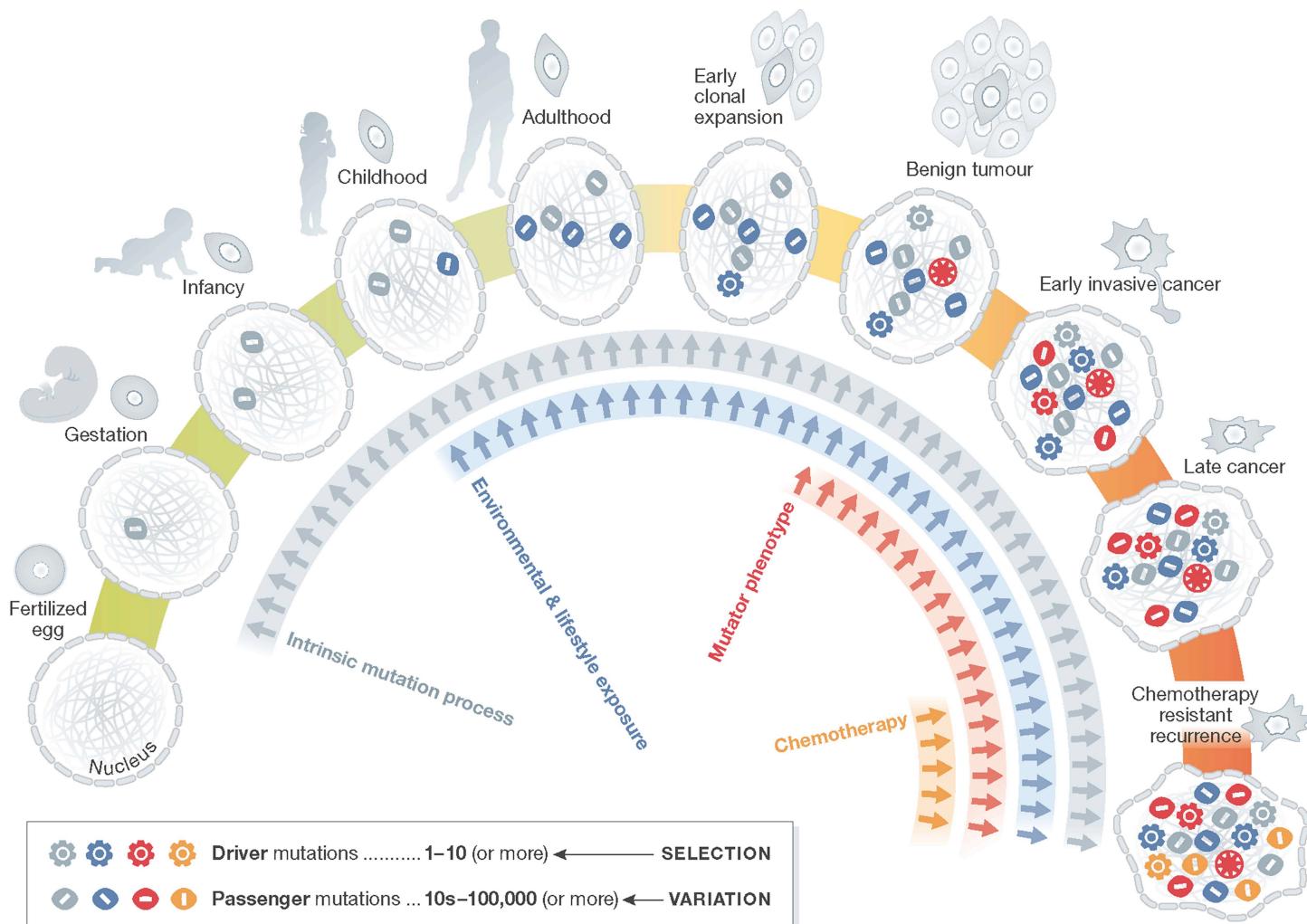


Mutational signature analysis

Introduction

