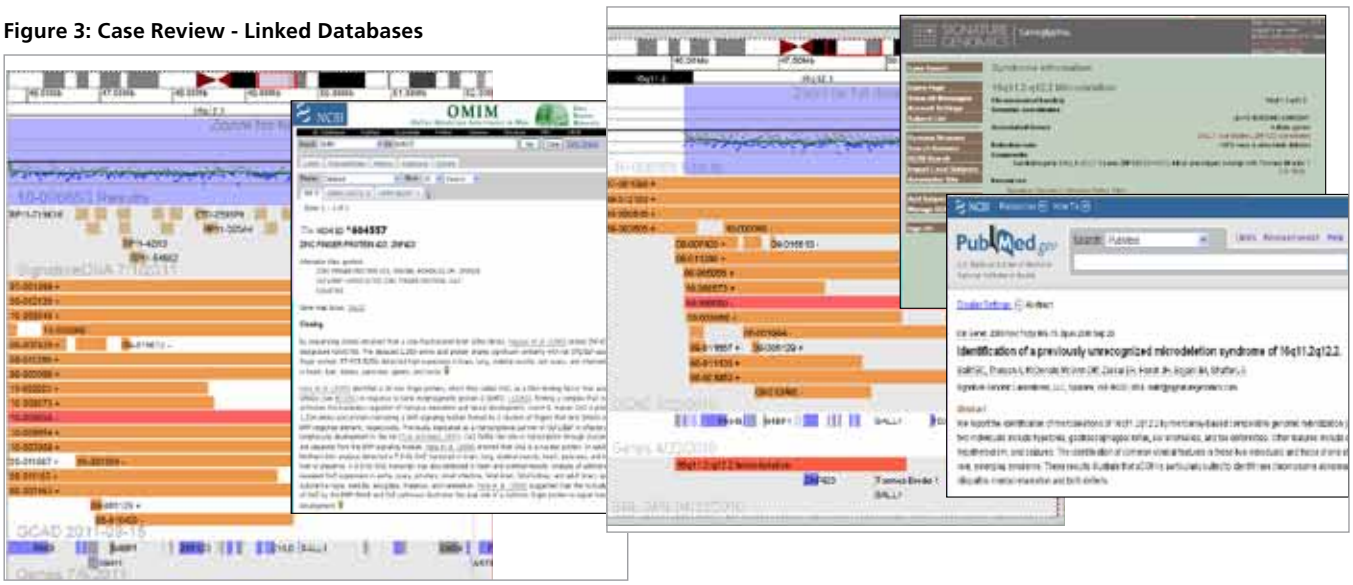
**Figure 2: Case Review - Genoglyphix Genome Browser**


< Graphical representation of aberrations in the Genoglyphix Genome Browser

< Identify Signature BAC clones available for FISH confirmation

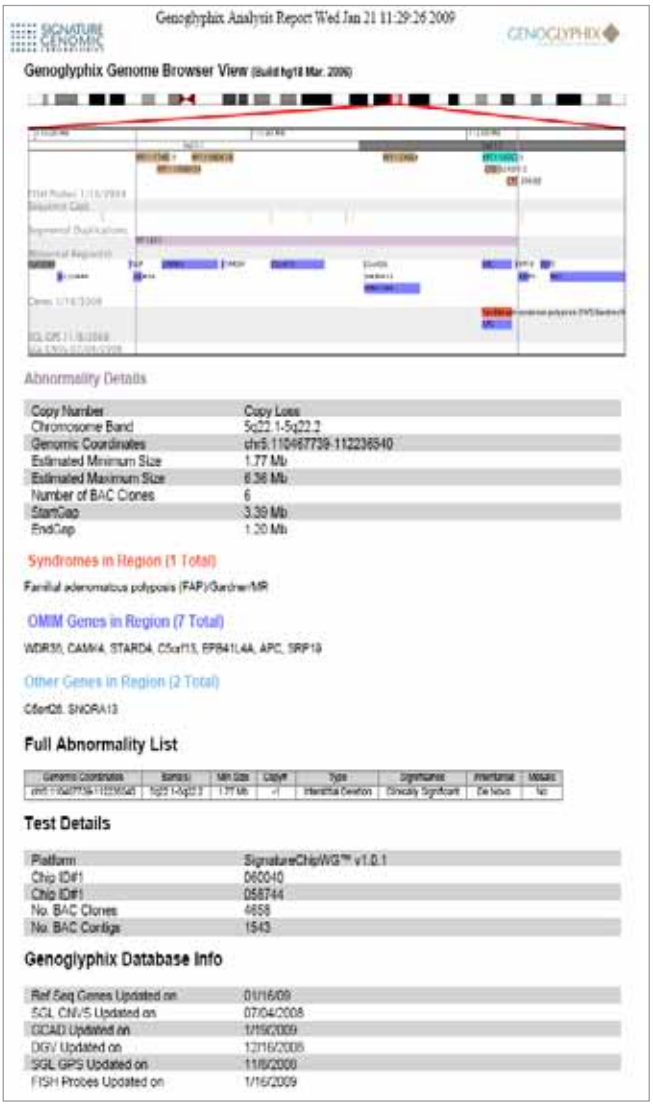
< Identify similar aberrations in Signature Genomics database of over 12,000 aberrations

