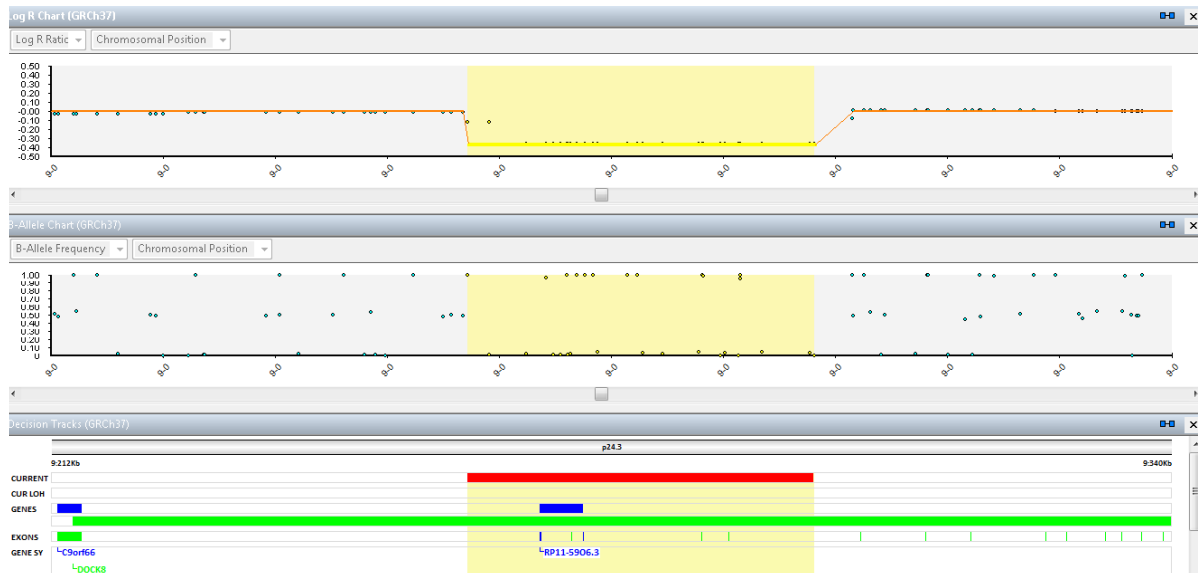


Figure 4: Visualization of a Gene Deletion



A deletion within the DOCK8 gene covers three exons.

Clear Visualization for Rapid Data Confirmation

The key to rapid, confident results interpretation is clear, unambiguous data presentation. In the BlueFuse Software, data can be viewed horizontally or vertically (classical cytogenetics view). The opening view shows the entire genome with the automated calling displayed, facilitating user confirmation of the calls. The user can zoom in to any region on the genome for more detailed analysis at any time by selecting the desired region or abnormality. The user may also annotate the data manually for segmental changes or override the automated calls. The region comments box supplements any changes with an explanation, enabling an audit trail and case history and facilitating the addition of any other relevant information used in the analysis (Figure 2).

Intuitive Navigation

BlueFuse Software links all data for each view, making navigation as simple as a click of the mouse. For example, click a region in a chart to see specific information in a table format, or click a line in a table to see that data highlighted in the charts (Figure 3).

Helpful Data Interpretation

In addition to clear data presentation, BlueFuse Software helps users understand the significance of any detected changes. This is achieved through the import of a secondary file called the annotation database. The annotation database is comprised of summary information from the key public databases (ISCA, DGV, Ensembl, and Decipher) and is updated regularly. To make sure that the most recent information is used, BlueFuse Software facilitates direct link-out to the live websites for region, gene, and OMIM searches, simply by right-clicking on the

relevant region in the decision track. Laboratories can import custom BED or TXT files to supplement this information. Visualization features in the decision tracks enable data comparison between current experiments and published regions, and a closer look at gene and exon content within each region (Figure 4).

Full History of Laboratory Data

BlueFuse Software incorporates a number of measures to ensure a transparent audit trail of laboratory data and decisions made regarding data interpretation.

Record of Data Assessment

The Region View dialog within BlueFuse Software provides a laboratory record of the decisions made for every region identified. Every region can be assigned an assessment of Benign, Probably Benign, Unknown, Probably Pathogenic, and Pathogenic, following ISCA guidelines. Users can add specific comments regarding the assessment, supplying additional support of the decision. This non-deletable audit trail delivers a history of analysis and a paperless format of case management between users. All information appears on the generated report (Figure 5).

Sample and QC Comparison

Complete information for all samples is stored in a central database. A summary of all regions found in all experiments, including frequency of detection and previous region assessments, is available to guide decisions. Data are permanently available and can be revisited and interrogated at any time ensuring that any follow-up questions can be readily answered. QC metrics for each experiment are also stored

AAAGAATGATAACAGTAACACACTTCTGTTAAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAAGAGTACCGTCTTCTGTTAAACCTTAAGATTACTTGATCCACTGATTCA
AATCAACGTACCGTAACGAACGTATCAATTAAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAAGAGTACCGTCTTCTGTTAAACCTTAAGATTACTTGATCCACTGATTCA
AACGACGAAAGAATGATAACAGTAACACACTTCTGTTAAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAAGAGTACCGTCTTCTGTTAAACCTTAAGATTACTTGATCCACTGATTCA
TTAAAGTACCATTAAGAGCTACCGTCAACAGTAACACACTTCTGTTAAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAAGAGCTACCGTCAACGACGAAAGAATGAT
AAAGAAATGATAACAGTAACACACTTCTGTTAAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAAGAGTACCGTCTTCTGTTAAACCTTAAGATTACTTGATCCACTGATTCA
AAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAAGAGTACCGTCTTCTGTTAAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTTACCG
AACGTATCAATTGAGACTAAATATTAACGTACCATTAAAGAGTCTGTTAAACCTTAAGATTACTTGATCCACTGATTCAACGTACCGTAACGAACGTATCAATTGAGACTAAATATTAACGTACCATTAAAGAGTACCGTCTTCTGTTAAACCTTAAGATTACTTGATCCACTGATTCA

