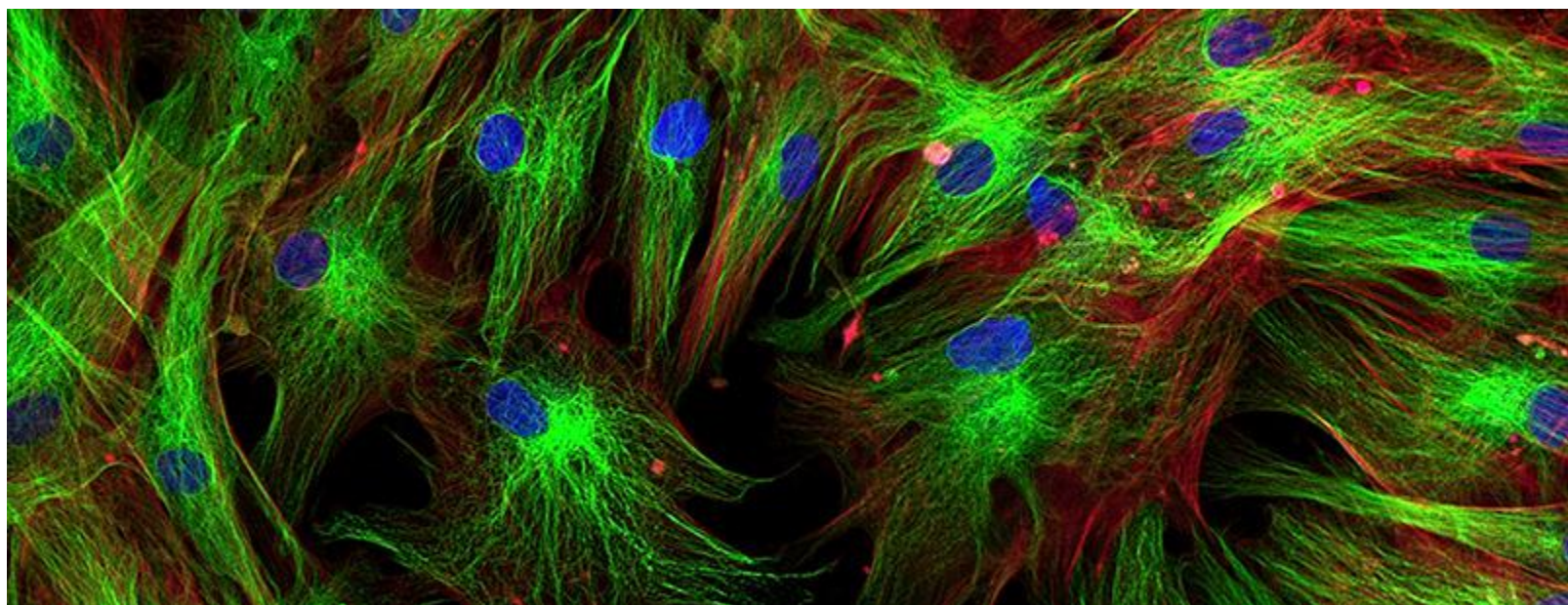
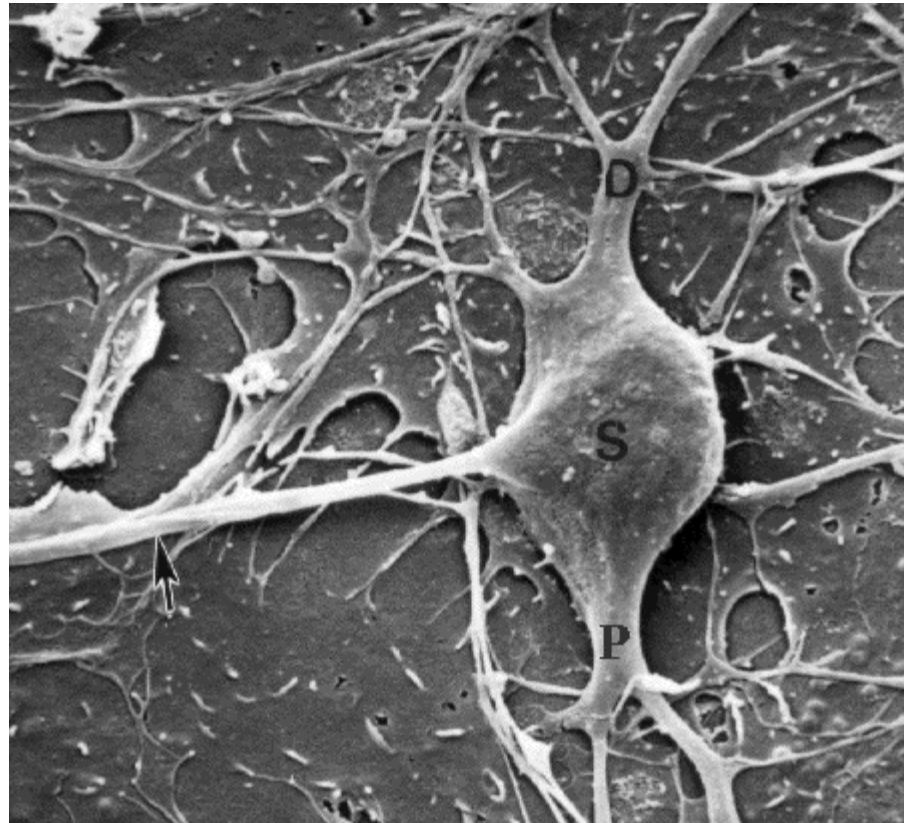


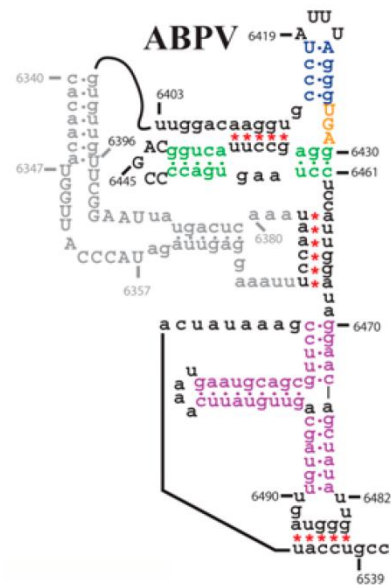
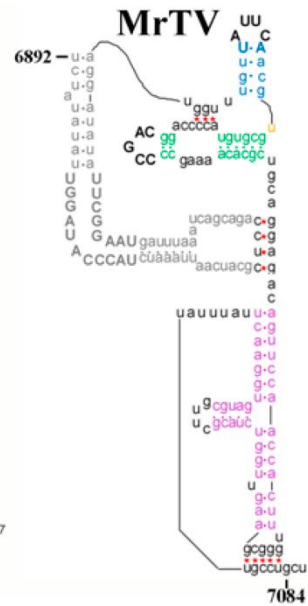
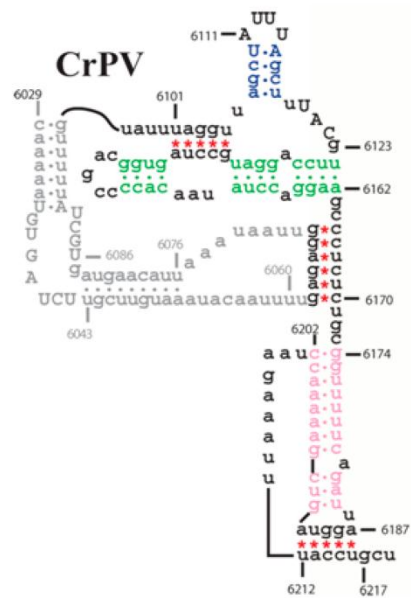
Plot Your Data!

