

SoftGenomics[®]

Computer software consisting of a suite of applications that support ordering, testing, documentation, and reporting in laboratories that perform testing using cytogenetic and molecular technologies, including chromosome analysis and microarrays, with instrument and foreign system interfacing capabilities and Web-based online ordering, reporting, and collaboration functionality.



FEATURES AND BENEFITS

Feature: Variant/Mutation Table and Gene Master Table

Benefit: The functionality supports generation of the laboratory's variant data base, categorization of variants, versioning, and linkage to relevant databases (OMIM, NCBI, etc.) for ease of variant entry, management, and interpretation. The Gene Master table allows users to capture information from other Internet databases for variant interpretation on a single gene level.

Feature: NGS reporting provides easy identification of the reportable variants into distinct category sections (reflecting states of pathogenicity, whether the associated disorder is medically actionable, and whether the finding is related to the patient's phenotype). Hyperlinks can be included for pertinent websites associated for each of the variants. Web-based reporting is available as well as options for various levels of reports.

Benefit: Users will be able to provide their clients clear and concise reports representing only the results opted for and with easy access to pertinent Internet information.

Feature: Seamless interfacing with sequencing and fragment analysis instruments

Benefit: Replacing manual data result entry with instrument interfaces eliminates the resource investment and inherent error rate associated with manual entry, thus providing greater efficiency and accuracy.

Feature: Rules-driven alerts or notifications based on client, patient demographic, test, or result parameters

Benefit: Reduce paper or telephone communications ensuring timely and accurate delivery of special instructions or critical information. Reduce risk of errors and inefficiencies that can be associated with frequent personnel interruptions without interrupting personnel at inopportune times.

SAMPLE SCREENSHOTS

NGS Variant/Mutation Table

SoftGenomics supports NGS (panels, WES, and WGS) and includes the Variant/Mutation Table, which presents the variant/mutation from the internal database, as well as hyperlinks to relevant Internet databases or websites that contributed to the interpretation. Versioning of the information is also available allowing users to track the available information used for each interpretation in cases where new information impacts the variant category or the interpretation.

The screenshot shows a detailed view of a variant. Key fields include:

- Nucleotide Change: c.2900C>T
- Amino Acid Change: p.A89V
- Location: 4
- Zygosity: Hem
- Position: ChrX:153296472
- RefSeq: NM_004992
- Reference: PMID:15557528

 An interpretation message states: "Whole exome sequencing has identified a hemizygous c.2900C>T (p.G289V) mutation affecting exon 4 of the MECP2 gene. This mutation has been previously reported both in patient with c of Fan, Am J Hum Genet 85:1520-1595; PMID:15573905, and also in a male patient with early-onset encephalopathy (V. Leuz, Neurology 63:1969-2004; PMID:1557528)."

Test Name	Status	Reported	Version
MOL-13-152	Signed Out	2	2
MOL-13-152	Whole Exome Sequencing	Signed Out	3
MOL-13-154	Whole Exome Sequencing	Unsigned	2
MOL-13-154	Whole Exome Sequencing	Unsigned	3
MOL-13-156	Whole Exome Sequencing	Unsigned	2
MOL-13-156	Whole Exome Sequencing	Unsigned	3
MOL-13-158	Whole Exome Sequencing	Unsigned	2
MOL-13-158	Whole Exome Sequencing	Unsigned	3
MOL-13-160	Whole Exome Sequencing	Unsigned	2
MOL-13-160	Whole Exome Sequencing	Unsigned	3

Tasklist Resulting

Analysis functionality combines patient results with the information in the Gene Master Table and Variant/Mutation Table for ease of interpretation. A direct link provides easy bridging between tables. The tasklist supports PCR, qPCR, array, MLPA, fragment analysis, and various sequencing testing.

The screenshot displays a tasklist with the following columns: MARK, Gene, Location, NT Change, AA Change, State, Web Ref, Mit Effect, Coverage, Category, Legacy Name. The table lists various tests and their results, such as:

- MARK: MOL-12-14, Gene: E1, Location: c.50dupT, AA Change: p.Ser18Gln>X27, State: het, Mit Effect: 6% (80) / 5% (100), Coverage: 6% (49) / 36% (242), Category: benign.
- MARK: MOL-12-14, Gene: E1, Location: c.132-1delK, AA Change: het, Mit Effect: 8% (179) / 4% (114), Coverage: 8% (112) / 7% (217), Category: benign.
- MARK: MOL-12-14, Gene: E2, Location: c.164delC, AA Change: p.Lys52Asn>X39, State: het, Mit Effect: 9% (230) / 7% (262), Coverage: 4% (108) / 6% (157), Category: benign.
- MARK: MOL-12-14, Gene: E3, Location: c.233dupT, AA Change: p.Trp78>Leu>X32, State: het, Mit Effect: 4% (130) / 18% (515), Coverage: 6% (207) / 6% (166), Category: benign.
- MARK: MOL-12-14, Gene: E6, Location: c.639delG, AA Change: p.Leu214>Terf>X1, State: het, Mit Effect: 47% (1472) / 48% (111), Coverage: 9% (288) / 16% (164), Category: benign.
- MARK: MOL-12-14, Gene: E7, Location: c.832delK, AA Change: p.Glu278>Leu>X7, State: het, Mit Effect: 3% (85) / 6% (87), Coverage: 43% (595) / 40% (543), Category: benign.
- MARK: MOL-12-14, Gene: E8, Location: c.850delA, AA Change: p.Met281>Terf>X1, State: het, Mit Effect: 25% (588) / 6% (199), Coverage: 45% (1108) / 49% (141), Category: pathogenic, Legacy Name: N143K.
- MARK: MOL-12-14, Gene: E8, Location: c.1040G>C, AA Change: p.Arg347>Pro, State: het, Mit Effect: 21% (187) / 3% (16), Coverage: 6% (80) / 5% (100), Category: benign.
- MARK: MOL-12-14, Gene: E8, Location: c.870>5delT, AA Change: het, Mit Effect: 6% (49) / 36% (242), Coverage: 8% (179) / 4% (114), Category: benign.
- MARK: MOL-12-14, Gene: E8, Location: c.942delK, AA Change: p.Pha315>Ser>X13, State: het, Mit Effect: 43% (595) / 40% (543), Coverage: 25% (588) / 6% (199), Category: benign.
- MARK: MOL-12-14, Gene: E11, Location: c.1408G>A, AA Change: p.Val470>Met, State: rs213950, Mit Effect: 45% (1108) / 49% (141), Coverage: 21% (187) / 3% (16), Category: benign.
- MARK: MOL-12-14, Gene: E11, Location: c.1633delT, AA Change: p.Ser492>Pro>X32, State: het, Mit Effect: 6% (80) / 5% (100), Coverage: 6% (49) / 36% (242), Category: benign.
- MARK: MOL-12-14, Gene: E1, Location: c.164delC, AA Change: p.Lys52Asn>X39, State: het, Mit Effect: 8% (179) / 4% (114), Coverage: 8% (112) / 7% (217), Category: benign.
- MARK: MOL-12-14, Gene: E2, Location: c.166delA, AA Change: p.Lys52Asn>X39, State: het, Mit Effect: 9% (152) / 7% (217), Coverage: 4% (108) / 6% (157), Category: benign.

FEATURES AND BENEFITS

Feature: Client-editable ISCN dictionary

Benefit: Selection of results for the most common chromosome abnormalities eliminates manual typographical error and assures consistent resulting.

Feature: Linking of family members

Benefit: Easy reviewing, correlating, and referencing of familial rearrangements in reports

Feature: Full patient testing history access from current file

Benefit: Easy reviewing and correlating of both historical and concurrent testing results and reports supports accurate ordering and interpretation.

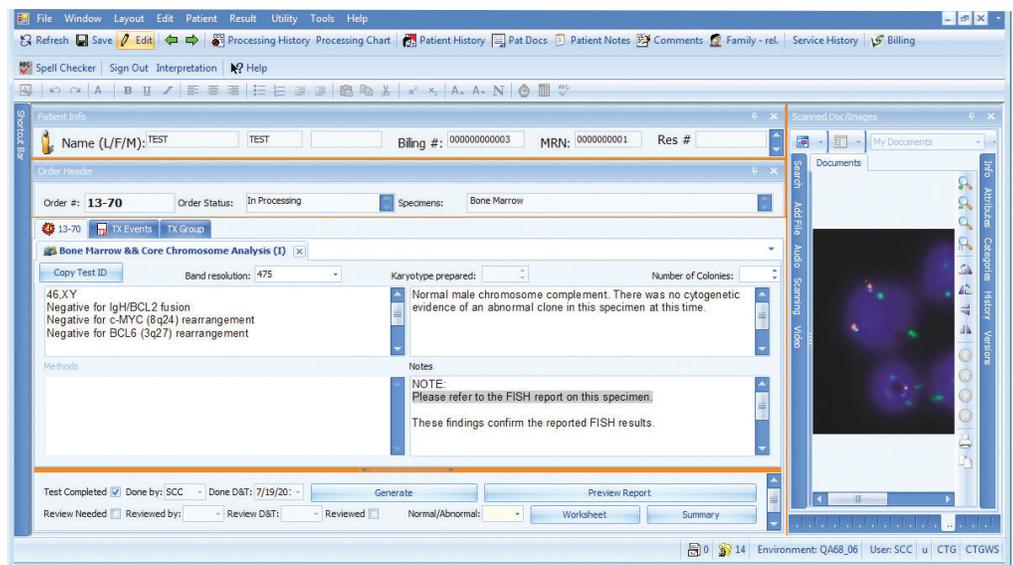
Feature: Auto reflex testing across technologies based on client-defined rules

Benefit: Eliminates manual error inherent in manual reflex ordering and reduces turnaround time of reflex tests.