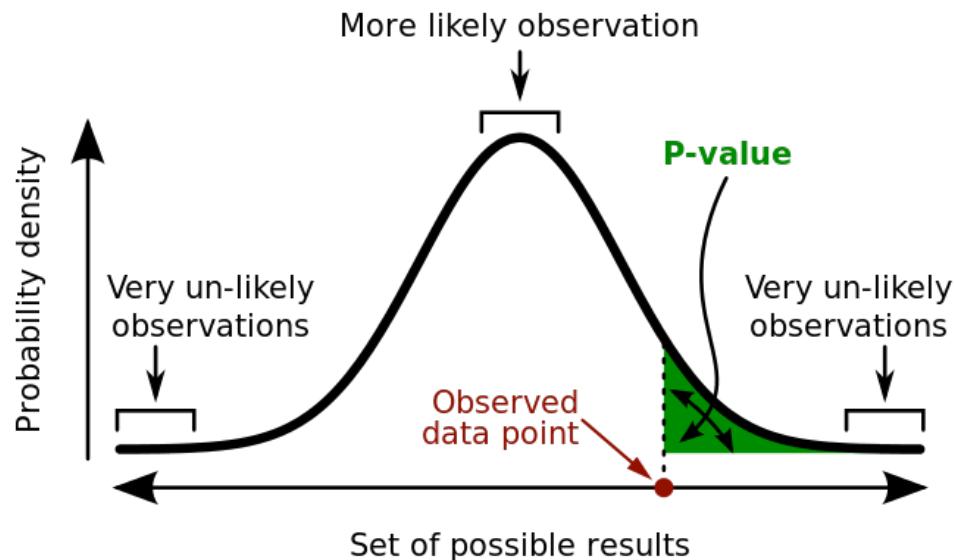
Cohort analysis



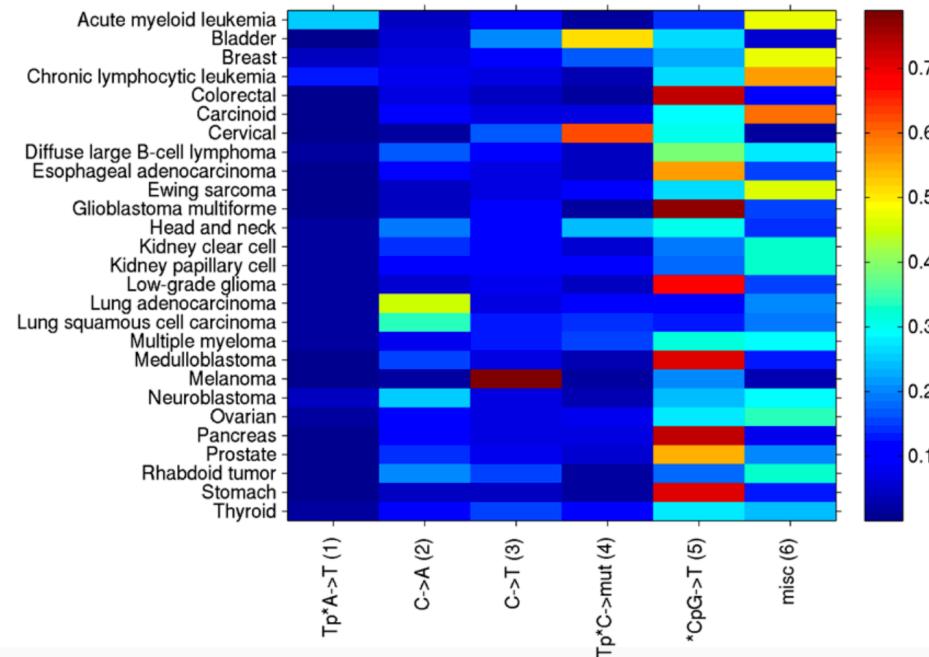