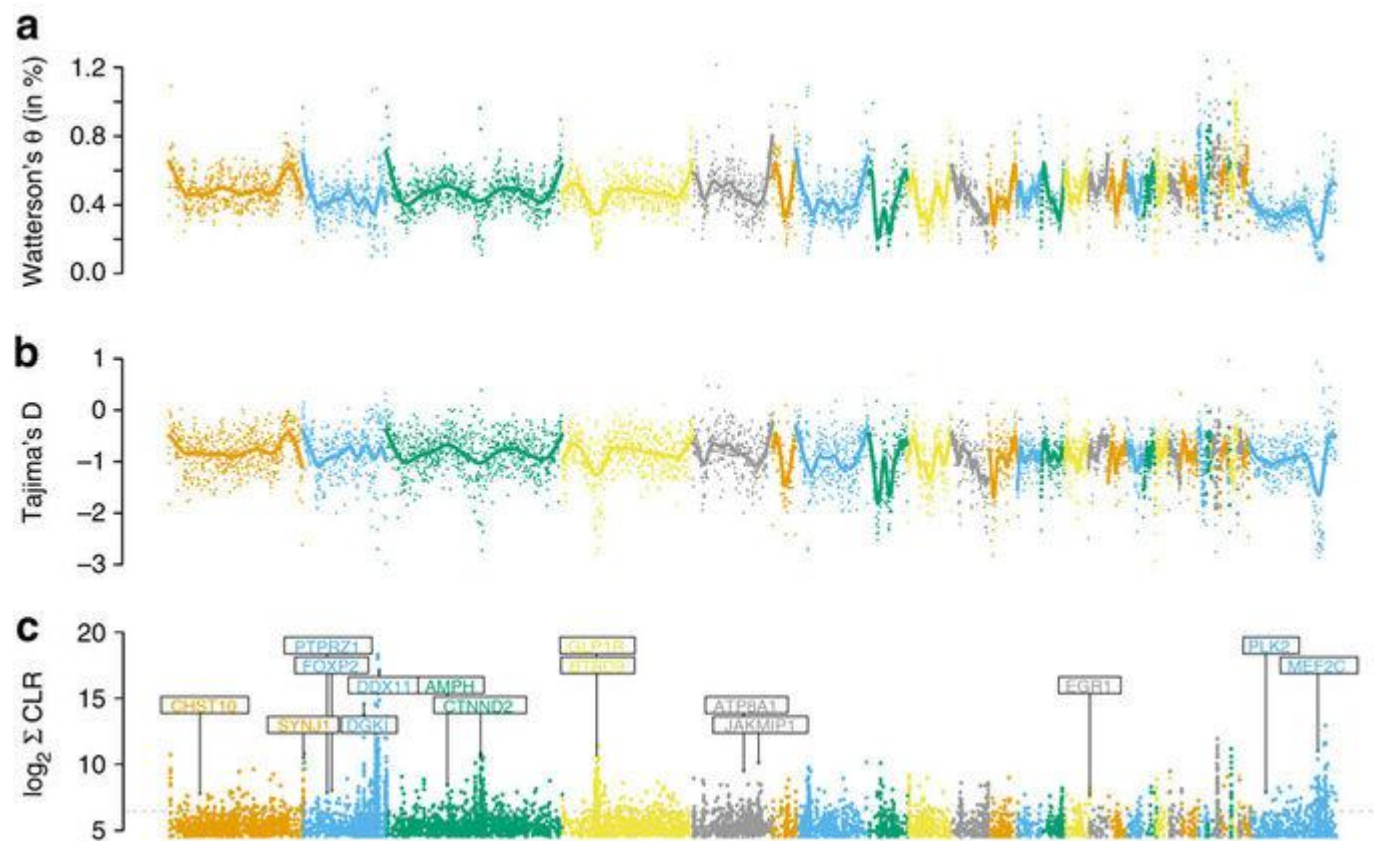
We all like pretty images in papers

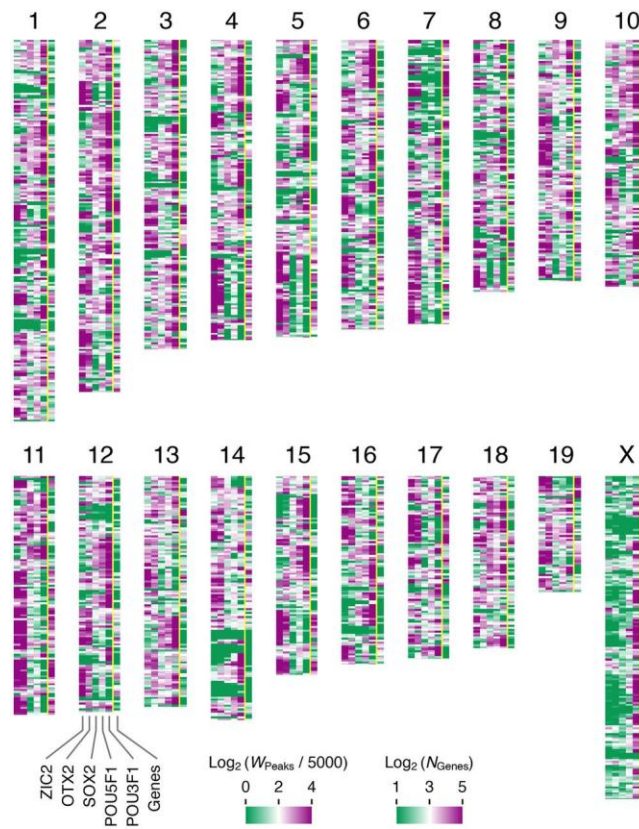


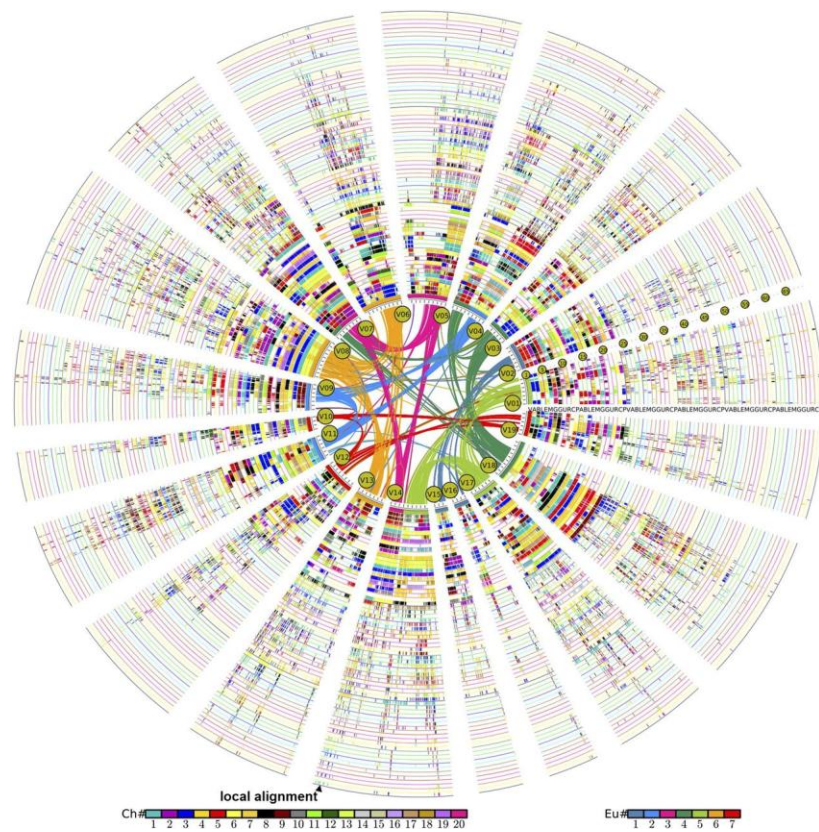




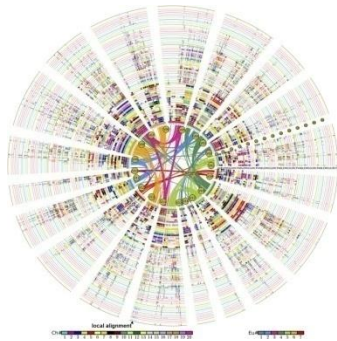




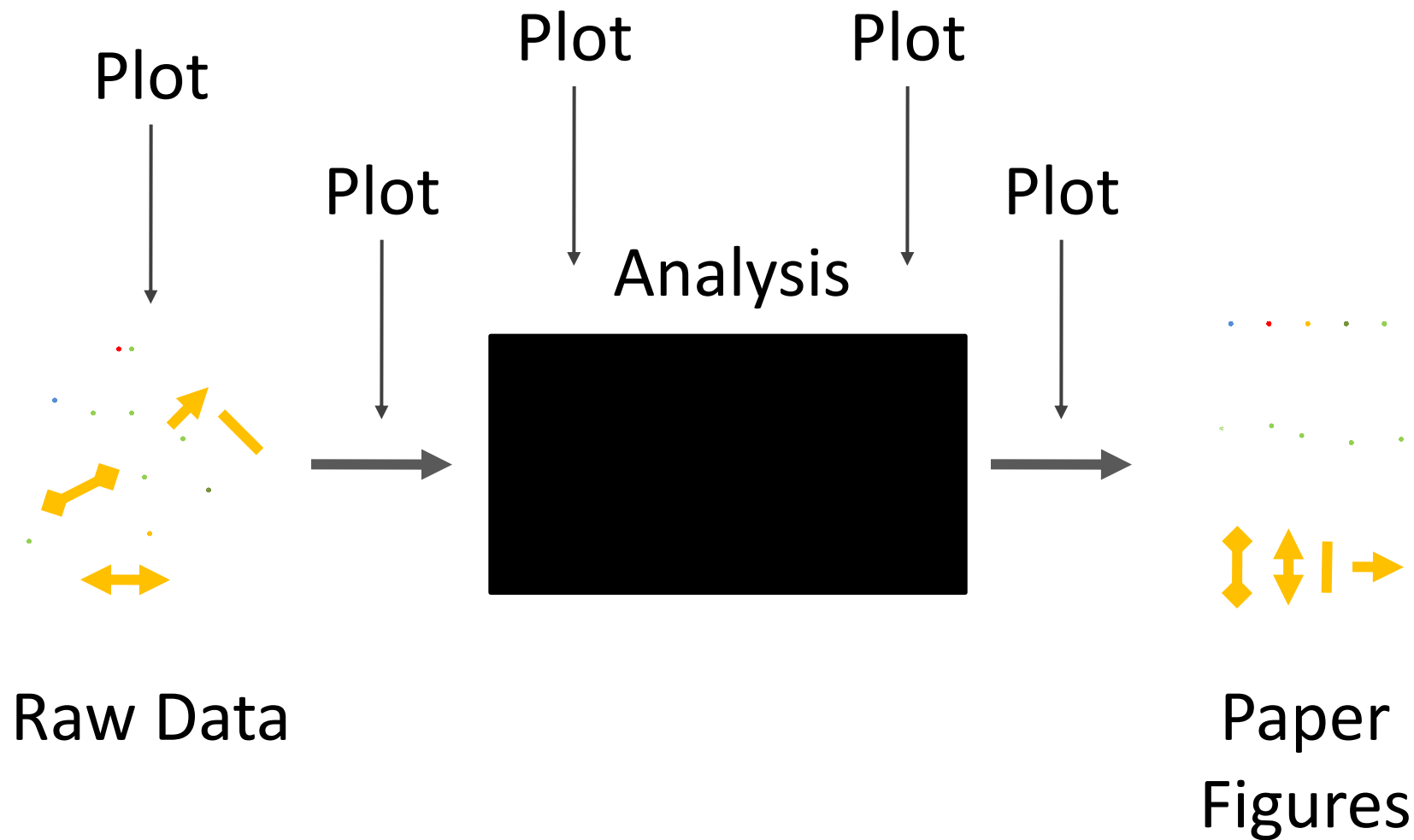




Images (might) help us convey a message



... but they can do more than that



Plot Everything!

aCGH

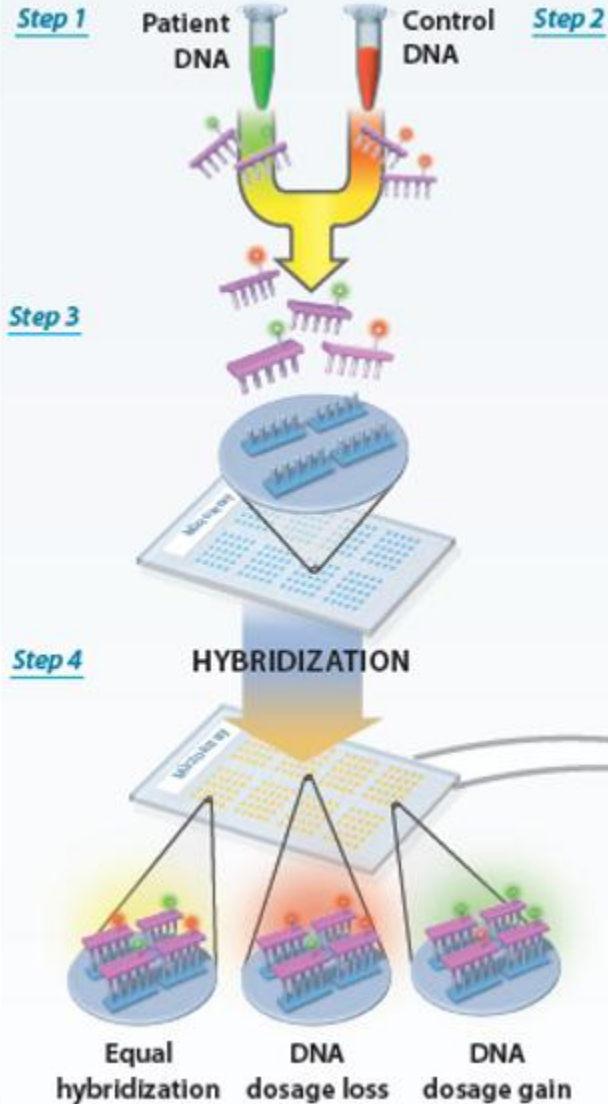
Array CGH: The Complete Process

Steps 1-3 Patient and control DNA are labeled with fluorescent dyes and applied to the microarray.

Step 4 Patient and control DNA compete to attach, or hybridize, to the microarray.

Step 5 The microarray scanner measures the fluorescent signals.

Step 6 Computer software analyzes the data and generates a plot.



Step 5

Step 6



COMPUTER
SOFTWARE



DATA PLOT
(Chromosome 7)



