GENETICS, SOFTWARE

CNV (NOT LONDON) CALLING

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Having been asked now by several people, I am compiling here a list of programs can be used for CNV calling.

- QuantiSNP: is an analytical tool for the analysis of copy number variation using whole genome SNP genotyping data. In its first implementation it was developed for data arising from IlluminaÂ® platforms and this is fully described in Colella and Yau et al., 2007
- CNVFinder: The CNVFinder algorithm has been designed to detect copy number variants (CNVs) in human population from large-insert clone DNA microarray covering the entire human genome in tiling path resolution (WGTP platform).
- SNPtools:The purpose of this program is to provide analysis tools for and visual representations of array data. The program was initially developed for Affymetrix SNP arrays, but has subsequently been modified to allow other array types such as Affymetrix gene expression arrays, Illumina arrays and various types of CGH arrays. However, some features only work for SNP arrays in the current implementation.
- TriTyper: We present a new method (TriTyper) that can infer genotypes in case-control data sets for deletion CNVs, or SNPs with an extra, untyped allele at a high-resolution single SNP level. By accounting for linkage disequilibrium (LD), as well as intensity data, calling accuracy is improved.
- PennCNV: The PennCNV software is cross-platform compatible with appropriate compilation of source codes. However, you need a Windows machine to use the Illumina BeadStudio software to export the signal intensity values for analysis.
- SNPtrio: Single nucleotide polymorphism (SNP) data can provide information about genotypes and chromosomal copy number. SNPtrio offers an additional category of information by visualizing and analyzing inheritance patterns in trios (mother, father, and child).
- VanillaICE: Hidden Markov model for identifying chromosomal alterations in high--throughput SNP arrays
- GEMCA: The program that enables CNVs to be detected for a test sample compared to a reference sample or a reference set.