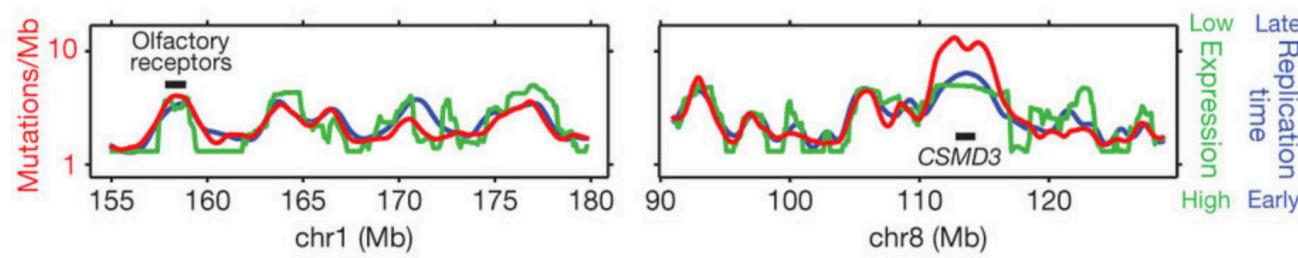
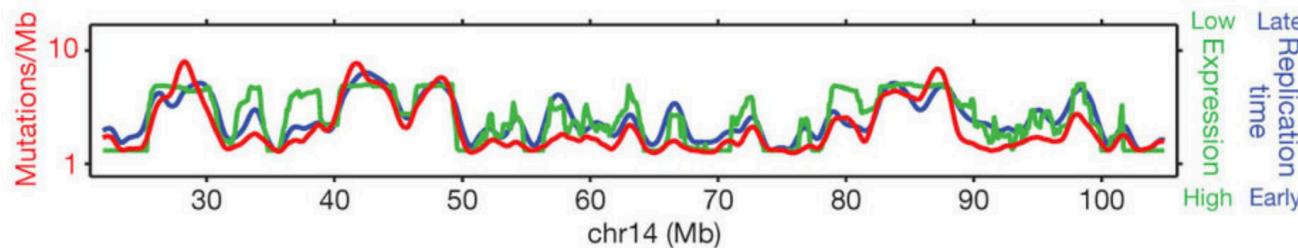
Stratton (EMBO Molecular Medicine 2013)