Dídac Barroso

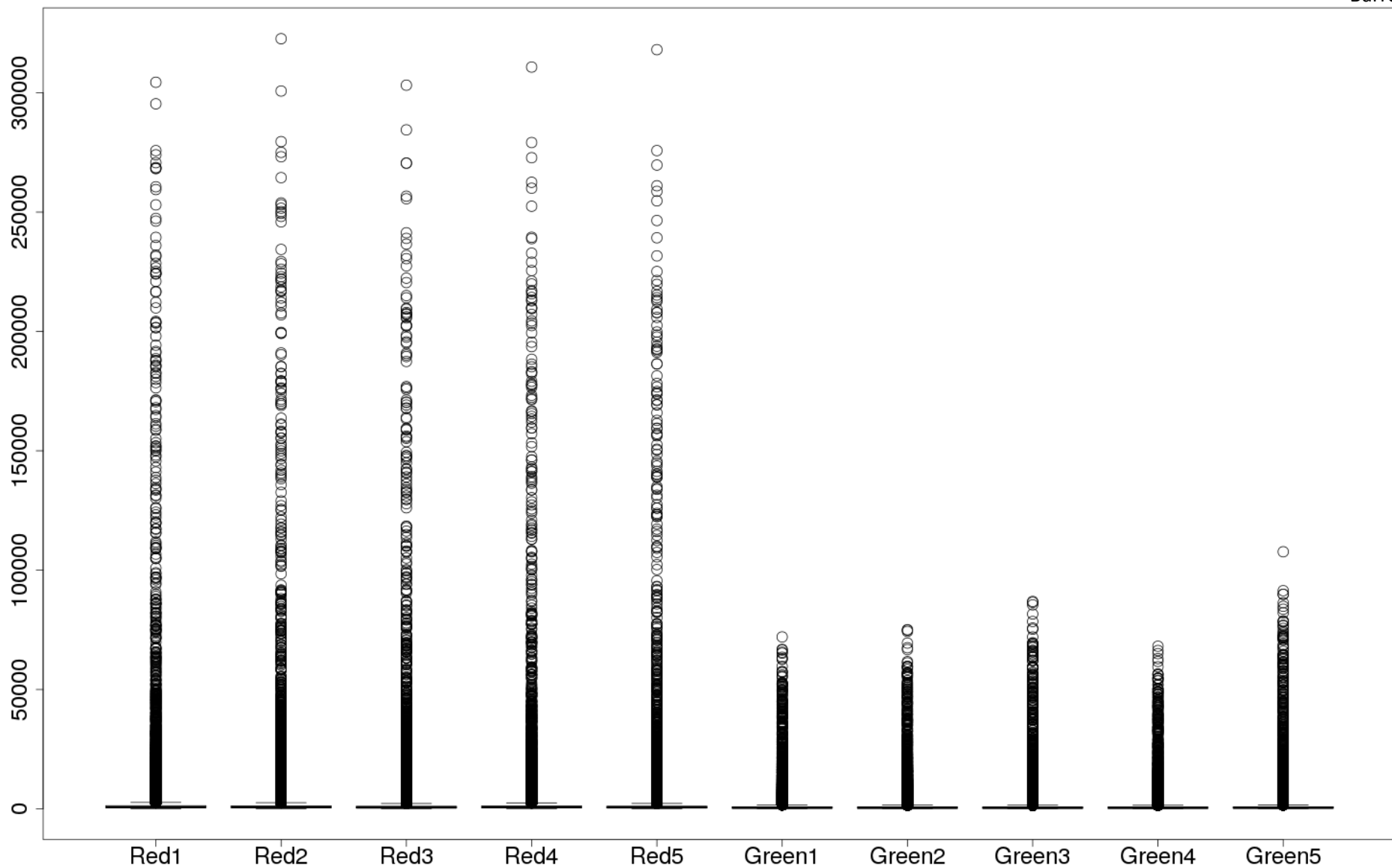
Custom aCGH design

Very noisy probes → Custom analysis



Dídac Barroso

5 samples from blood with small
insertions/deletions



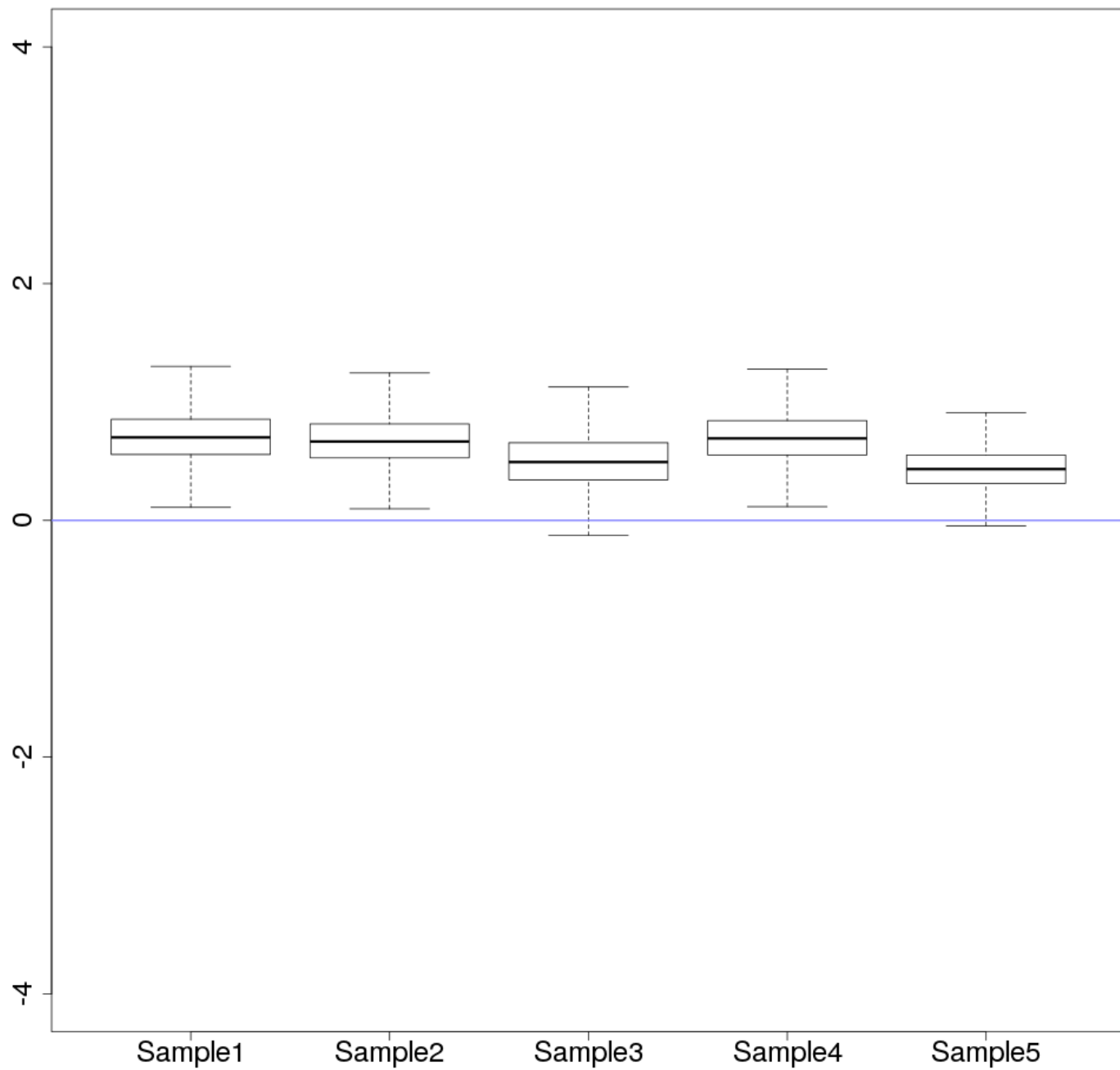


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$$\log_2 \left(\frac{\text{Red Signal}}{\text{Green Signal}} \right)$$




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rCGH Bioconductor package



Bioconductor
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build [warnings](#)

DOI: [10.18129/B9.bioc.rCGH](#) [f](#) [t](#)

Comprehensive Pipeline for Analyzing and Visualizing Array-Based CGH Data

Bioconductor version: Release (3.7)

A comprehensive pipeline for analyzing and interactively visualizing genomic profiles generated through commercial or custom aCGH arrays. As inputs, rCGH supports Agilent dual-color Feature Extraction files (.txt), from 44 to 400K, Affymetrix SNP6.0 and cytoScanHD probeset.txt, cychp.txt, and cnchp.txt files exported from ChAS or Affymetrix Power Tools. rCGH also supports custom arrays, provided data complies with the expected format. This package takes over all the steps required for individual genomic profiles analysis, from reading files to profiles segmentation and gene annotations. This package also provides several visualization functions (static or interactive) which facilitate individual profiles interpretation. Input files can be in compressed format, e.g. .bz2 or .gz.

Author: Frederic Commo [aut, cre]

Maintainer: Frederic Commo <fredcommo at gmail.com>

Citation (from within R, enter `citation("rCGH")`):

Commo F (2017). rCGH: Comprehensive Pipeline for Analyzing and Visualizing Array-Based CGH Data. R package version 1.10.0, <https://github.com/fredcommo/rCGH>.

Installation

To install this package, start R and enter:

```
## try http:// if https:// URLs are not supported
source("https://bioconductor.org/biocLite.R")
biocLite("rCGH")
```

Documentation

To view documentation for the version of this package installed in your system, start R and enter:

```
browseVignettes("rCGH")
```

[PDF](#) [R Script](#) using rCGH package

[PDF](#) [Reference Manual](#)

[Text](#) [NEWS](#)

Documentation »

Bioconductor

- Package [vignettes](#) and manuals.
- [Workflows](#) for learning and use.
- [Course and conference](#) material.
- [Videos](#).
- Community [resources](#) and [tutorials](#).

R / [CRAN](#) packages and [documentation](#)

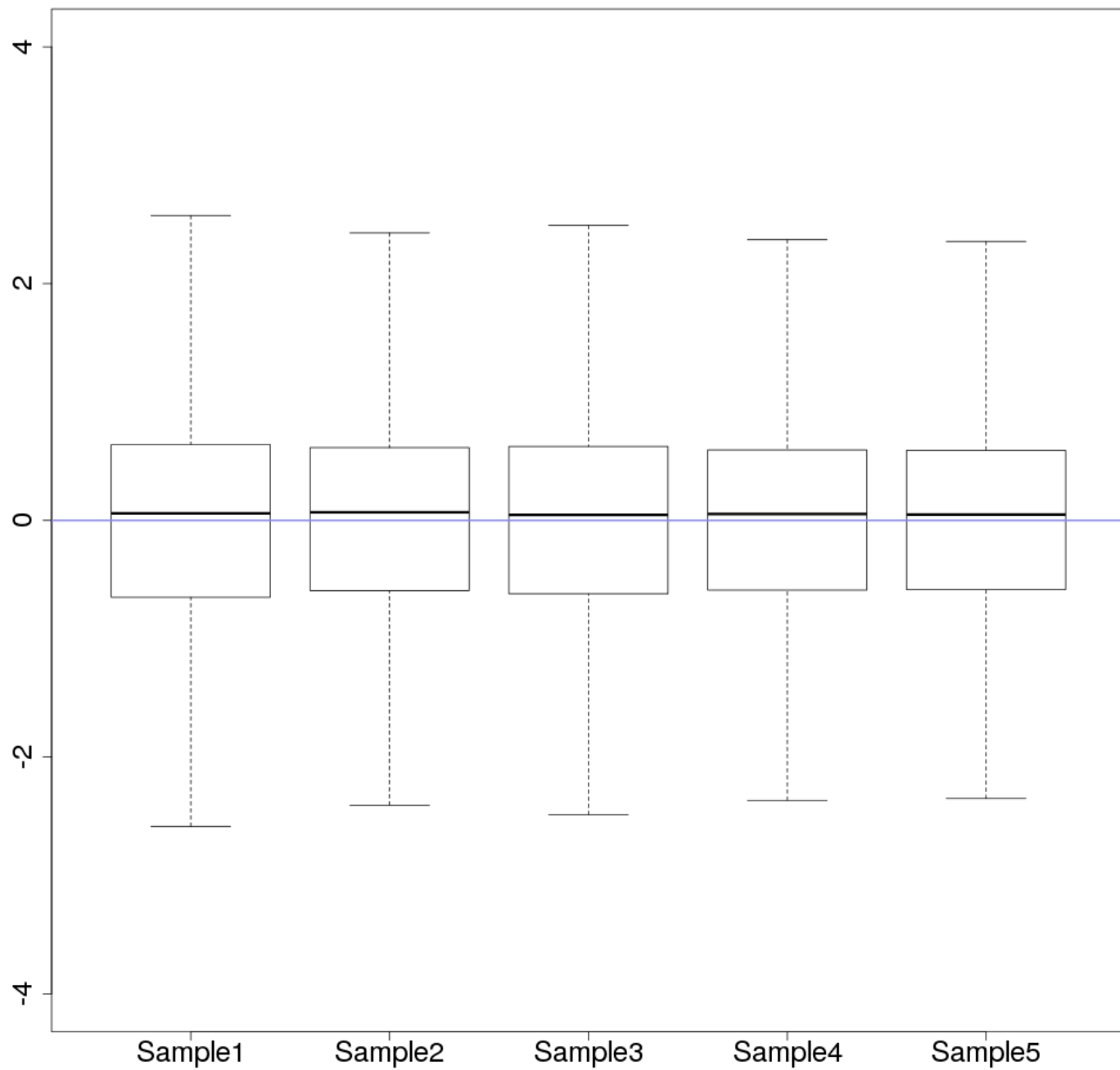
Support »

Please read the [posting guide](#). Post questions about Bioconductor to one of the following locations:

- [Support site](#) - for questions about Bioconductor packages
- [Bioc-devel](#) mailing list - for package developers

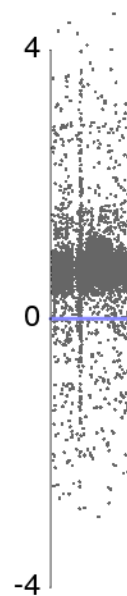


```
cgh <- readAgilent(my.data.file)
cgh <- adjustSignal(cgh)
cgh <- segmentCGH(cgh)
cgh <- EMnormalize(cgh)
```



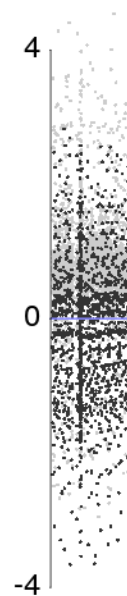


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Barroso



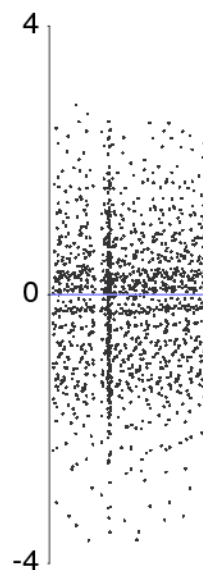


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Barroso



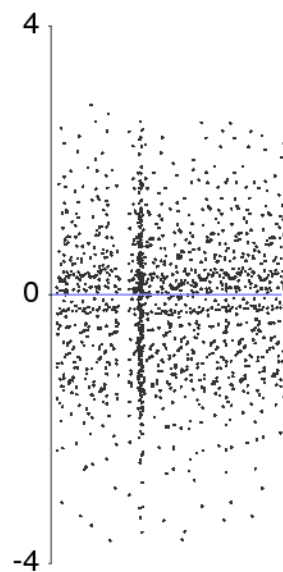


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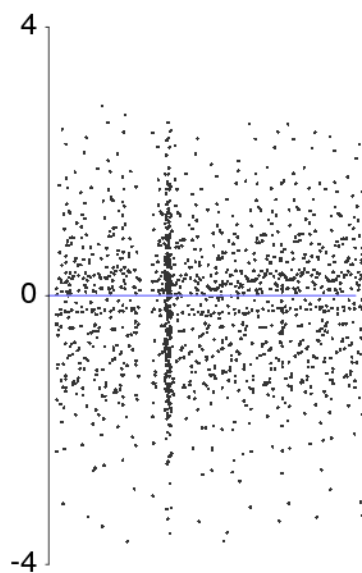


Dídac
Barroso



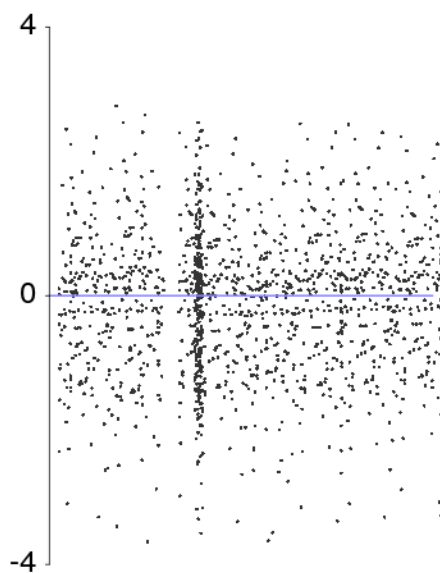


Dídac
Barroso



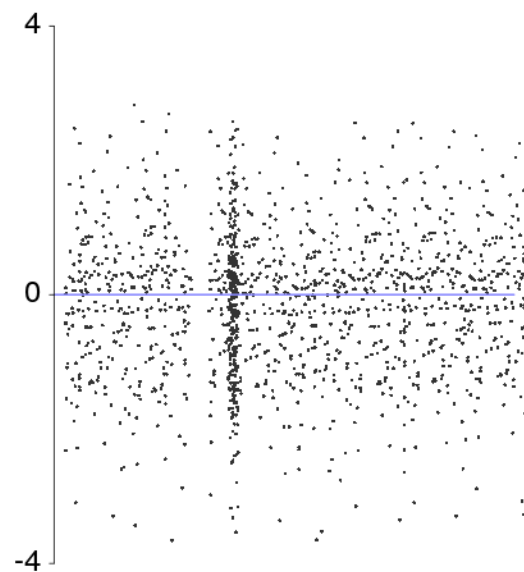


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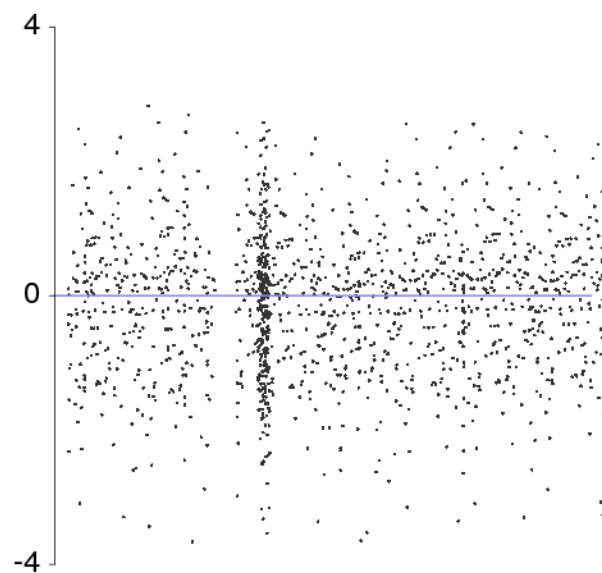


