VUmc Cancer Center Amsterdam

Implementation of a copy number analysis pipeline for shallow sequencing in Galaxy framework

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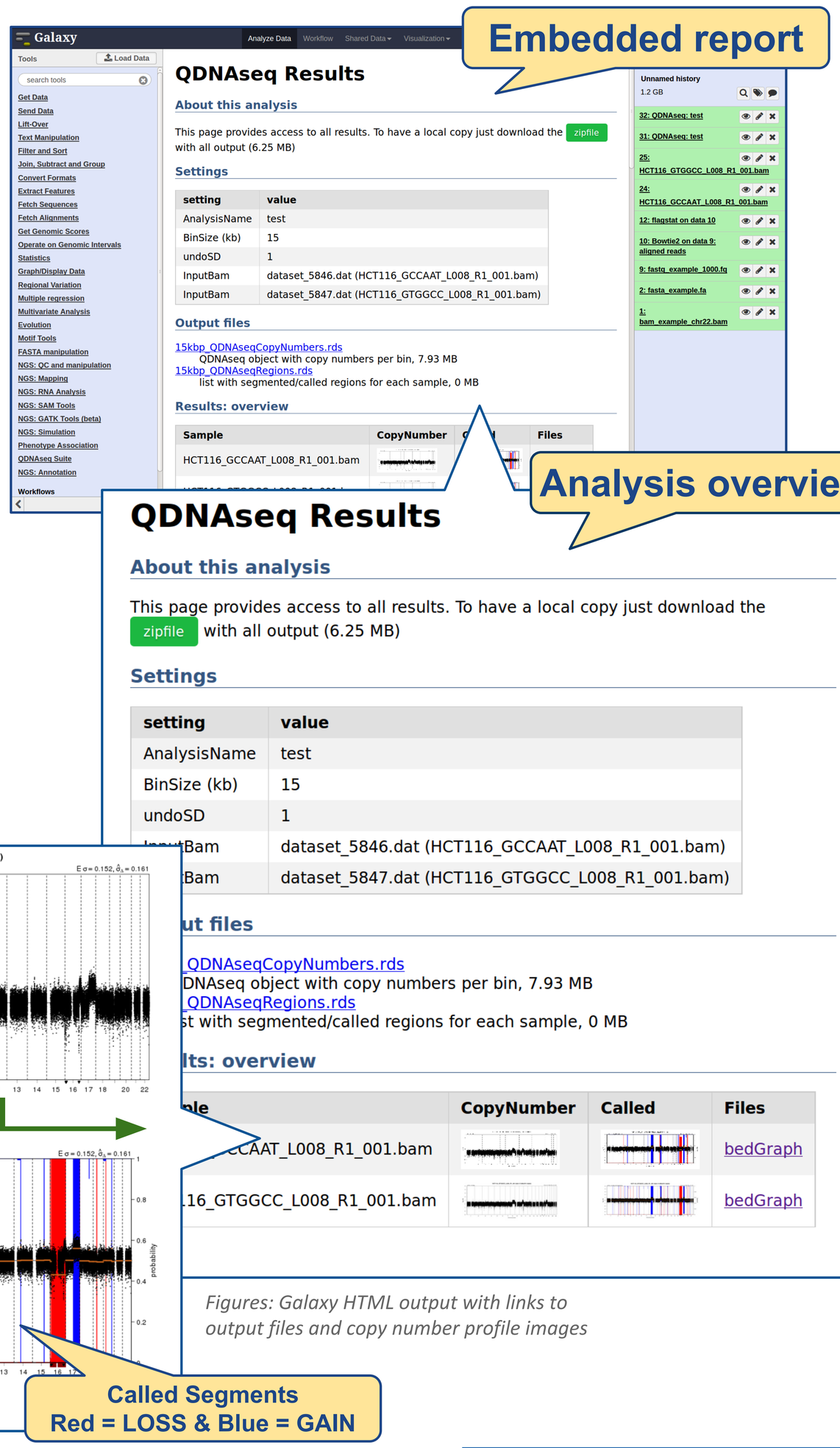
Introduction

DNA copy number aberrations are a hallmark of cancer and can be quantified by **shallow whole-genome sequencing** (WGS). A robust method has been developed [1] that detects copy number aberrations. This method is currently being integrated into the **Galaxy platform** [3] within the Center for Translational Molecular Medicine - Translational Research IT (CTMM TraIT) project [4].

Methods

Sequence reads are **binned and counted in non overlapping windows** of 15kb and a combined LOESS correction for mappability and GC content is applied. Then a **comprehensive filtering** excludes genomic regions from both ENCODE project blacklists and a novel blacklist we have developed based on sequence depth of 38 individuals from the 1000 Genomes project. Procedures for copy number detection have been **optimized for** use in combination with DNA isolated from **formalin-fixed paraffin-embedded** (FFPE) samples.

- ✓ **highly concordant with array CGH** at considerably lower cost [1]
- ✓ also available as a **Bioconductor** package, QDNAseq [2]
- ✓ the popular **BAM format** as input
- ✓ reports results in a clear and concise **HTML based view** within Galaxy itself
- ✓ **R data structure** file for downstream analysis available
- ✓ Zipped archive with all output



Figures: Galaxy HTML output with links to output files and copy number profile images

References

1. I. Scheinin, D. Sie et al. DNA copy number analysis of fresh and formalin-fixed specimens by whole-genome sequencing: improved correction of systematic biases and exclusion of problematic regions (submitted).
2. http://www.bioconductor.org/
3. http://galaxyproject.org
4. http://www.ctmm-trait.nl/

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trait a ctmm project

decode a ctmm project

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