Frequently mutated genes - MutSigCV

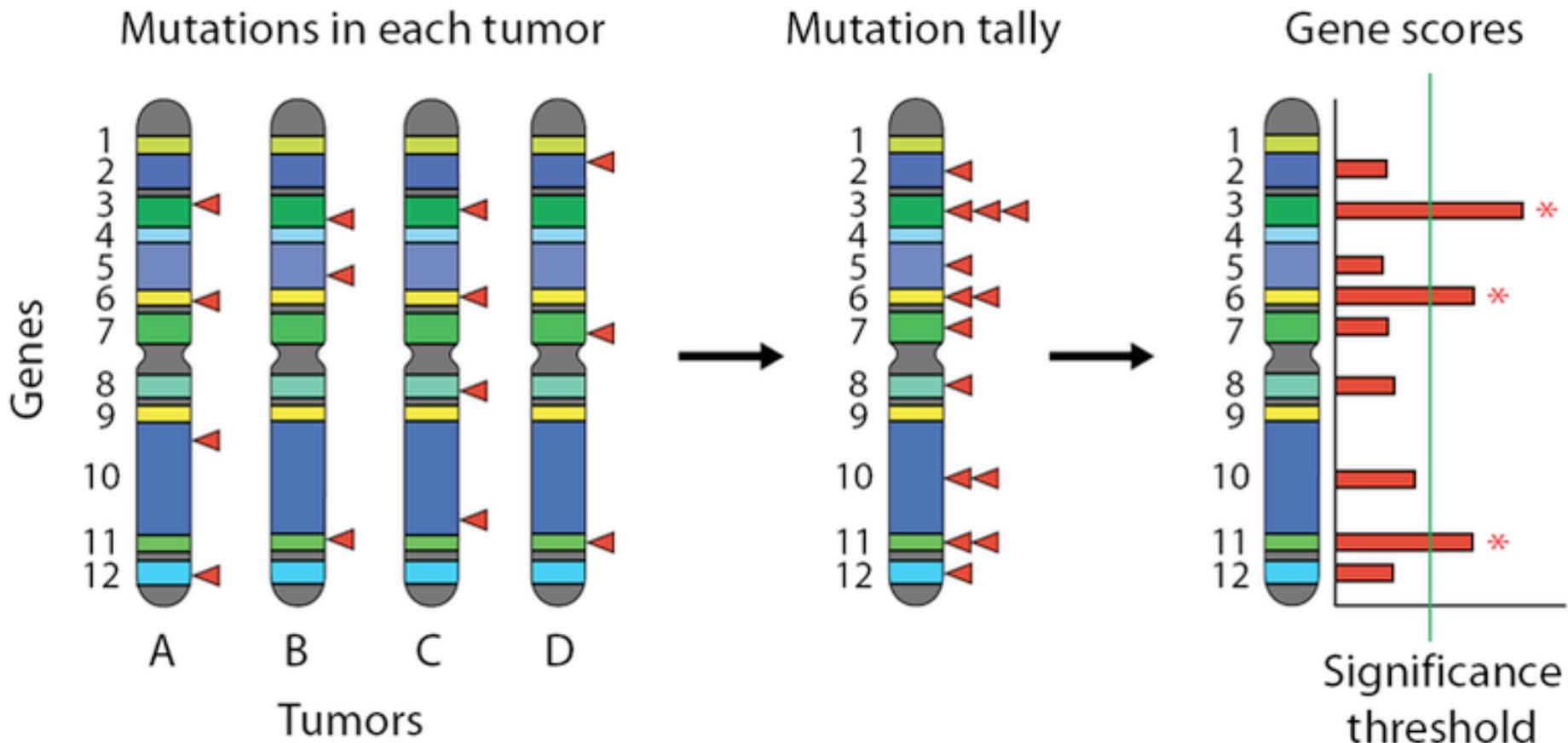
Question:
Which gene is hit by an SNV
more often than expected
in a cohort?



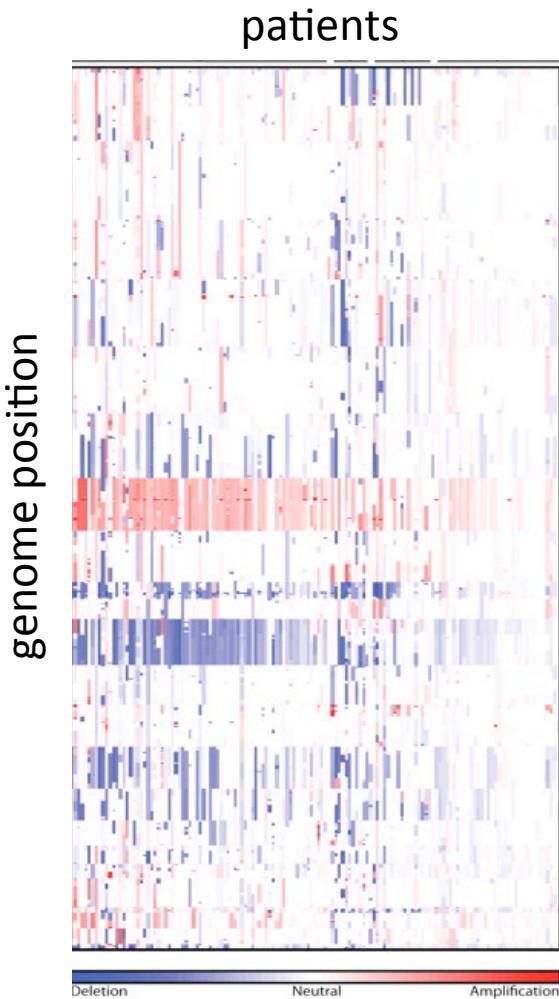
Characteristics of frequently mutated regions