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Barroso



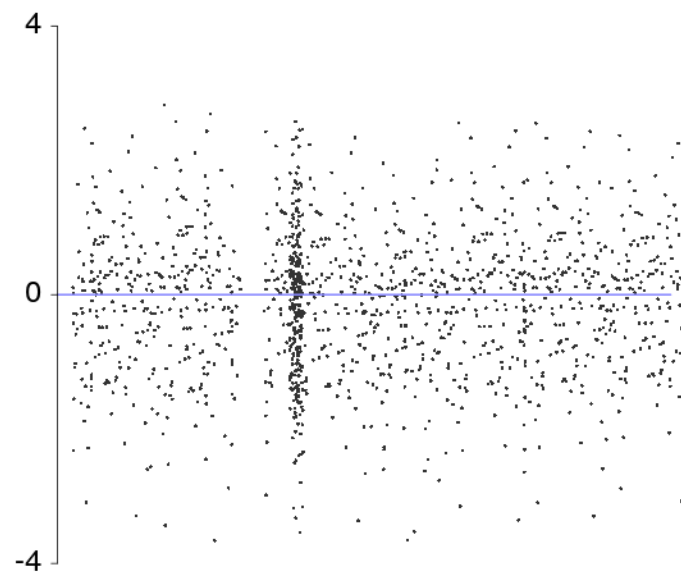


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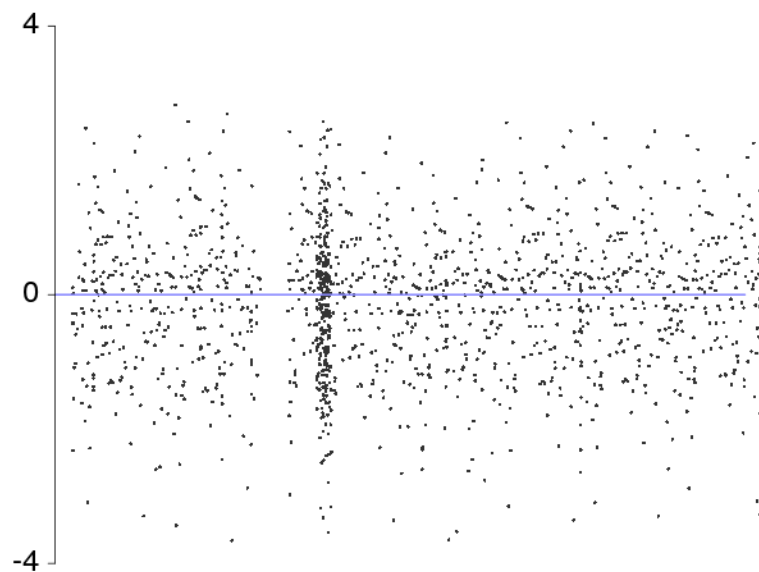


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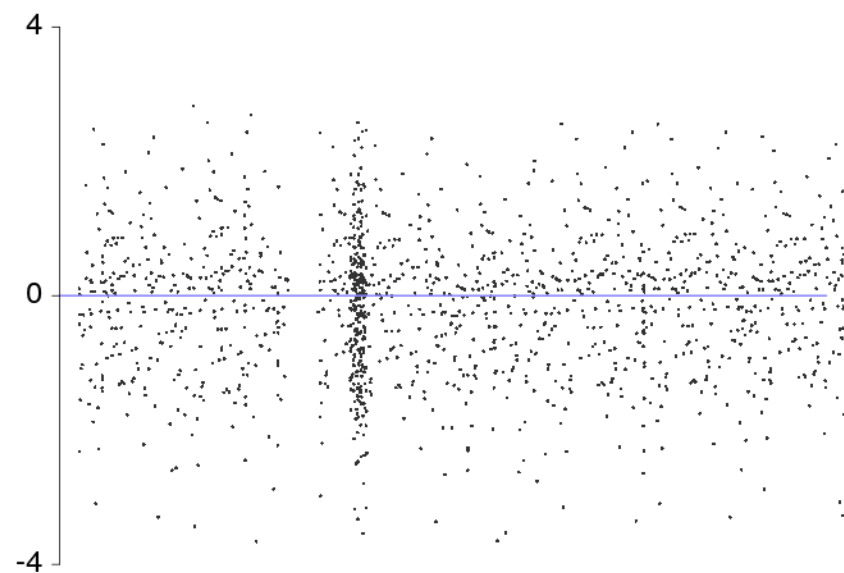


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Barroso



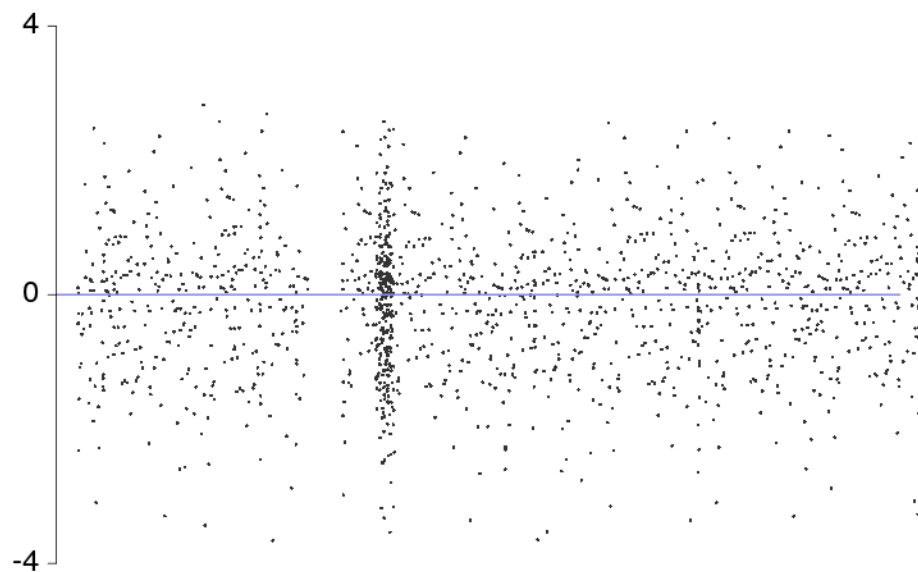


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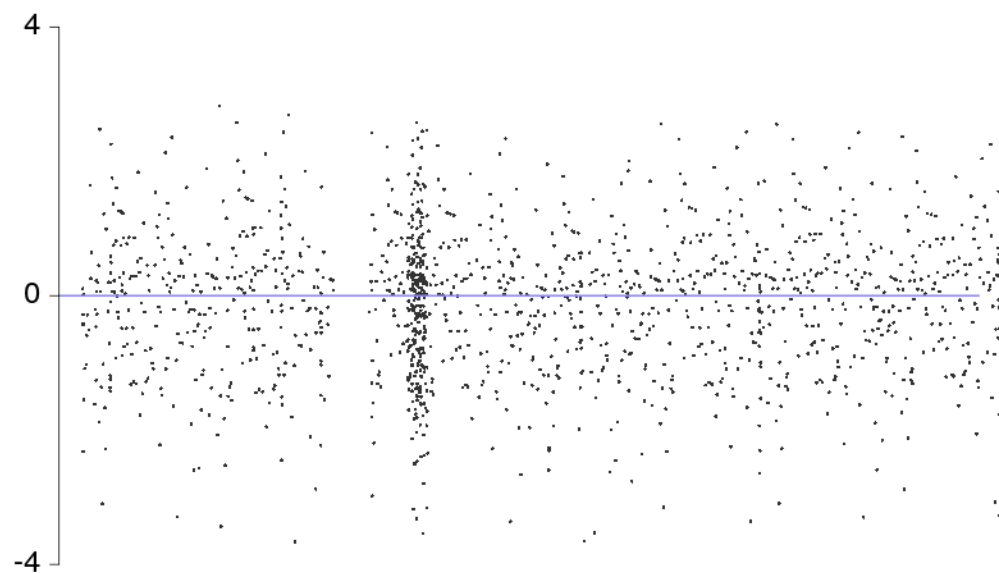


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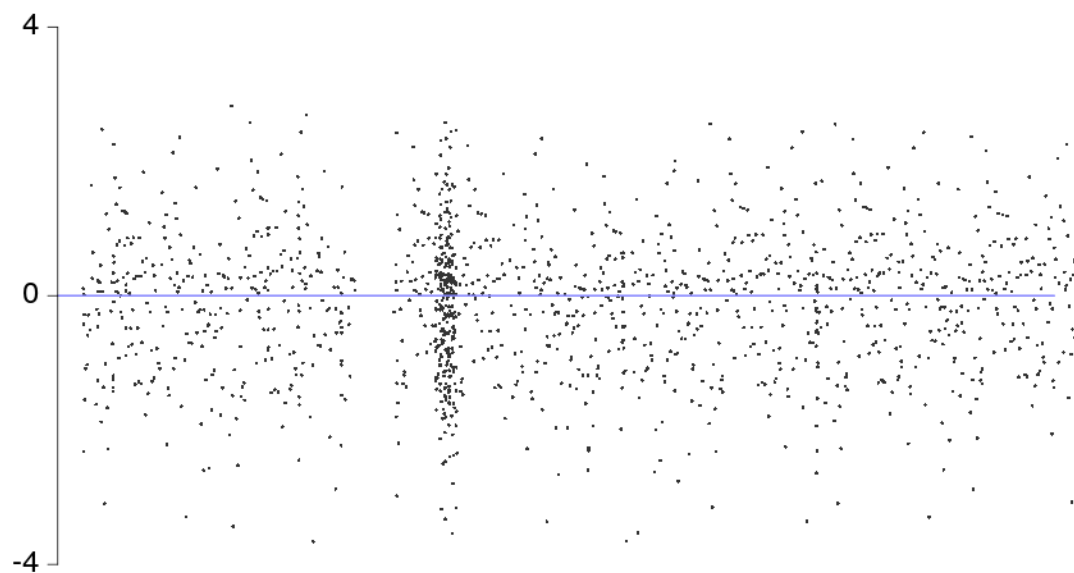


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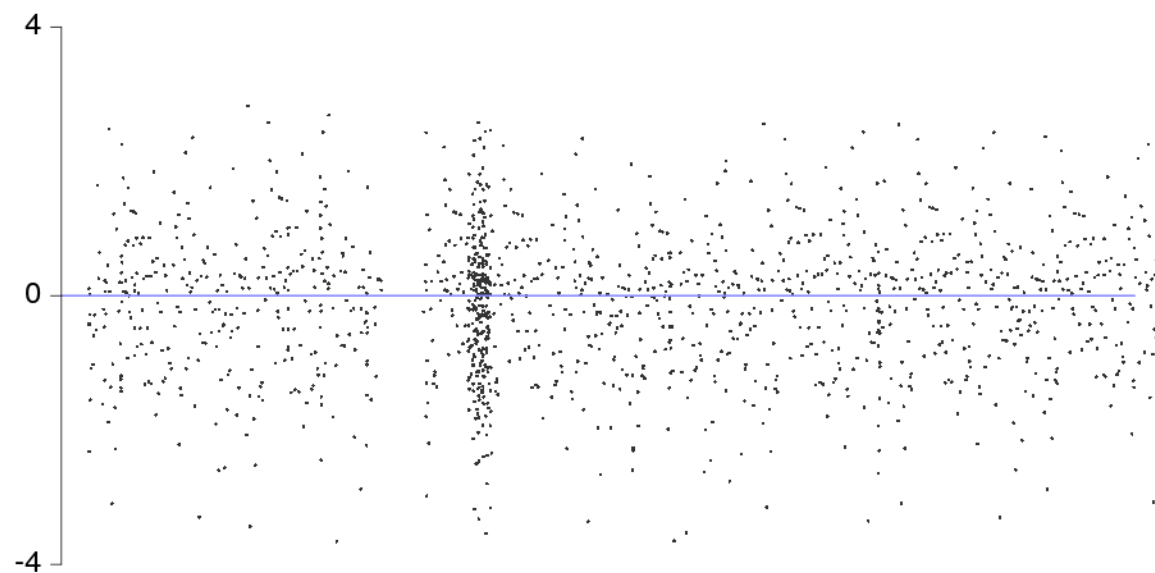


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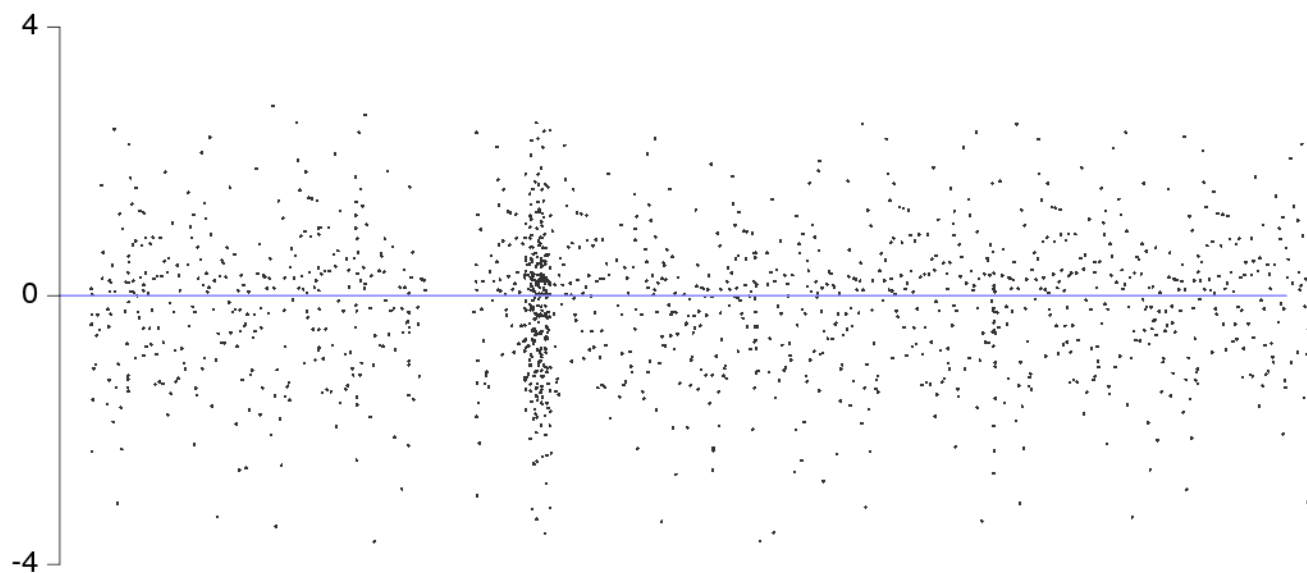


