

SoftMolecular®-NGS



SoftMolecular—SCC's intuitive workflow solution for the diagnostic molecular genetics laboratory—enables geneticists and pathologists to create customized protocols for managing testing workflows, derive data and images from automated instruments by interfaces, auto-populate report text based on result-driven reporting rules, and easily access patient and family medical and testing histories.

SoftMolecular empowers pathologists, geneticists, and forensic scientists to automate workflow and manage data and analysis stemming from current and future molecular testing methods in order to generate accurate interpretations and meaningful reports with minimized effort. This advanced genetics LIS platform supports both clinical and research applications of Next Generation Sequencing (NGS) including Targeted Panels, Whole Exome Sequencing (WES), or Whole Genome Sequencing (WGS) as well as other common molecular technologies. Robust and user-friendly, SoftMolecular enables users to design and streamline workflows, interface with automated instrumentation, manage patient and family cases, track sample storage, and control inventory. As part of SCC's Genetics Information Systems Suite, SoftMolecular supports interfacing and communication with other laboratories and health agencies, and represents the most comprehensive suite of genetics LIS applications in the world.

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.

Benefit: This feature 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. 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: Images on reports.

Benefit: Images and graphics are available on patient reports providing professional and sophisticated reports for your clients and patients.

Feature: Ability to choose a re-run of an entire batch or auto-assign a specific sample to the next pending batch with a single mouse-click.

Benefit: This feature enables users to manage repeat testing quickly and easily to avoid any delays or mismanagement that can occur with manual processes. This ensures the most optimal turnaround time, thus enhancing client satisfaction.

Spotlight Feature:

This innovative software supports both clinical and research applications of Next Generation Sequencing (NGS) whether it involves Targeted Panels, Whole Exome Sequencing (WES), or Whole Genome Sequencing (WGS). Discrete structured genomic variant result reporting is now available for result reporting integration to EMR system.

FEATURES AND BENEFITS

- Feature:** Ability to link controls to a sample or batch.
- Benefit:** Control documentation will be accurate, can be accessible per patient or batch, and will remain electronically linked for historical reference or investigation. Users will no longer need to sort through manual logs or files to identify or trend control data for quality control troubleshooting.
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- Feature:** Auto-calculation of master mixes with auto-deduction of volume from inventory.
- Benefit:** Reduction of manual calculations and inventory control leading to error-free calculations and effective inventory management.
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- 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.
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- Feature:** Auto reflex and auto rerun testing across technologies based on client-defined rules.
- Benefit:** Eliminates manual error inherent in manual reflex/rerun ordering and reduces turnaround time of tests.

SAMPLE SCREENSHOTS

NGS Variant/Mutation Table

SoftMolecular 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 displays the 'Variants/Mutation Table' interface. The main table lists variants with columns: Gene, Variant, Inheritance, Type, and various scores. A 'Properties' panel on the right shows details for a selected variant, including its title, location, and associated publications. The bottom section shows a list of previously loaded variants with columns: Order #, Sample ID, Title ID, Patient, and various scores.

Gene	Variant	Inheritance	Type	Score
BRCA2	157046	AD	DEL	1000000
BRCA2	157046	AD	DEL	1000000
BRCA2	157046	AD	DEL	1000000

Properties panel:

Registration ID: 157046-2019-01-19

Publications:

Pub Med ID	Title	Location	Publication Type	Authors	Publication Date	URL
157046	Characterization of BR...	BRCA2	Journal Article	Briggs, A., Hsieh, R. H. L., ...	2019	http://www.ncbi.nlm.nih.gov/pubmed/30812450
157046	The founder mutation ...	BRCA2	Journal Article	Radman, D., Kessler, ...	2007	http://www.ncbi.nlm.nih.gov/pubmed/17333
157046	Breast and ovarian ca...	BRCA2	Journal Article	Friedman, S. S., Rubin, ...	2012	http://www.ncbi.nlm.nih.gov/pubmed/22554
157046	Double heterozygotes ...	BRCA2	Journal Article	Frederick, C. B., Gad, ...	1998	http://www.ncbi.nlm.nih.gov/pubmed/9500000
157046	Functional evaluation ...	BRCA2	Journal Article	Hsu, K., Newman, S. R., ...	2005	http://www.ncbi.nlm.nih.gov/pubmed/157046

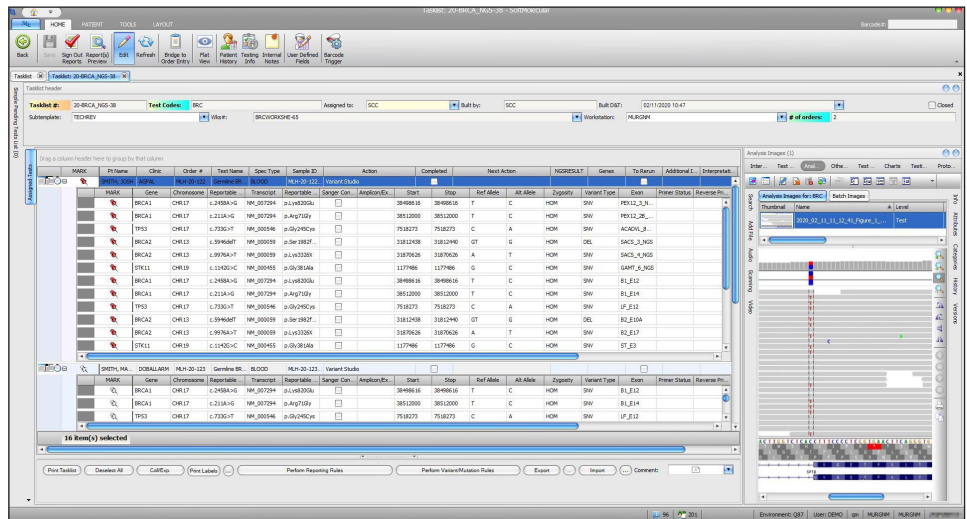
Previously loaded variants:

Order #	Sample ID	Title ID	Patient	Variant	Score	Reported	Transcript	Location	Category	Status
1	157046	157046	157046	157046	1000000	1	Reported	BRCA2	AD	Signed Out
2	157046	157046	157046	157046	1000000	1	Reported	BRCA2	AD	Signed Out
3	157046	157046	157046	157046	1000000	1	Reported	BRCA2	AD	Signed Out
4	157046	157046	157046	157046	1000000	1	Reported	BRCA2	AD	Signed Out
5	157046	157046	157046	157046	1000000	1	Reported	BRCA2	AD	Signed Out
6	157046	157046	157046	157046	1000000	1	Reported	BRCA2	AD	Signed Out
7	157046	157046	157046	157046	1000000	1	Reported	BRCA2	AD	Signed Out
8	157046	157046	157046	157046	1000000	1	Reported	BRCA2	AD	Signed Out
9	157046	157046	157046	157046	1000000	1	Reported	BRCA2	AD	Signed Out
10	157046	157046	157046	157046	1000000	1	Reported	BRCA2	AD	Signed Out

SAMPLE SCREENSHOTS

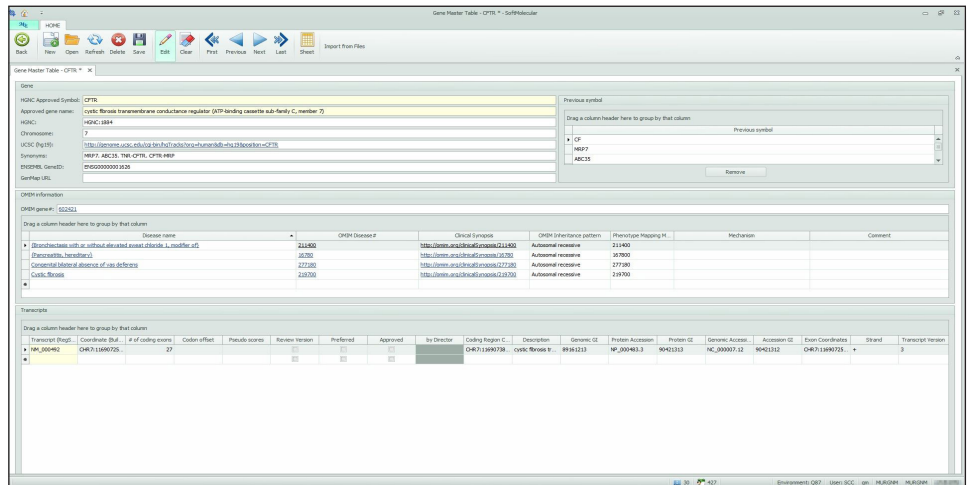
Tasklist Resulting

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.



NGS Gene Master Table

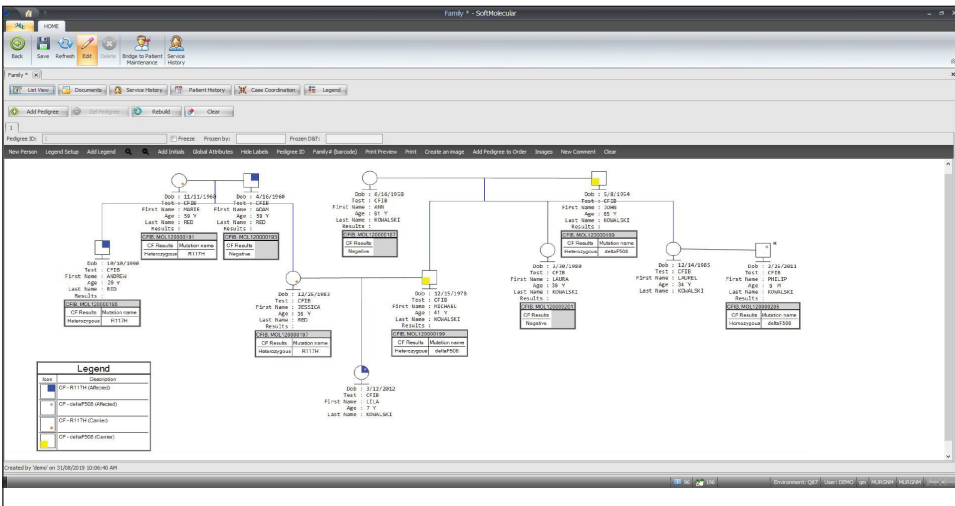
This table provides at-a-glance information (transcripts, associated diseases, etc.) on exons for a given gene and provides a repository of relevant data from internet sources including links to these sources.



SAMPLE SCREENSHOTS

Family Pedigrees

SCC's Genetics Information System Suite Pedigree functionality allows linking of family members based on their relationships. User-defined attributes such as carrier or affected status, mutations, and phenotypic traits can be captured and displayed within the pedigree.



Trending Graphs

SoftMolecular offers an extensive library of trending chart templates that can be used to create customized graphs. Users can select the attributes/fields to be included, the number of recent values, and the scale of the graph. SoftMolecular's flexibility enables users to combine different result values on one graph for comparison purposes or have multiple graphs for a single test.