Frequently mutated genes



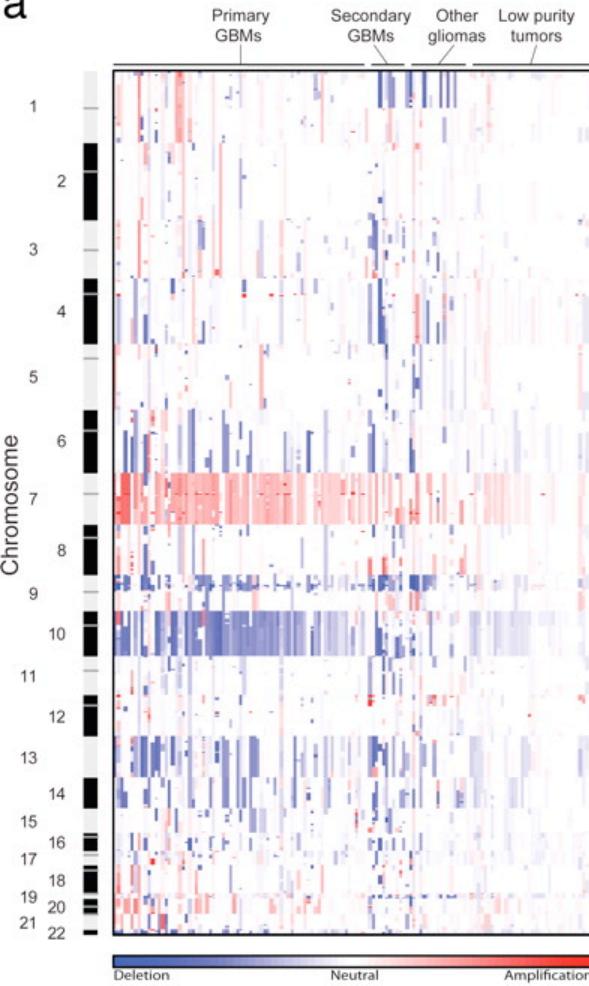
Frequent CNV changes - GISTIC



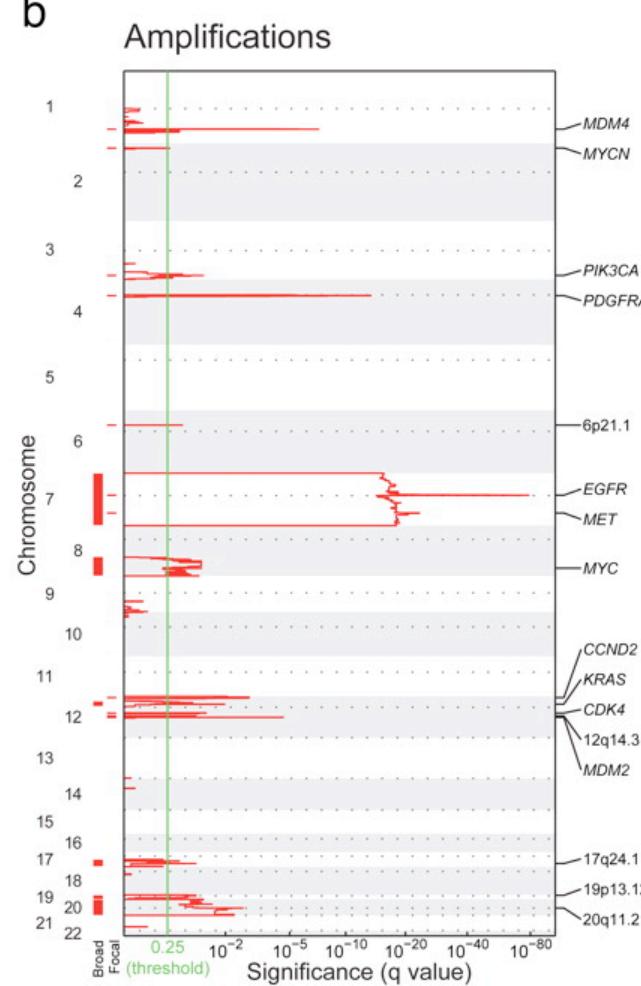
Question:
Which regions are
frequently amplified or
deleted in the genome?