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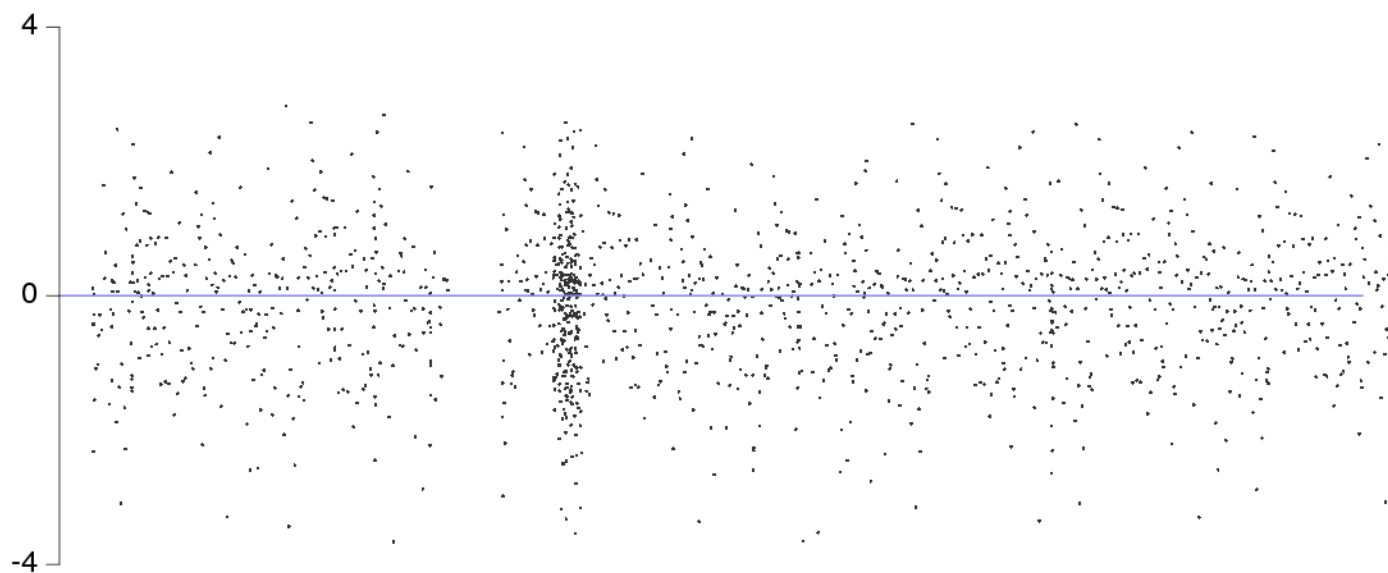


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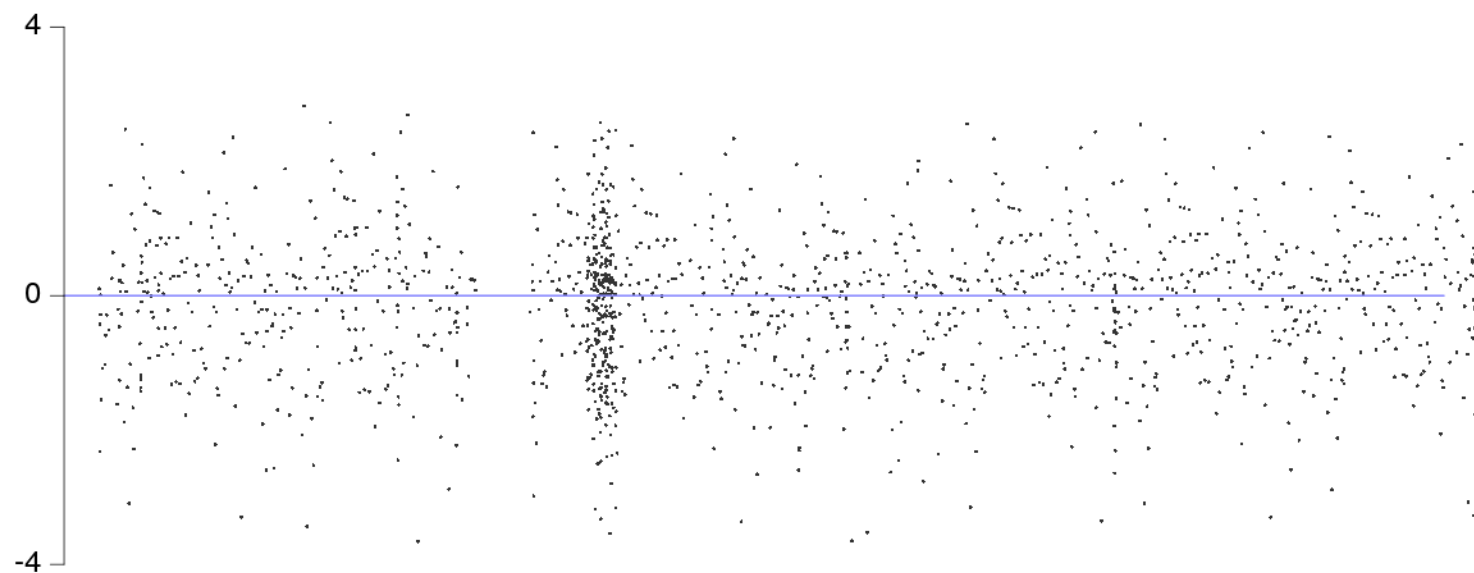


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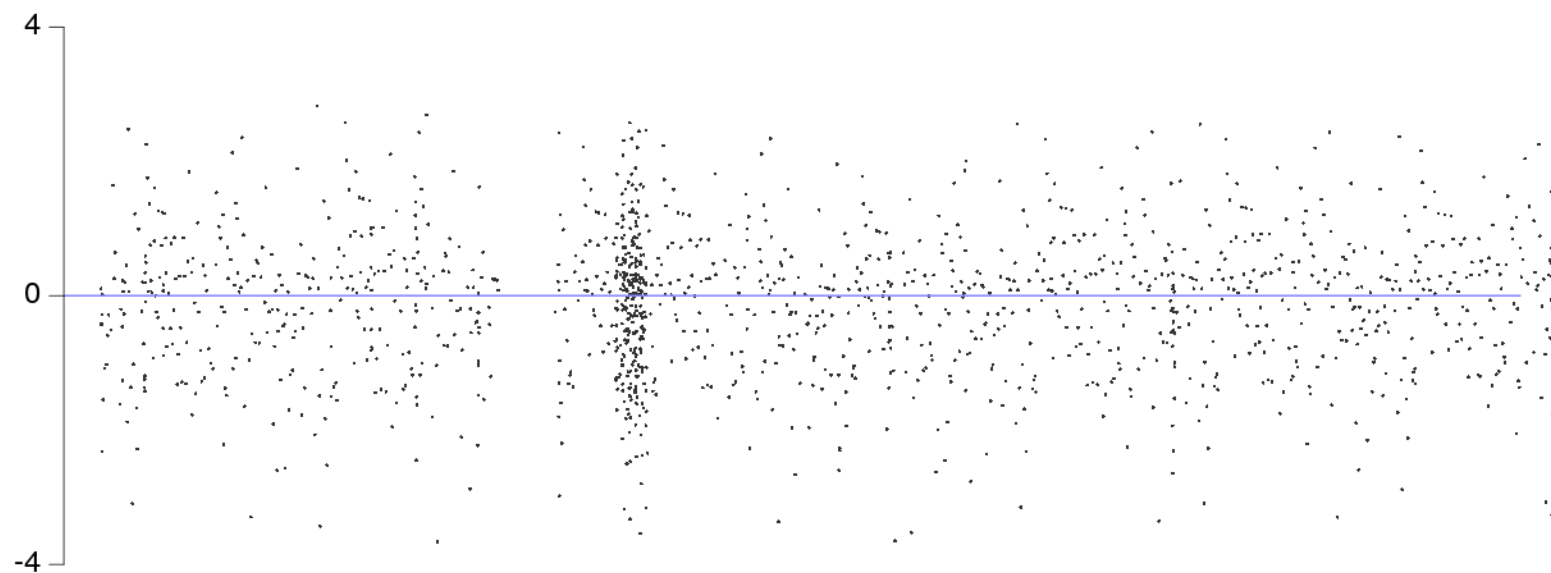


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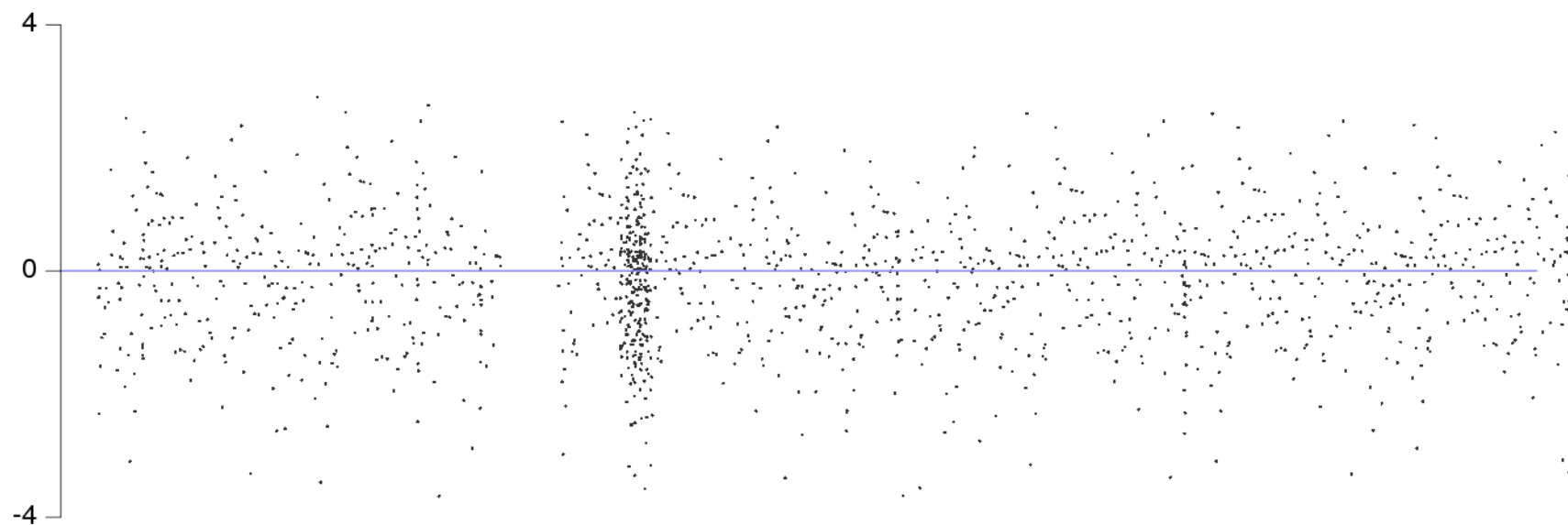


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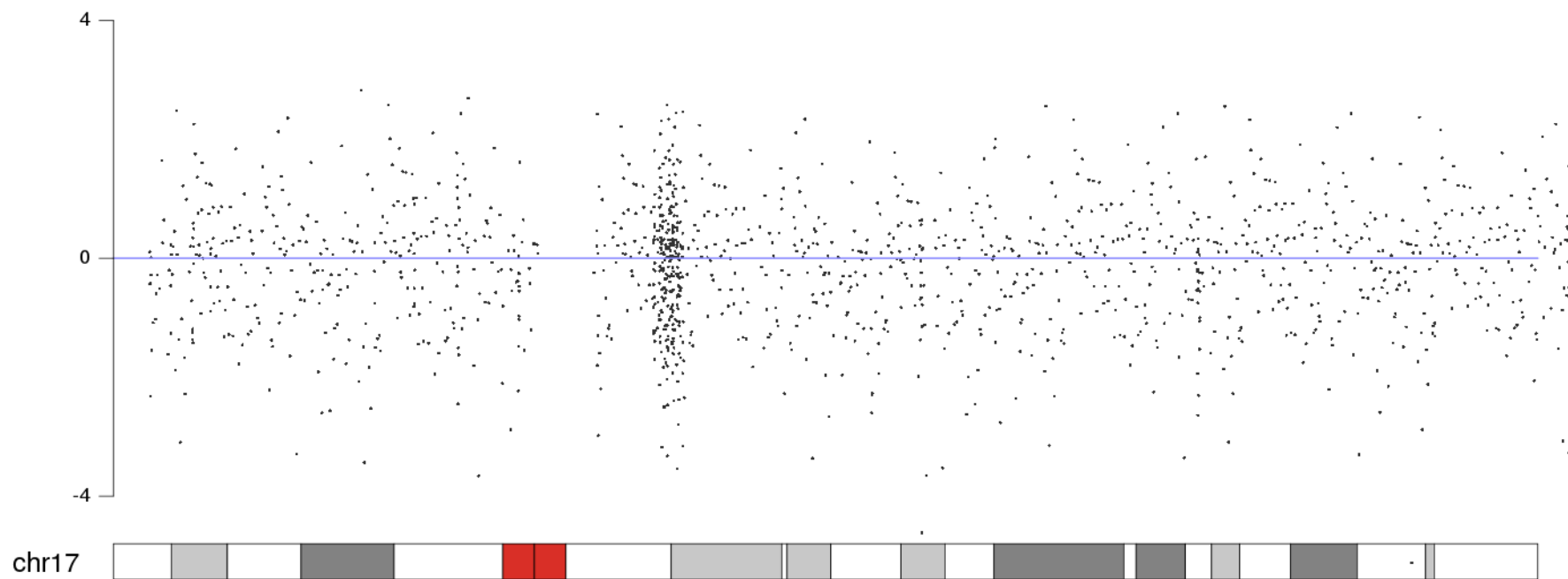


