

- Associar uma variante a um fenótipo

  A/G

  G/G

  A/G

  G/G

  A/A

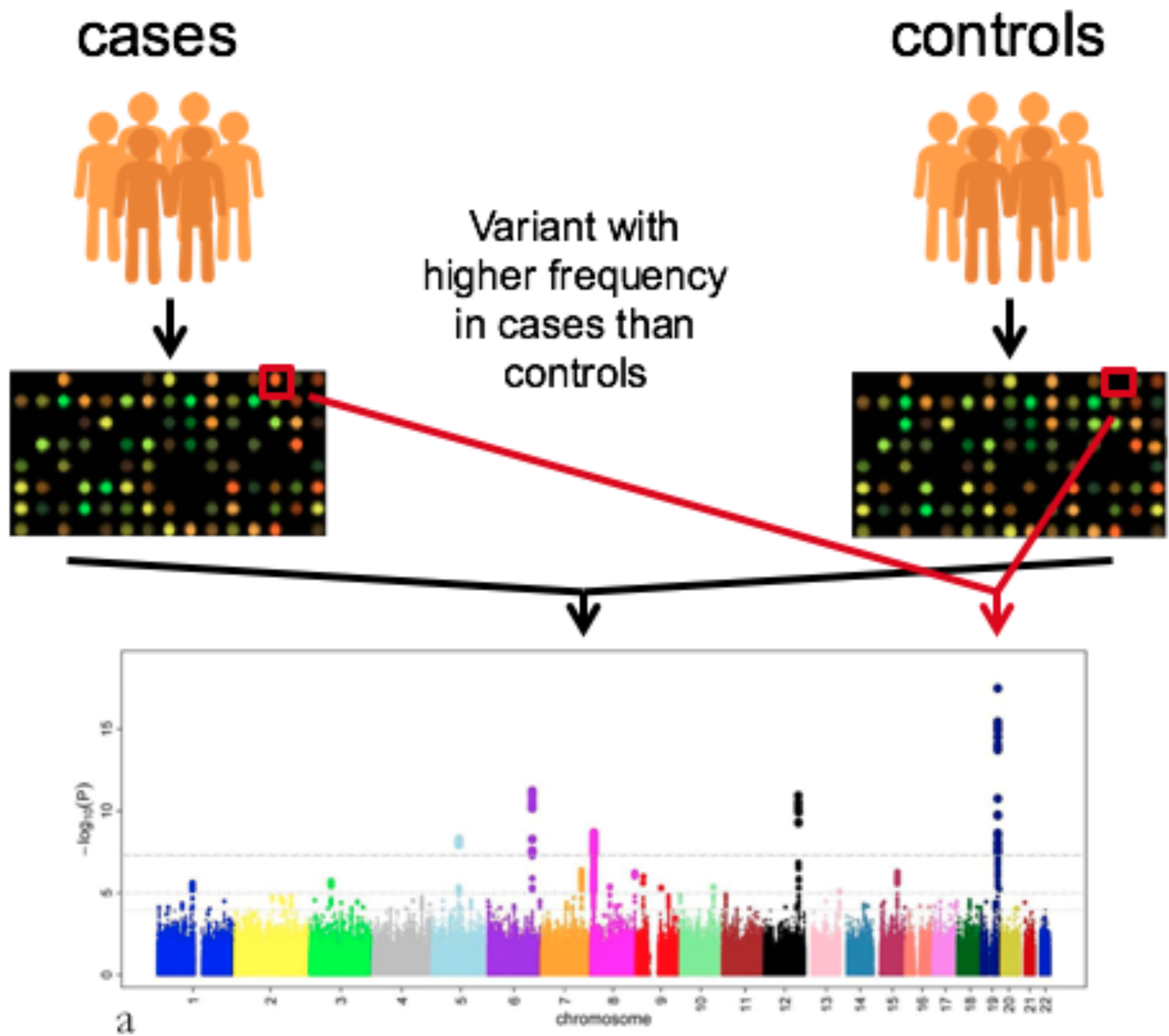
  G/G

  A/A

  A/A

  A/G

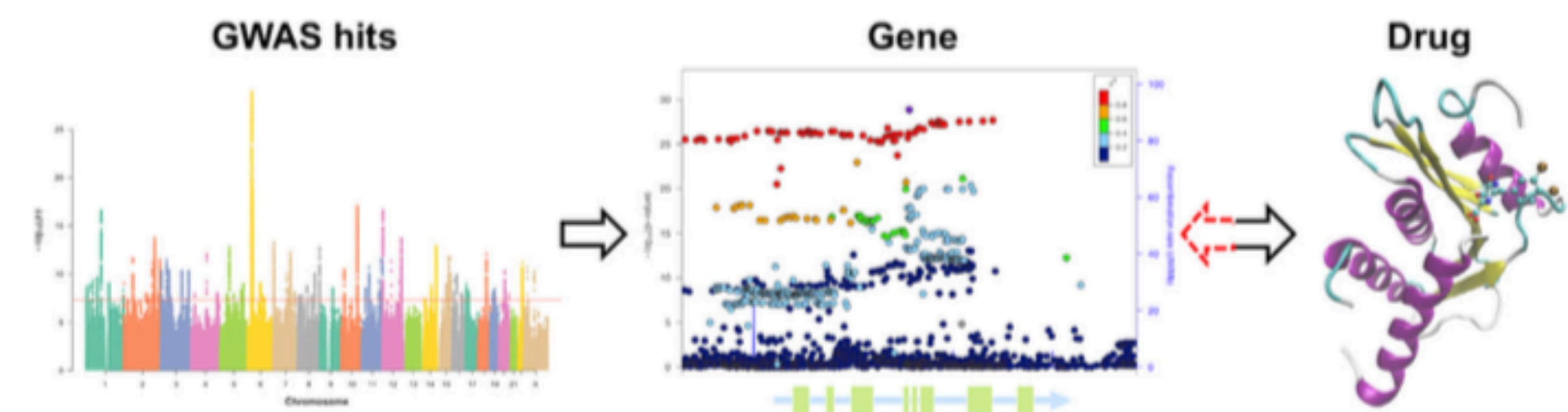
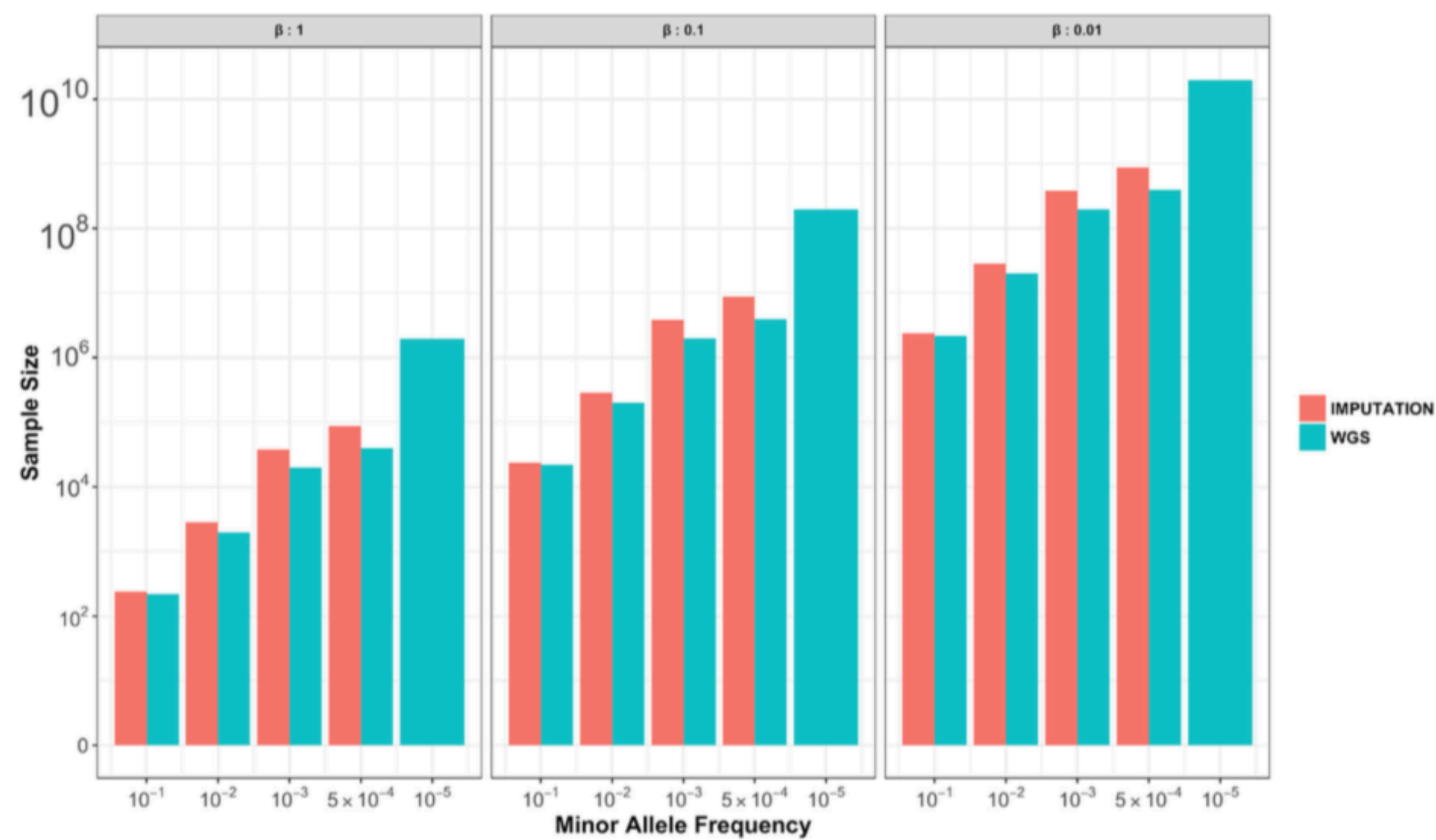
  A/A



$p = 1$	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

- Quanto menor a associação, mais amostras necessárias.
- Quanto menor a MAF (Minor Allele Frequency), mais amostras necessárias.
- Permite identificar genes alvo.
- Desenvolvimento de drogas.



Trait	Gene with GWAS hits	Known or candidate drug
Type 2 Diabetes	<i>SLC30A8/KCNJ11</i>	ZnT-8 antagonists/Glyburide
Rheumatoid Arthritis	<i>PADI4/IL6R</i>	BB-CI-amidine/Tocilizumab
Ankylosing Spondylitis(AS)	<i>TNFR1/PTGER4/TYK2</i>	TNF-inhibitors/NSAIDs/fostamatinib
Psoriasis(Ps)	<i>IL23A</i>	Risankizumab
Osteoporosis	<i>RANKL/ESR1</i>	Denosumab/Raloxifene and HRT
Schizophrenia	<i>DRD2</i>	Anti-psychotics
LDL cholesterol	<i>HMGCR</i>	Pravastatin
AS, Ps, Psoriatic Arthritis	<i>IL12B</i>	Ustekinumab