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PanelMaps: a web tool for detection and visualization of altered regions for targeted sequencing

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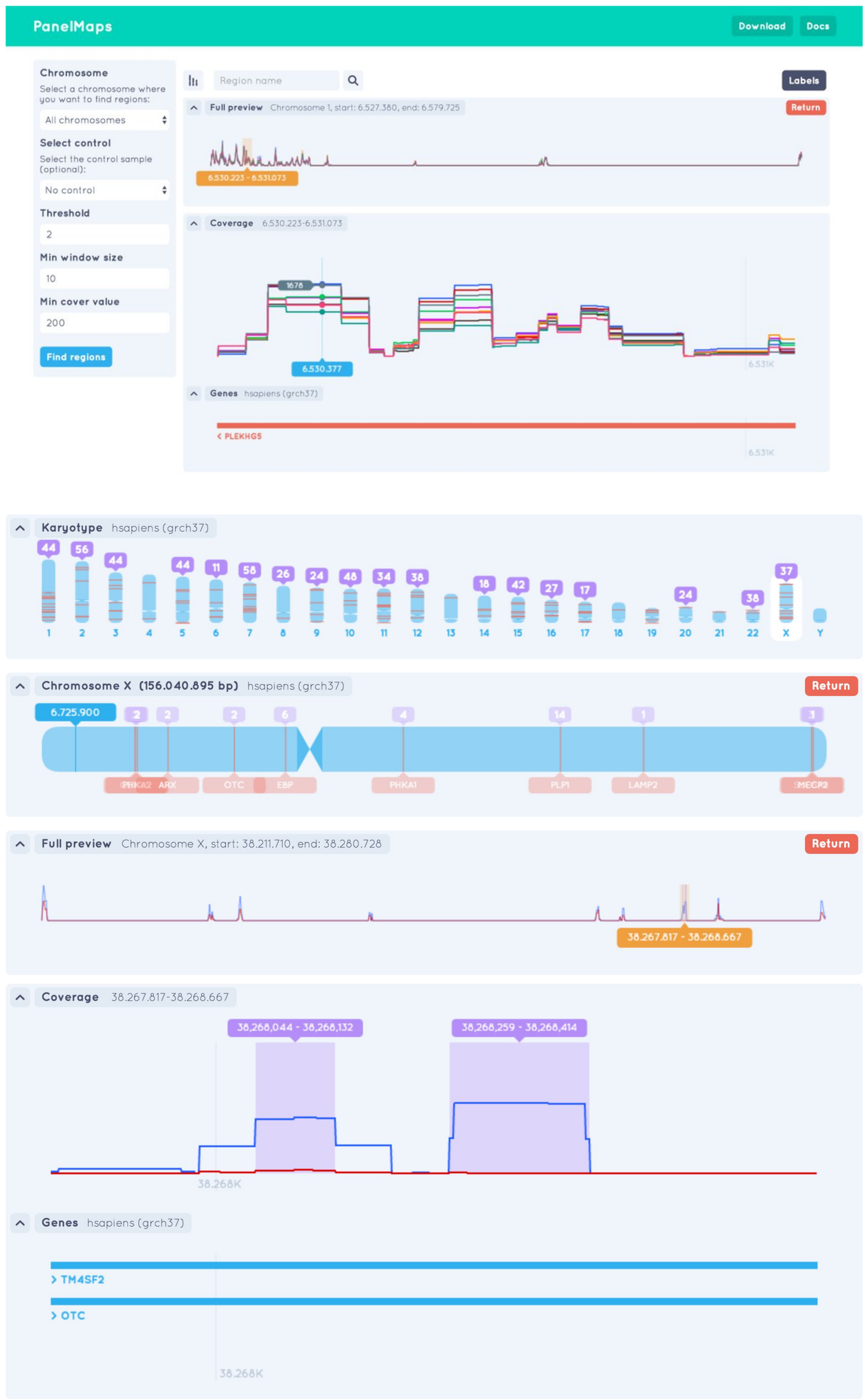
INTRODUCTION

Gene panel sequencing allow us to detect variants associated with different diseases. Sometimes, the possible cause of the disease is not due to variations in SNPs (Single Nucleotide Polymorphism) or INDELs (small insertions-deletions) and can be motivated by the presence of a larger variation: deletion or insertion.
The aim of this work is the design and development of a web tool for detection and visualization of altered regions from targeted sequencing data.

HOW DOES PANELMAPS WORK? http://panelmaps.juanes.xyz

- **Inputs**. One or several BAM files (one file for each individual) and a BED file including all regions for gene panel.
  After loading data, the coverage of each sample is calculated and these values are normalized between all samples considering the total number of reads of each sample.
- **Methods**. The tool includes two modules: visualization of genes or regions from coverage data and a module analysis to detect regions of interest. For detection of altered regions, PanelMaps uses a control sample selected by the user or combines all samples to get a common reference. Users can also specify comparisons between subgroups of samples of interest. The analysis incorporates a sliding window algorithm with various parameters adjustable by the user and related to the precision and characteristics of the region to be detected.
- **Outputs**. PanelMaps shows a graphical description of coverage levels for genes and samples to confirm that all regions are covered. This web tool visualizes all regions included in the panel and shows a selection of altered regions between samples.



CONCLUSIONS

Panelmaps is a useful tool for detection and visualization regions of genes altered in panels that improves the knowledge of the genetic basis of diseases and produces useful information for diagnosis in clinical contexts.
This web tool is an alternative to the use of molecular biology techniques such as MLPA (Multiplex Ligation-dependent Probe Amplification), which are very costly and sometimes have some technical problems such as failure to detect variants or micro-deletions in positions where primers are incorporated.

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