SAMPLE SCREENSHOTS

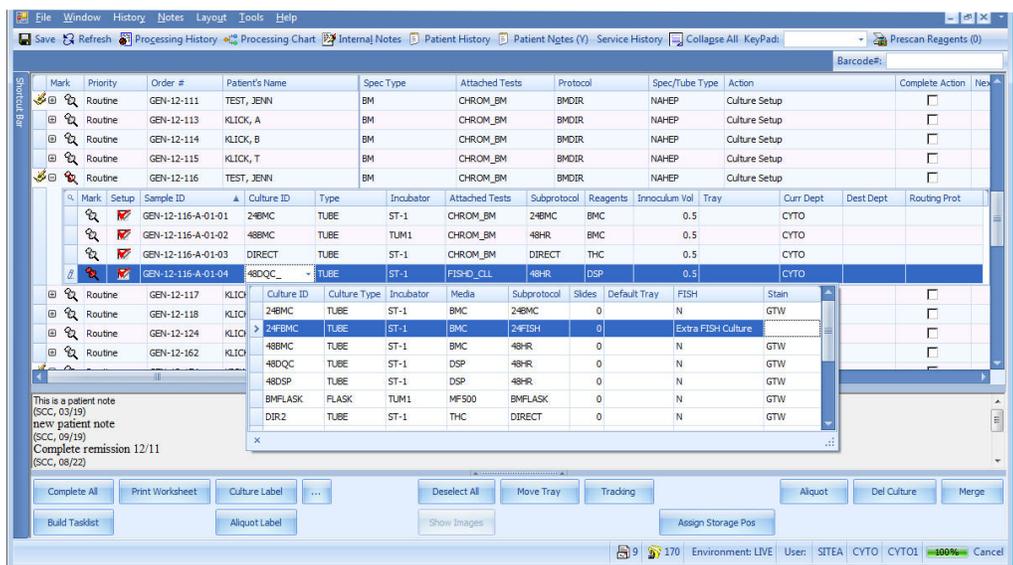
Interpretation Result Entry window

The Interpretation Result Entry interface provides users with a patient's previous and concurrent order information as well as previous or concurrent order information on family members. Interpretation generation can be driven by automated reporting rules using canned messages, manual insertion of canned messages, or free text editing.



Setup window

During culture setup, the technologist can easily print labels, assign media, add and remove cultures, and create aliquots as the system records critical documentation including the user identification, the date, and the time.



FEATURES AND BENEFITS

Feature: Online ordering now enhanced with Wizard functionality that provides real-time information on incoming volumes, ensures that required data is provided, automatically prompts the ordering party at accessioning entry with appropriate test ordering guidance, and enables users to request any unique additional information required for specific testing.

Benefit: This functionality reduces resource needs in accessioning, eliminates delays experienced with incomplete paper requisitions, cuts costs associated with manual follow-up, and supports optimized patient care while reducing unnecessary testing.

Feature: SoftGenomics online notification capabilities replace phone and fax communications. Notifications can consist of the following: status updates of test results, online availability of results or reports, reflex or adjunct test guidance, hyperlinks to relevant journal articles or policy statements, and even forms such as required consent or add-on test attestation forms.

Benefit: Users can provide real-time notifications without delays often imposed by office hours or personnel availability. Efficiency gains and cost savings in clerical areas.

Feature: Patient Portal – Report access

Benefit: Patients can access their reports in compliance with the patient access law, recently enacted by the Department of Health and Human Services (HHS).

SAMPLE SCREENSHOTS

Online Ordering interface

The Online Ordering interface can be specifically configured for users and their clients' needs with as many template versions as desired. Required fields are user defined, and SCC's rules-based system allows users to trigger prompts based on user-defined criteria. These prompts can contain test ordering suggestions, educational links, or requests for additional information.

Interpretation Entry

Online connectivity supports remote consultations and technical component/professional component (TC/PC) split test options. Easy, HIPAA-compliant access to test results for remote or external physician review.

Crossmatch	Marker Name	Value	Unit	Panel	Run ID	Acquisition Date	Reportable	Include in Trend
CELLULARITY	Cellularity	435464		0			Y	N
D_GRANGATE	Granulocyte/Myeloid Gate	44	%	0			Y	N
D_LYMPHGATE	Lymphocyte Gate	434	%	0			Y	N

Phenotype
PHENOTYPE

Morphology
MORPHOLOGY
This interpretation is based on the clinical information provided and the current understanding of the molecular genetics of this condition. Although DNA-based testing is highly accurate, rare diagnostic errors may occur. Examples include misinterpretation because of genetic variants, blood transfusion, bone marrow transplantation, or erroneous representation of family relationships or contamination of a fetal sample with maternal cells.

Comment
COMMENT:
Genetic counseling is recommended to discuss the potential clinical and/or reproductive implications of this result, as well as recommendations for testing other family members and, when applicable, this individual's partner.

Interpretation
INTERPRETATION
This individual is a carrier of CF.
Order Number: MGS-13-0000004 Signed By Code: SCC D&T: 04/10/2013 05:05

Interpretation
The tumor is consistent with the previously diagnosed tumor.
A low-grade lymphoproliferative disorder cannot be excluded on the basis of morphology alone.
Pathologists needs to be consulted for this specific
Partial mastectomy

SCC Soft Computer
5400 Tech Data Drive
Clearwater, Florida 33760
(727) 789-0100
sales@softcomputer.com