**Figure 3: Case Review - Linked Databases**




Selection of the appropriate entries will automatically take you into databases such as OMIM, PubMed, and Genoglyphix Syndrome Detail Page

**Figure 4: Final Report**



Genoglyphix Analysis Report Wed Jan 21 11:29:26 2009

Genoglyphix Genome Browser View

Abnormality Details

| | |
|---|---|
| Copy Number | Copy Loss |
| Chromosome Band | 5q22.1-5q22.2 |
| Genomic Coordinates | chr5:110467739-112236540 |
| Estimated Minimum Size | 1.77 Mb |
| Estimated Maximum Size | 6.36 Mb |
| Number of BAC Clones | 6 |
| StartGap | 3.39 Mb |
| EndGap | 1.20 Mb |

Syndromes in Region (1 Total)

Familial adenomatous polyposis (FAP)/Gardner/MR

OMIM Genes in Region (7 Total)

WDR35, CAMK4, STARD4, C5orf13, EPB41L4A, APC, SRP19

Other Genes in Region (2 Total)

C5orf26, SNORA13

Full Abnormality List

| Genomic Coordinates | Band(s) | Min Size | Copy# | Type | Significance | Inheritance | Mosaic |
|---|---|---|---|---|---|---|---|
| chr5:110467739-112236540 | 5q22.1-5q22.2 | 1.77 Mb | <1 | Interstitial Deletion | Clinically Significant | De Novo | No |

Test Details

| | |
|---|---|
| Platform | SignatureChipWG™ v1.0.1 |
| Chip ID#1 | 060040 |
| Chip ID#1 | 058744 |
| No. BAC Clones | 4858 |
| No. BAC Contigs | 1543 |

Genoglyphix Database Info

| | |
|---|---|
| RefSeq Genes Updated on | 01/16/09 |
| SGL CNV5 Updated on | 07/04/2008 |
| GCAD Updated on | 1/15/2009 |
| DGV Updated on | 12/16/2008 |
| SGL GPS Updated on | 11/6/2008 |
| FISH Probes Updated on | 1/16/2009 |

## Genoglyphix Workflow

**1.** Once CGX array results are uploaded, the data can be displayed either within whole-genome or chromosome specific graphs, or alternatively, by use of the Karyogram view (Figure 1).

**2.** Selecting aberrations of interest will automatically upload the Genoglyphix Genome browser (Figure 2). From here, different data bases such as OMIM, RefSeq, PubMed, DGV, as well as Signature's Genoglyphix Chromosome Aberration Database (GCAD) can be directly accessed (Figure 3). This database contains over 12,000 abnormal cases that can be viewed in context of the selected sample.

**3**. Data can be shared between users and can be made available as a community resource. The information will be displayed as a separate track within the Genoglyphix Genome Browser.

**4.** Interpretation of each abnormality within a DNA sample can be carried out within the Notation Review by classifying for example the aberration type, its clinical importance, and inheritance.

**5.** The Final Review and Report option offers the possibility to summarize all classified aberrations and report those within their genomic context. FISH information can be uploaded to be displayed within the same report (Figure 4).

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