# Finding de novo copy number variants in case-parent trios for oral clefts 

Bioconductor Developers Meeting, Zurich, Switzerland

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December 13, 2012
father

offspring

offspring


False positives are problematic

## father



## minimum distance


()

MinimumDistance

## A de novo deletion

$\log _{2}$ ( R ratios) $\quad \mathrm{B}$ allele freqencies

$\log \{\operatorname{Pr}(332 \mid.) / \operatorname{Pr}(333 \mid)\}=.6257.8 / 4692.2$
$=1565.6$

## MinimumDistance

Scharpf RB, Beaty T, Schwender H, Younkin S, Scott AF, Ruczinski I: Fast detection of de novo copy number variants from SNP arrays for case-parent trios. BMC Bioinformatics 2012, 13:330.

Available from Bioconductor and collaboratively developed on GitHub (https://github.com/rscharpf/MinimumDistance ) with

- Sam Younkin
- Holger Schwender
- Ingo Ruczinski

