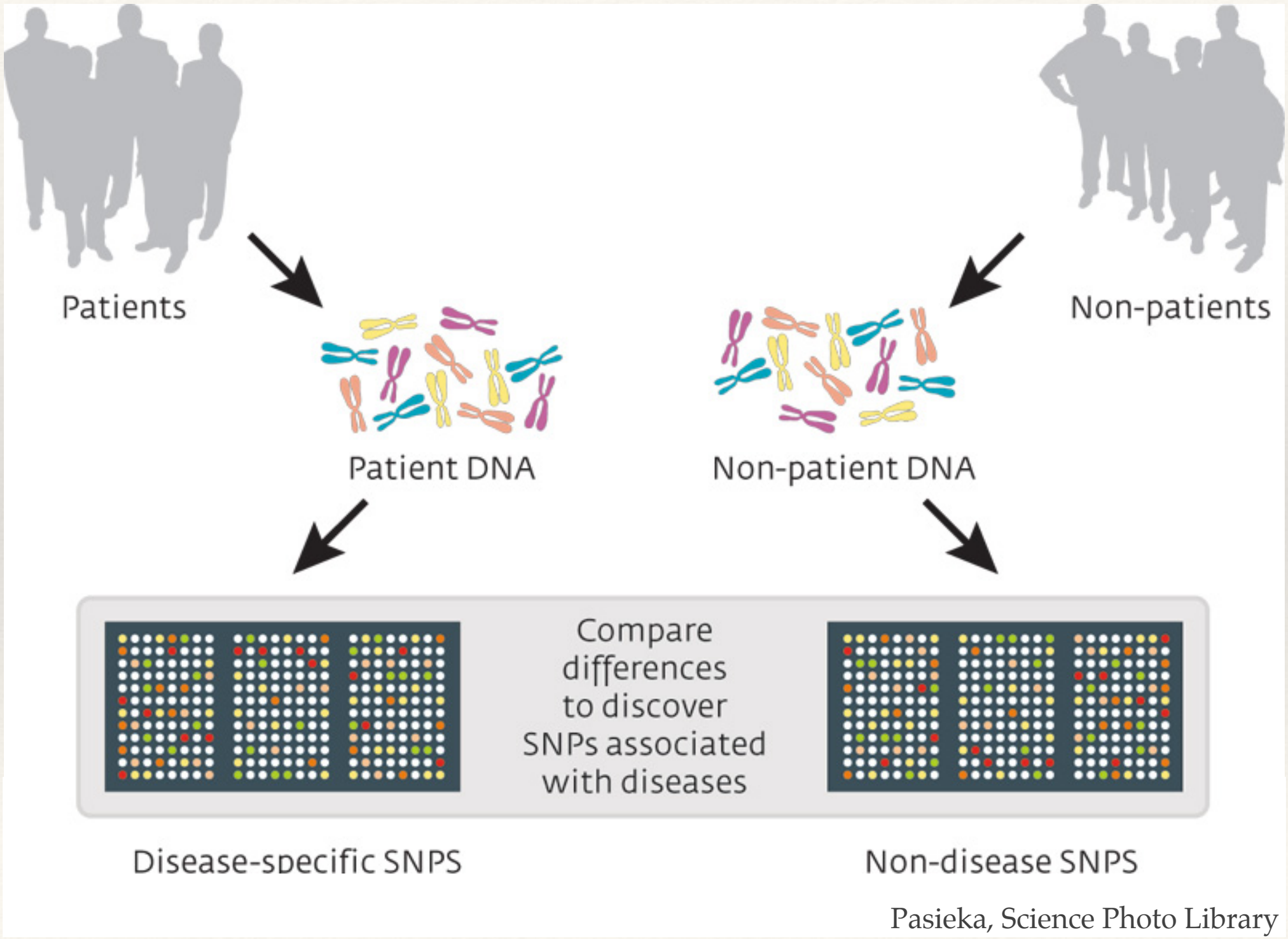
Borrowing from human genetics: Genome-wide Association studies (GWAS)



Pasieka, Science Photo Library

Sequence kernel association test (SKAT)

- Whole-genome sequencing can detect rare variants
- Group variants into genes and testing for association
- Variants can be assigned weights: $f(G_i)$
- Can easily obtain a p-value for each gene

Linear model:

disease/phenotype — covariants (*e.g.* age, sex) — variant sets (*e.g.* genes)

$$y_i = \alpha_0 + \boldsymbol{\alpha}'\mathbf{X}_i + \boldsymbol{\beta}'\mathbf{G}_i + \varepsilon_i,$$

null model:

$$y_i = \alpha_0 + \boldsymbol{\alpha}_1'\mathbf{X}_i + \varepsilon_i$$

Wu et al., *Am. J. Hum. Genet.*, 2011