Publicacions més rellevants de la línia de recerca:
Software per dades genètiques quantitatives

Referéncia: Pique-Regi, R, Cáceres A, González, JR. R-Gada: a fast and accurate tool for analyzing copy number data. R-GADA project accesible via
`http://groups.google.com/group/gadaproject`.

Abstract: The `gada` package is the implementation of a flexible and efficient analysis pipeline to detect genomic copy number alterations from microarray data. The package can import the raw copy number normalized intensities provided by Illumina BeadStudio, Affymetrix powertools, or any similar format.Probes of different samples are split into separate files and can be analyzed on a standalone workstation or in parallel using a cluster/multicore computer. The speed and accuracy of the genome alteration detection analysis (GADA) approach combined with parallel computing results in one of the fastest and most accurate methods available. GADA is especially suitable to extract copy number alterations (CNAs) on genomewide studies that utilize high density arrays of millions of markers to sample hundreds of subjects.

Referència: Subirana, I, González, JR. CNVassoc: an R package to analyze CNV association studies. CNVassoc project accesible via
`http://www.creal.cat/jrgonzalez/software.htm`.

Abstract: `CNVassoc` allows users to perform association analysis between CNVs and disease incorporating uncertainty of CNV genotype. This document provides an overview on the usage of the `CNVassoc` package. We illustrate how to analyze CNV data by using some real data sets. The first data set belongs to a case-control study where peaks intensities of MLPA assays were obtained for two diferent genes. The second example corresponds to the Neve's dataset that is available at Bioconductor. The data consists on 50 CGH arrays of 1MB resolution for patients diagnosed with breast cancer. All datasets are available directly from the `CNVassoc` package.

Referència: Cáceres, A, Vilatoro, S, Armengol, S, González, JR. MLPAstats: a R GUI tool for

detecting altered genes using MLPA data. *J Stat Software* , In first revision

Abstract: MLPAstats is software, written in R, design for the analysis of differences in CNVs using multiplex-dependent probe amplification (MLPA) data. The software is provided with a graphical user interface (GUI) that facilitates its use, in particular, for those not familiar with R. Here we present a step-by-step analysis of the data provided by the United Kingdom National Genetics Reference Laboratory of Manchester and another example for rare diseases. It is freely available from http://www.creal.cat/jrgonzalez/software.htm.