

Genomic Variant Studies

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1 Copy number variants (CNV)

Log R Ratio (LRR): The LRR is the normalized measure of total signal intensity and provides information relative to copy number or the number of alleles present in a biological sample at one SNP. **B Allele Frequency (BAF):** BAF is derived from the ratio of between intensities of the allele probes, measuring the allele proportion or allele composition of the sample. In normal samples, the expected values are:

- 0 and 1 for homozygous genotypes (AA, BB)
- 0.5 for heterozygous genotypes (AB)

1.0.1 CNV calling

Segmentation of LRR values along the chromosome together with BAF information are used to identify the regions with probable copy number alterations.

```
# Import packages  
library(gada)
```