Frequent CNV changes - Gistic

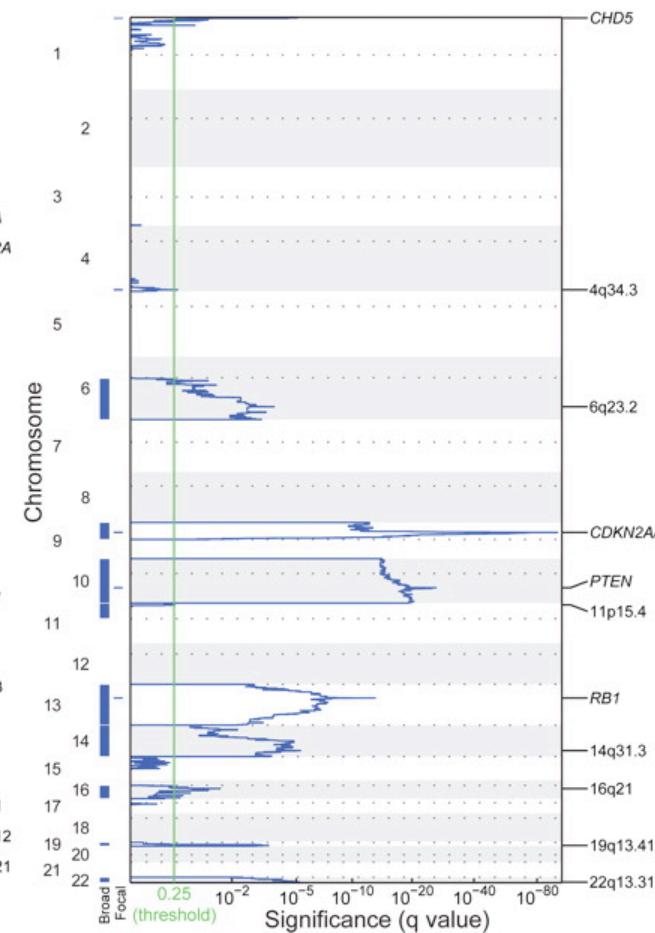
a



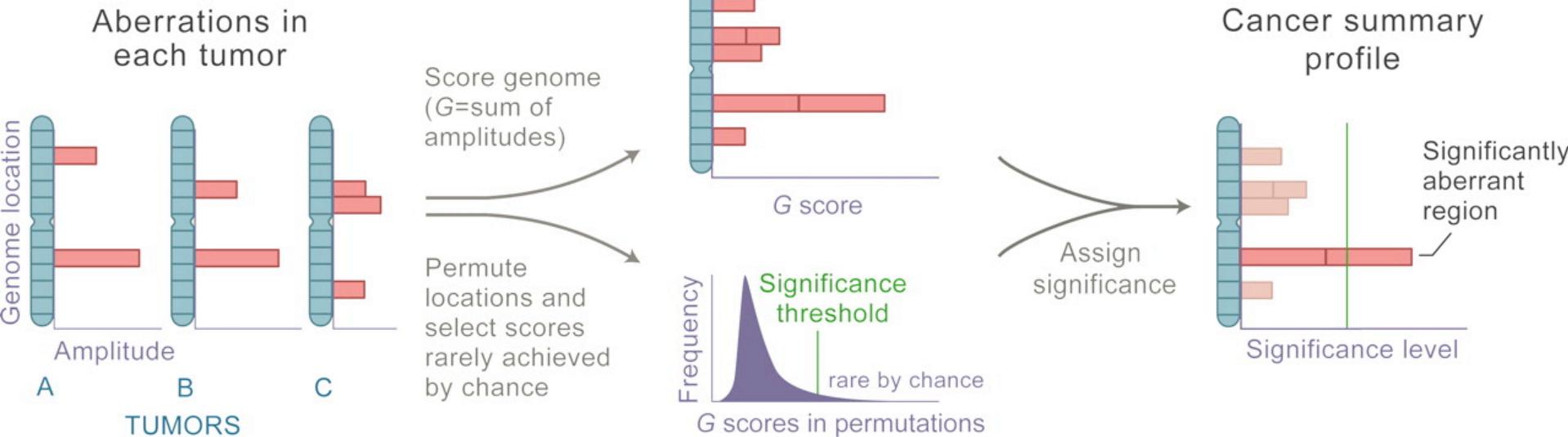
b