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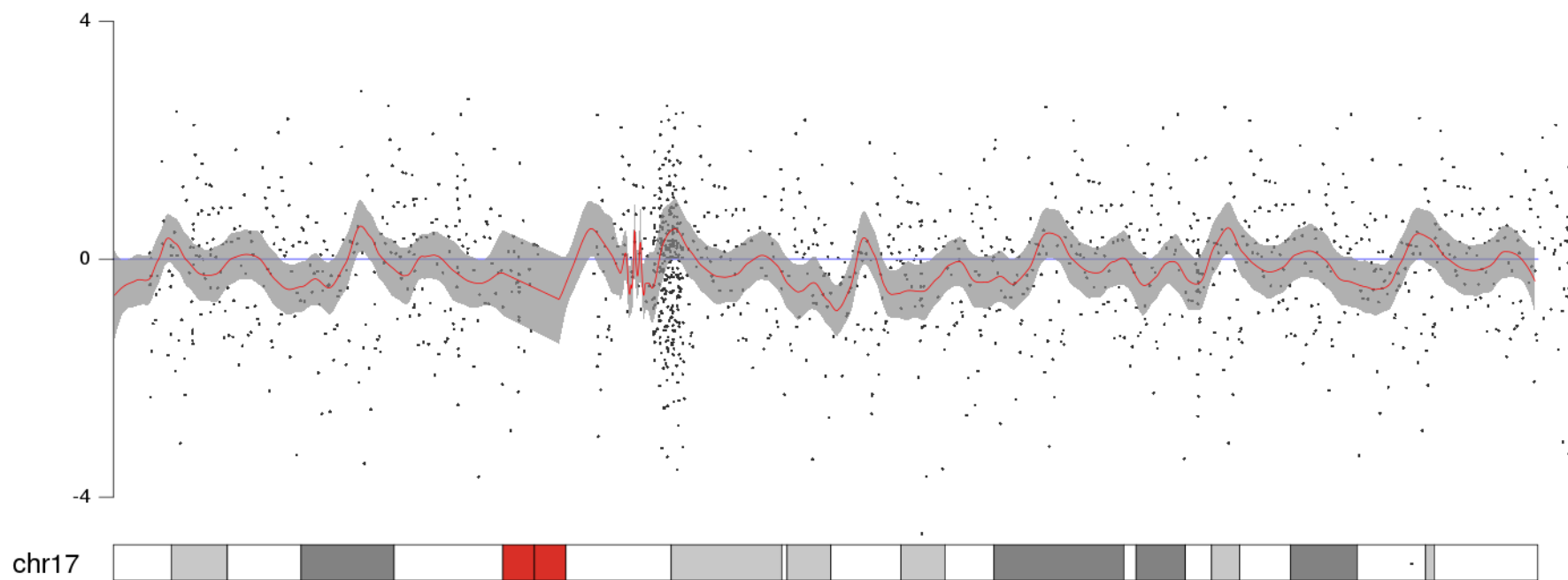


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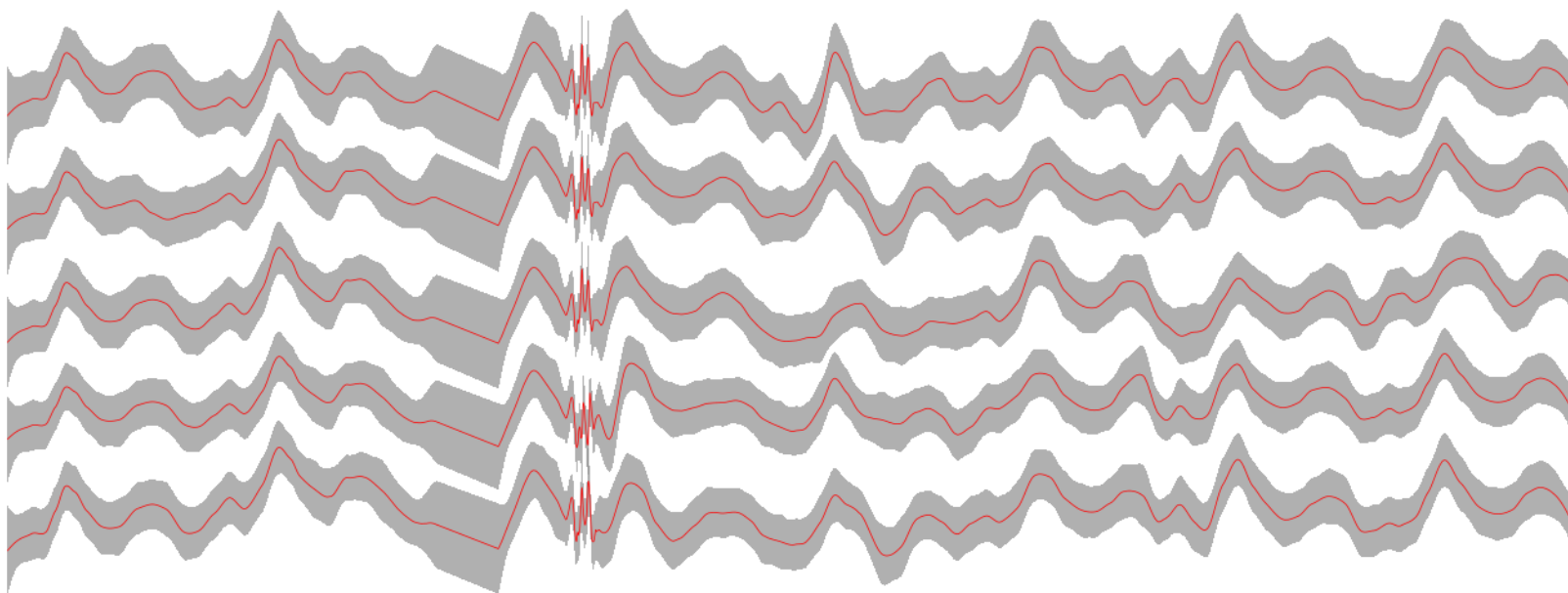


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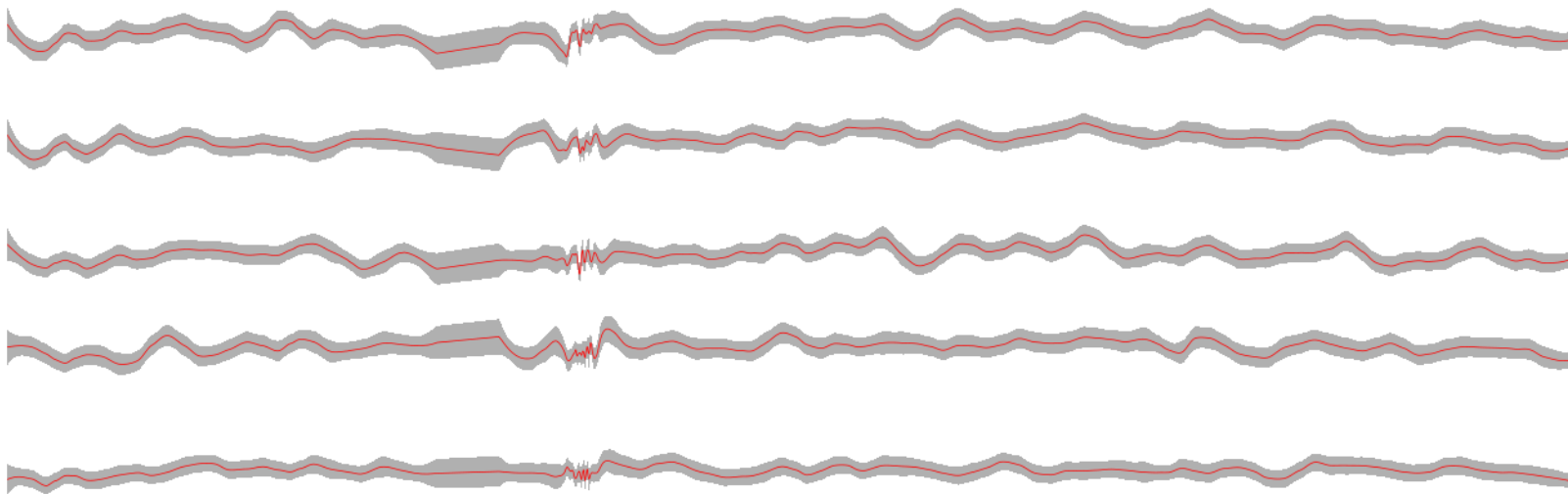


chr17





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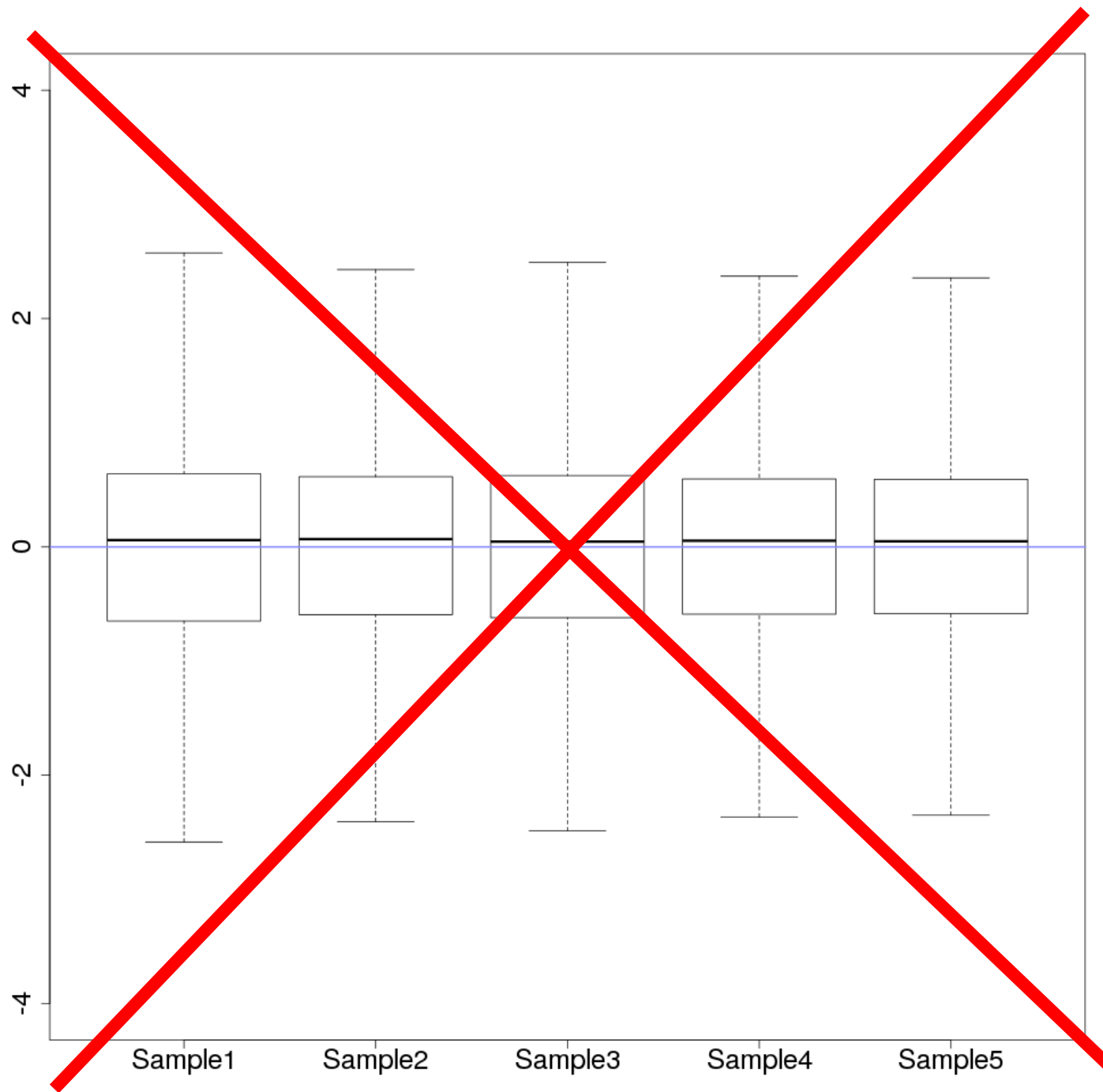


chr17





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Conclusion:
Custom normalization
and
Custom analysis

CNV



Jose Marcos
Moreno

Benchmark of algorithms for CNV calling from NGS data, at single exon level, for genetic diagnostics



Jose Marcos
Moreno

Testing 5 algorithms
4 real genetics diagnostics datasets
Sensitivity 100%
Maximum Specificity



Jose Marcos
Moreno

One one-exon deletion in NF1 was not
found by the algorithms

But is was validated by MLPA!



