



Instituto
René Rachou
FIOCRUZ MINAS

Identificarea variantelor

- Mapeamento de reads sequenciados.
- Sobreposição das sequências.
- Estatística para identificação da variante

[illegible]

CGATGCGTACGAGCGACGATGCGTACGAGCGCGT

ATGCGATCAGTCTAGCGCGATCGAGCG

Heterozigoto: Aa?

Homomorphisms: $G \rightarrow G$?

Identificação de variante

- Mapeamento de reads sequenciados.
- Sobreposição das sequências.
- Estatística para identificação da variante

CGATGCTACAGCATGCGATCAGTCTACGATGCATCAGCTAGCATGCGATCGATGCATCGATCGATCCGATGCGTCAGTCTAGCGAAT
CGATGCTACAGCATGCGATCAGTCTACGATGCATCAGCTAGC**GT**
ATGCTACAGCATGCGATCAGTCTACGATGCGTCAGCTAGC**GT**GTC
GCTACAGCATGCGATCAGTCTACGATGCATCAGCTAGCATGCGA
CTACAGCATGCGATCAGTCTACGATGCATCAGCTAGCATGCGAT
TACAGCATGCGATCCGTCTACGATGCATCAGCTAGC**GT**GCGATC
CAGCATGCGATCAGTCTACGATGCATCAGCTAGC**GT**GCGATCGA
CATGCGATCAGTCTACGATGCATCAGCTAGC**GT**GCGATCGATGC
ATGCGATCAGTCTACTATGCATCAGCTAGC**GT**GCGATCGATGCA
GCGATCAGTCTACGATGCATCAGCTAGC**GT**GCGATCGATGCATC
GATCAGTCTACGATGCGTCAGCTAGC**GT**GCGATCGATGCATCGA
ATCAGTCTACGATGCATCAGCTAGCATGCGATCGATGCATCGAT
CAGTCTACGATCCATCAGCTAGCATGCGATCGATGCATCGATCG
GTCTACGATGCATCAGCTAGCATGCGATTGATGCATCGATCGAT
TCTACGAAGCATCAGCTAGCATGCGATCGATGCATCGATCGATC
CTACGATGCATCAGCTAGCATGCGATCGATGCATCGATCGATCC
TACGATGCATCAGCTAGCATGCGATCGATGCATCGATCGATCCG
ACGATGCATCAGCTAGC**GT**GCGATCGATGCATCGATCGATCCTA

Heterozigoto: A/G?

- Associar uma variante a um fenótipo

  A/G

  G/G

  A/G

  G/G

  A/A

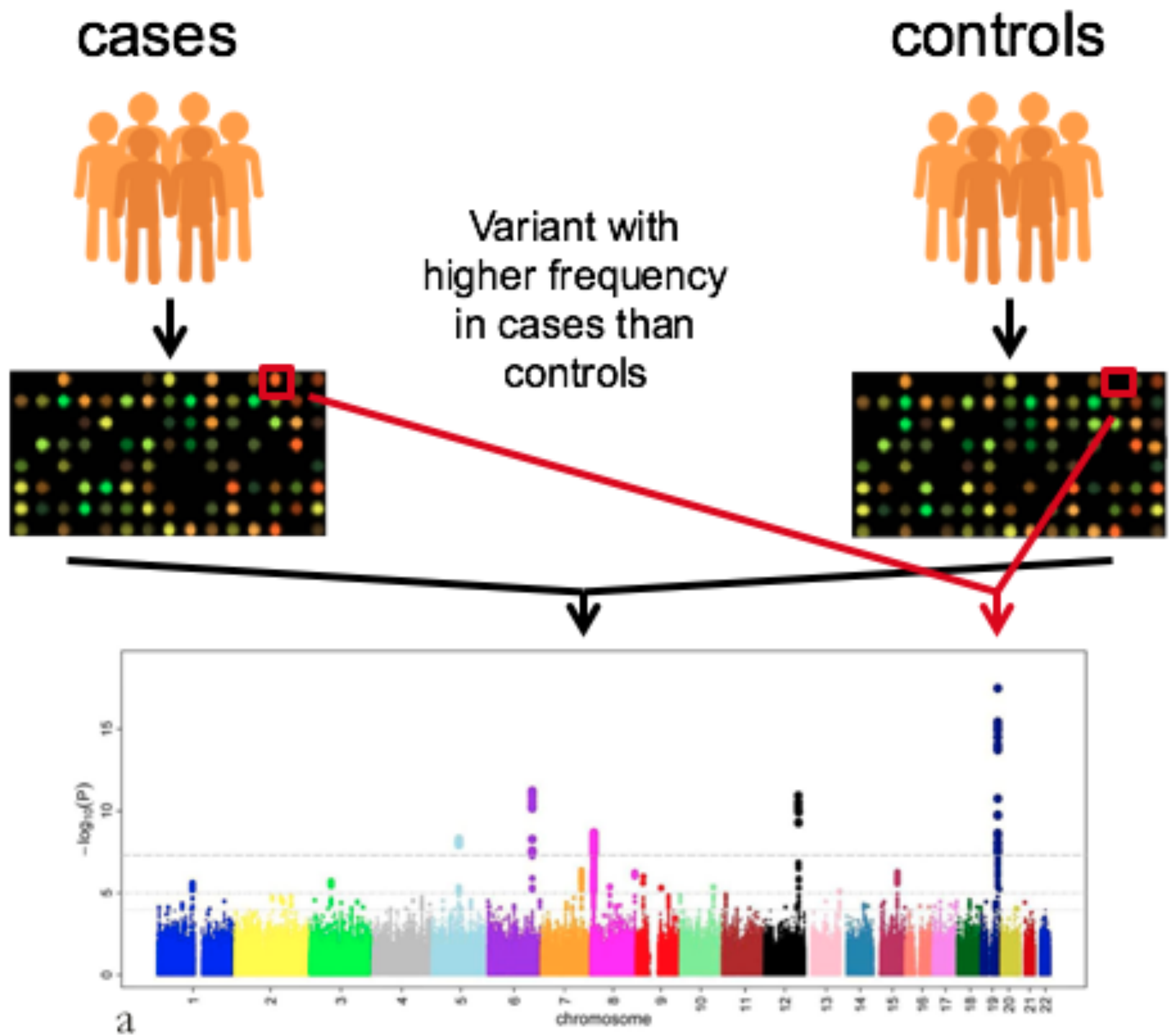
  G/G

  A/A

  A/A

  A/G

  A/A



$p = 1$	G/G	Não G/G
Doente	1	4
Saudável	2	3

$p = 6e-12$	G/G	Não G/G
Doente	100	400
Saudável	200	300