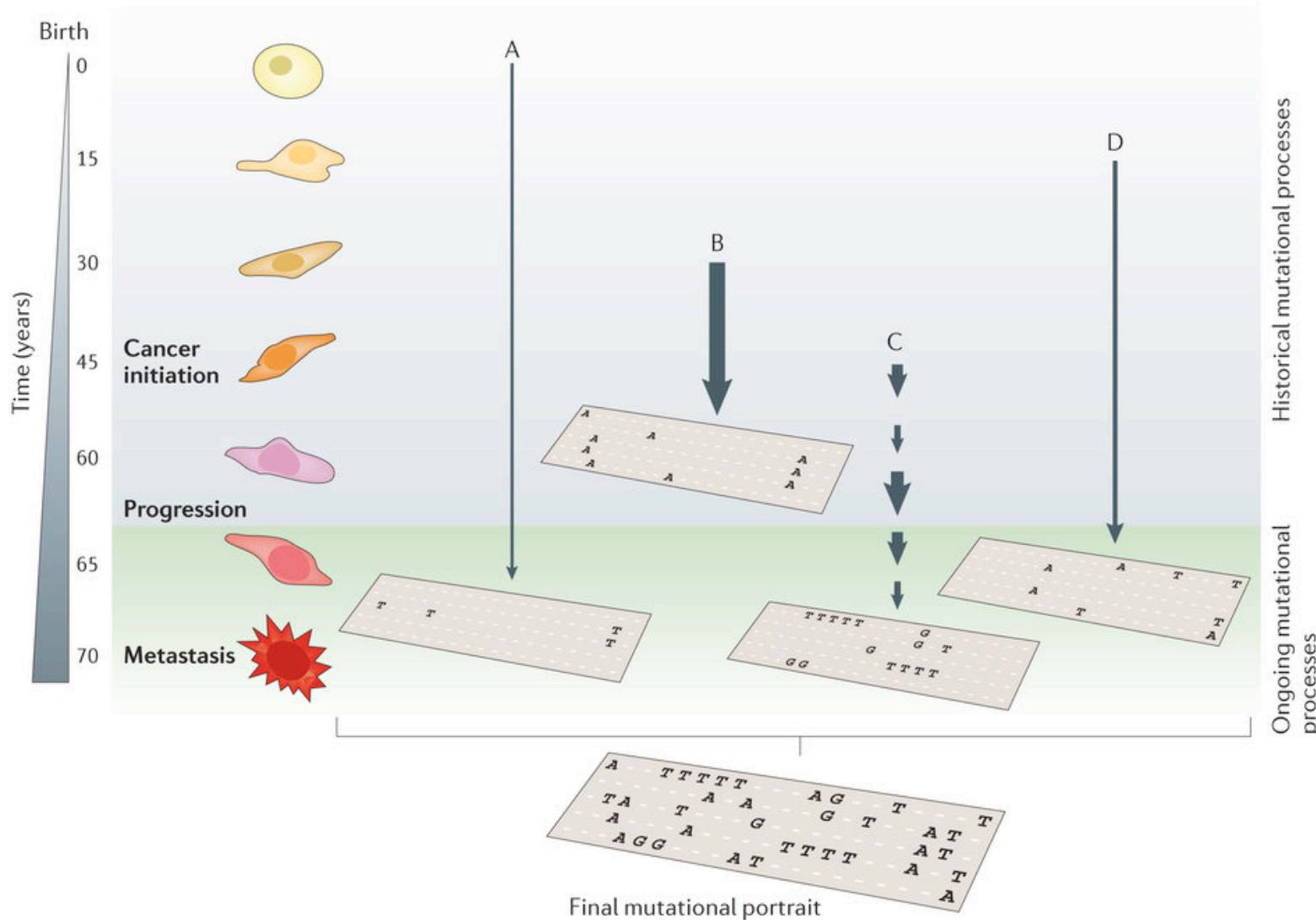
Deletions



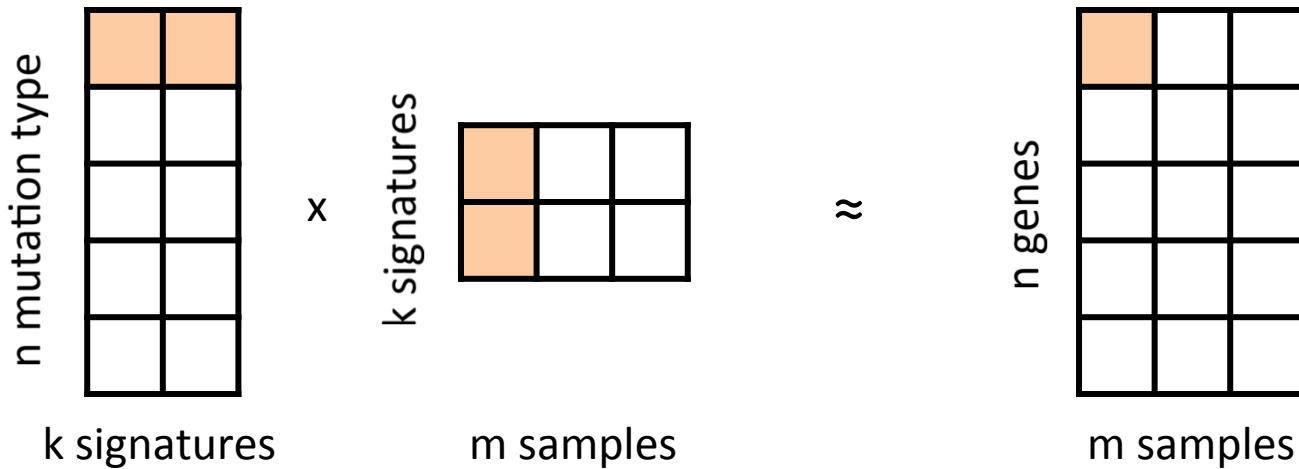
Frequent CNV changes