Jose Marcos
Moreno

We tried optimizing algorithm
parameters

We tried optimizing the optimizer

They never found the deletion

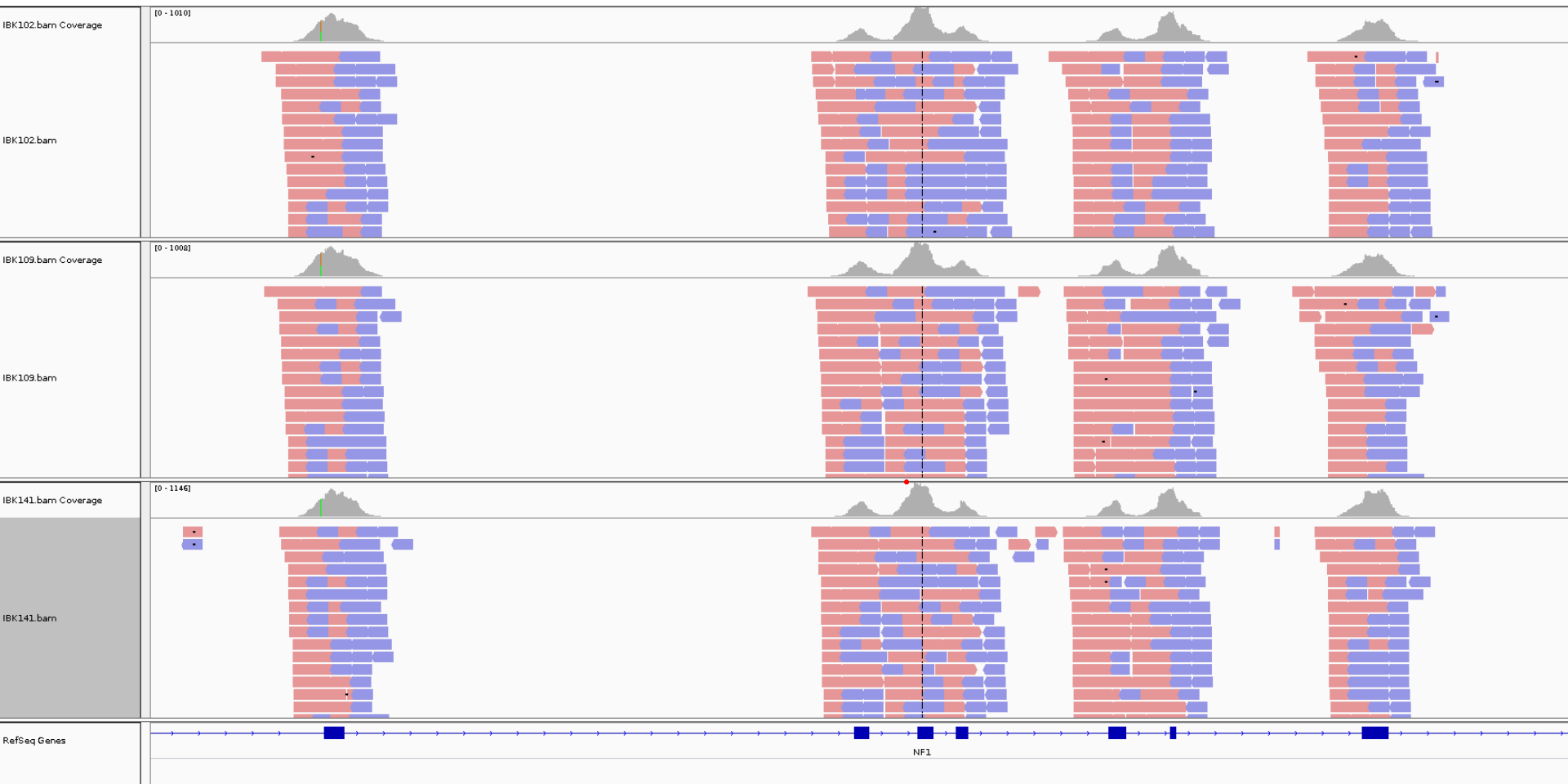


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Plot our data

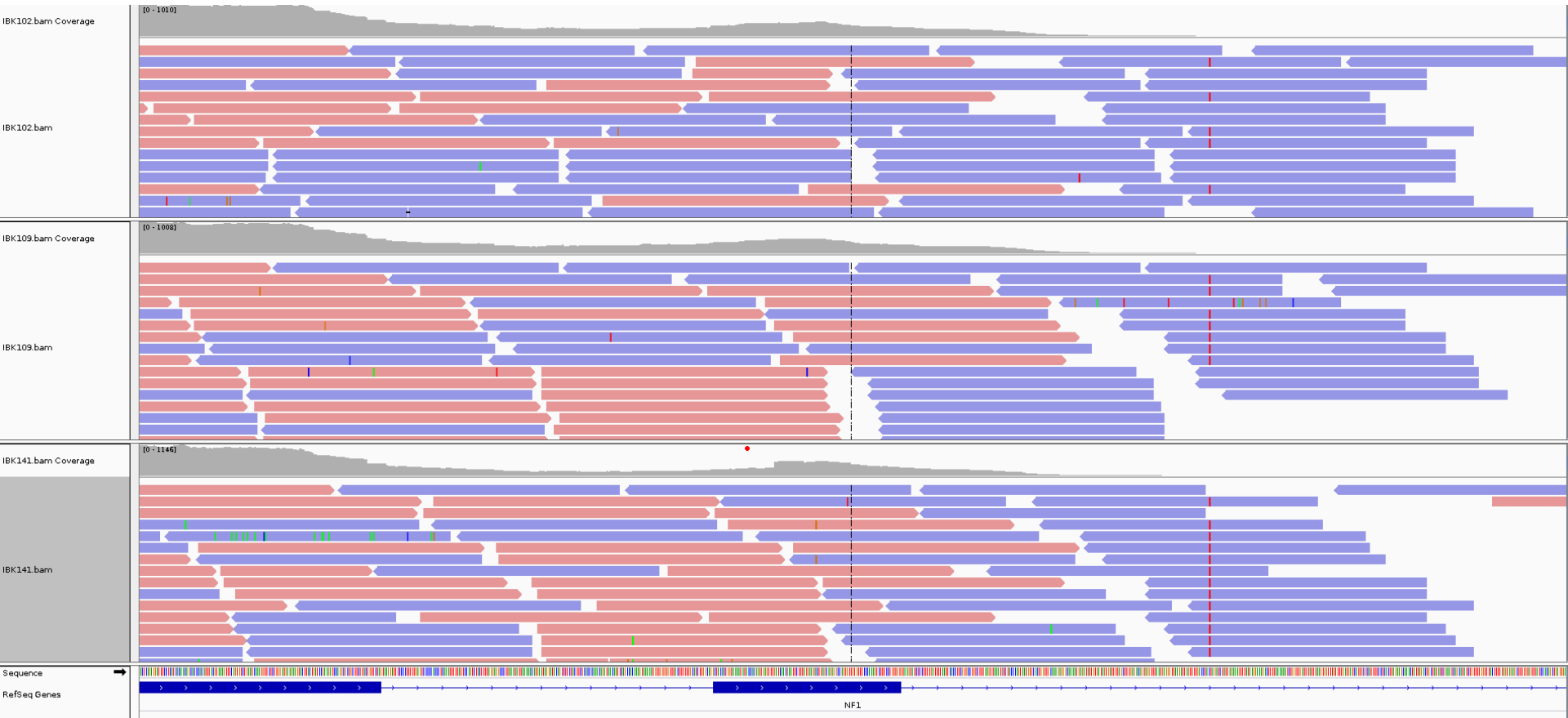


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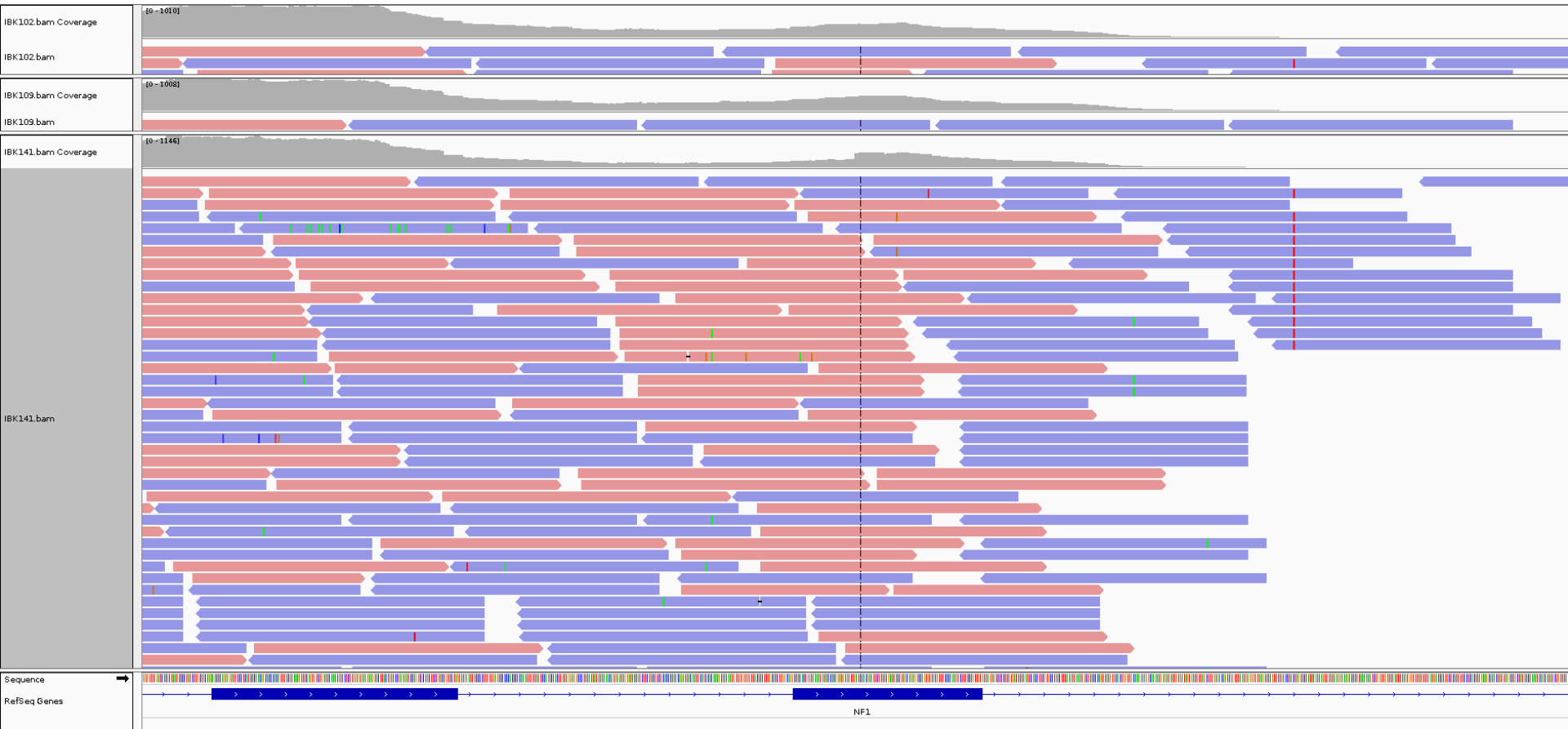


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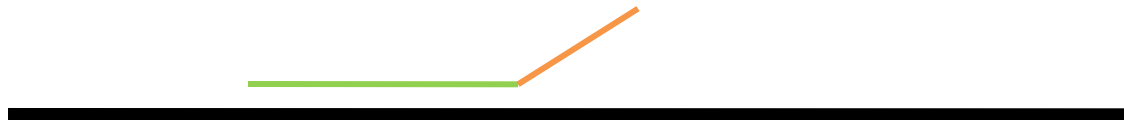
Jose Marcos
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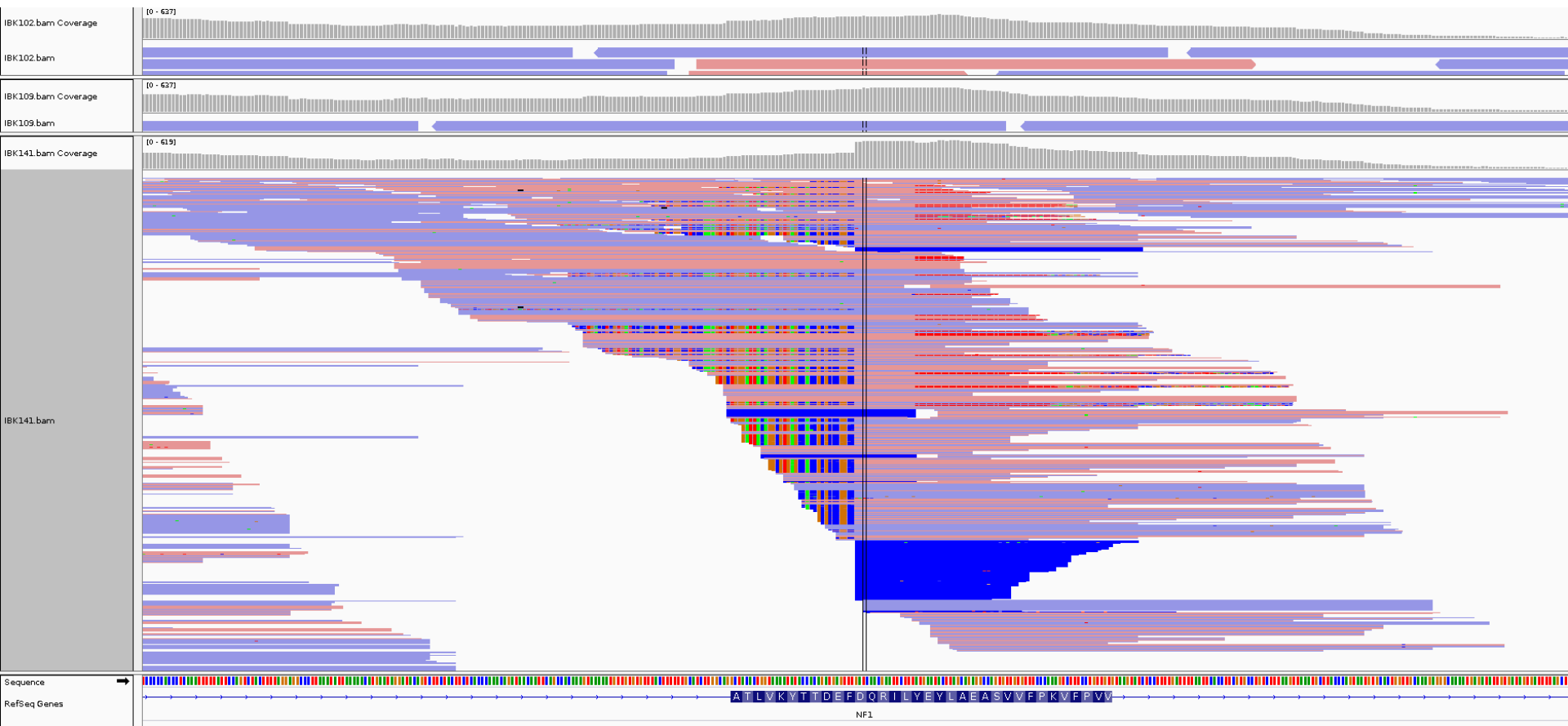
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Show soft-clipped reads





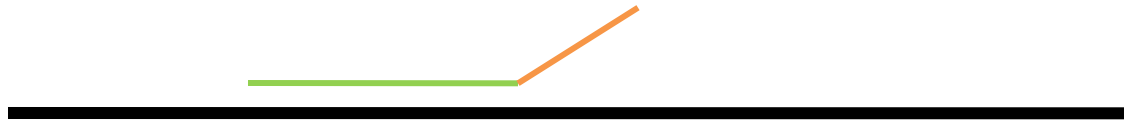
Jose Marcos
Moreno



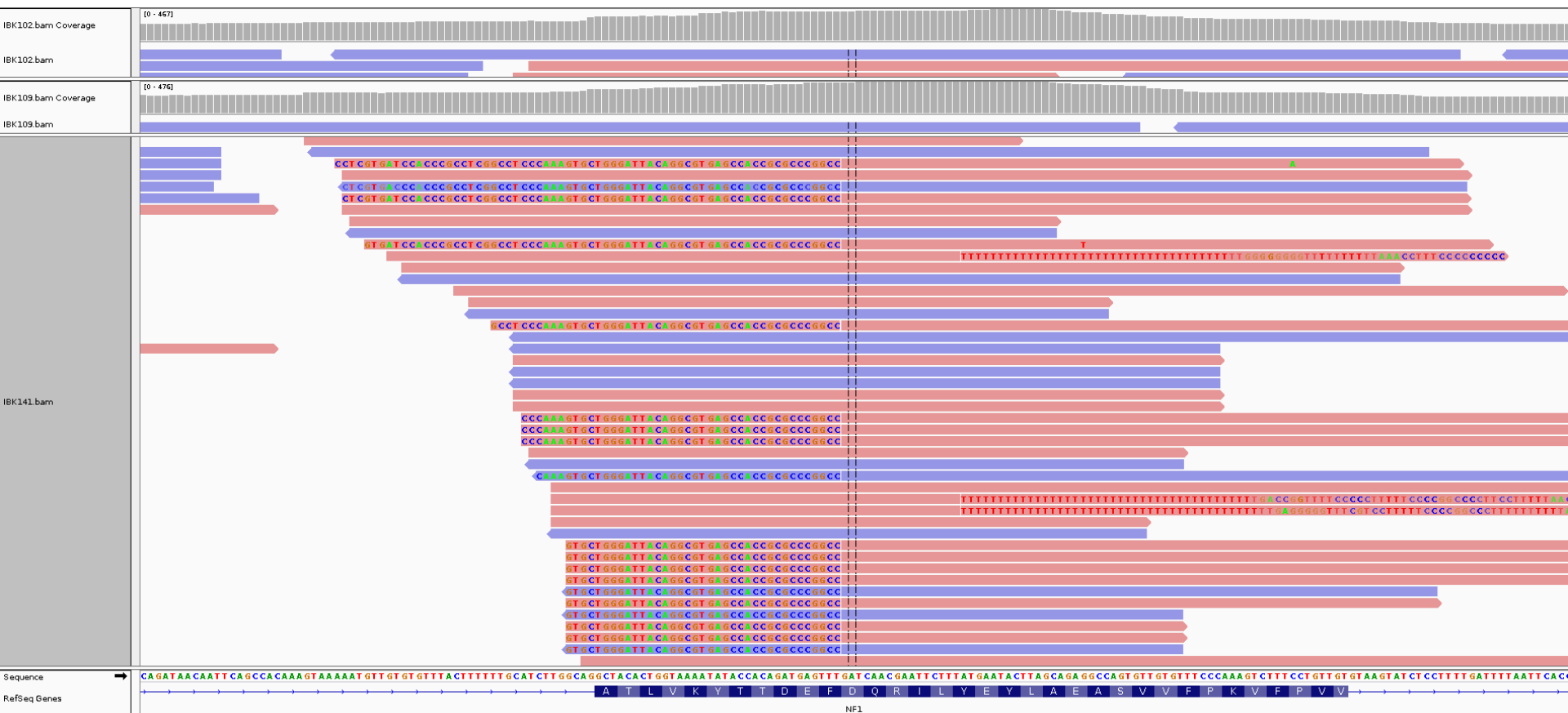
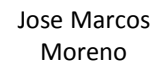


Jose Marcos
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Show soft-clipped reads

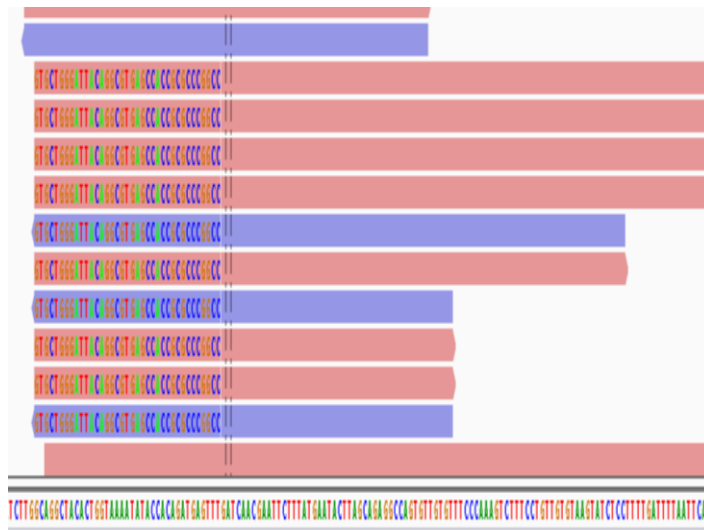


View -> Preferences -> Alignments
View soft-clipped bases





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BLAST



ALU

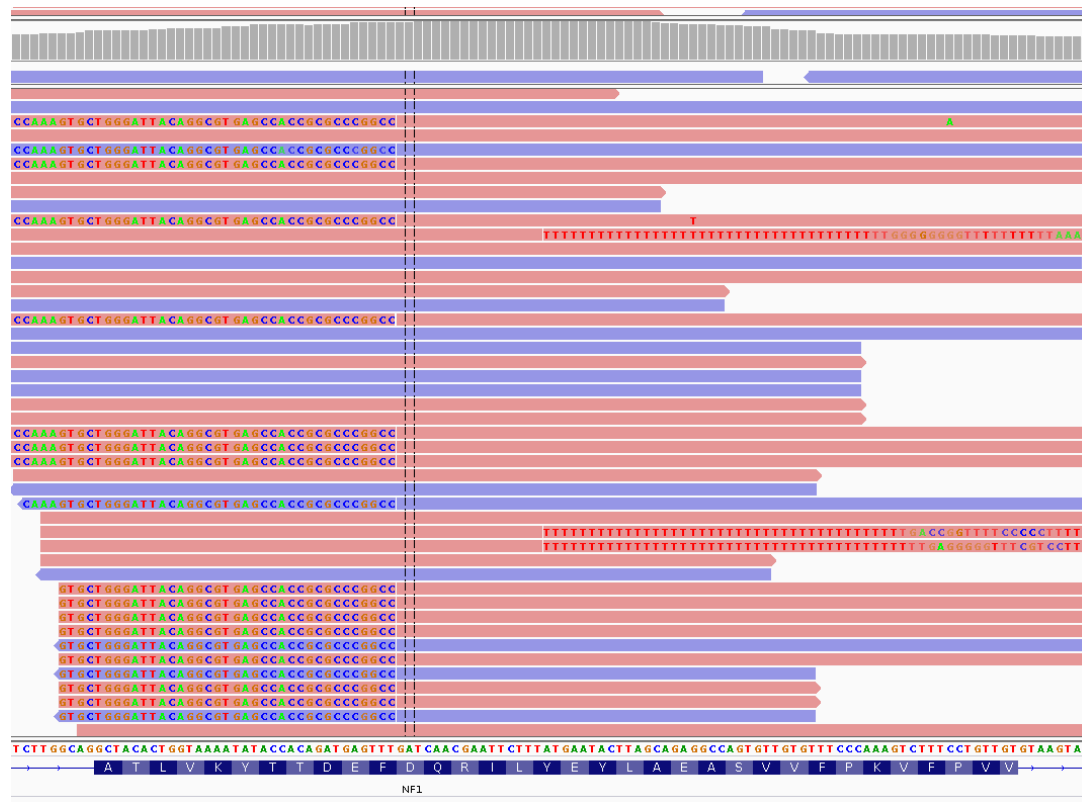


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What about the MLPA validation?



Jose Marcos
Moreno



MLPA probe



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What about the MLPA validation?

It was a false positive

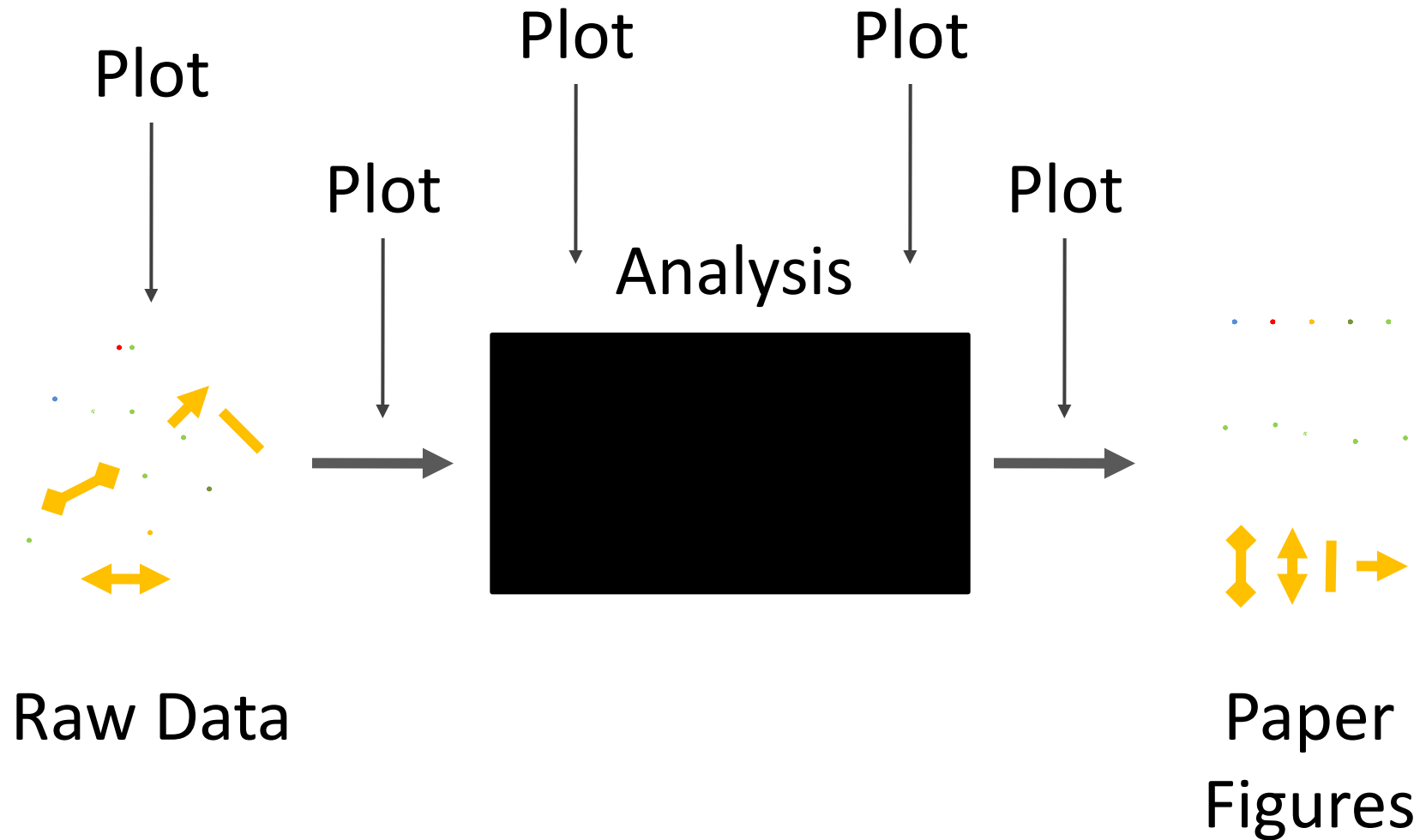


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We finally got the 100% sensitivity we
were looking for

Many other examples: variant calling,
SNP-arrays...

Plot Your Data!



Hereditary Cancer Group