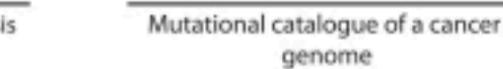
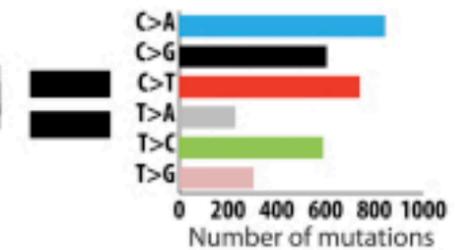
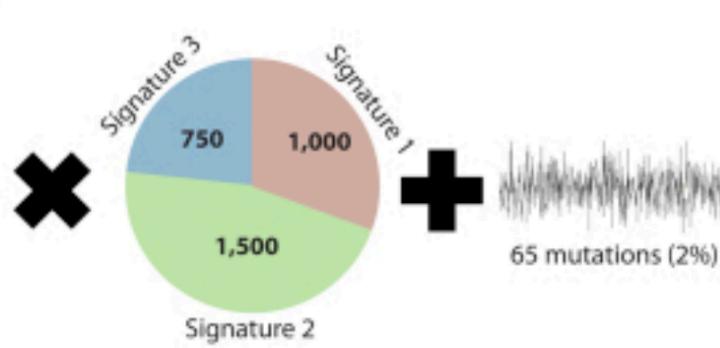
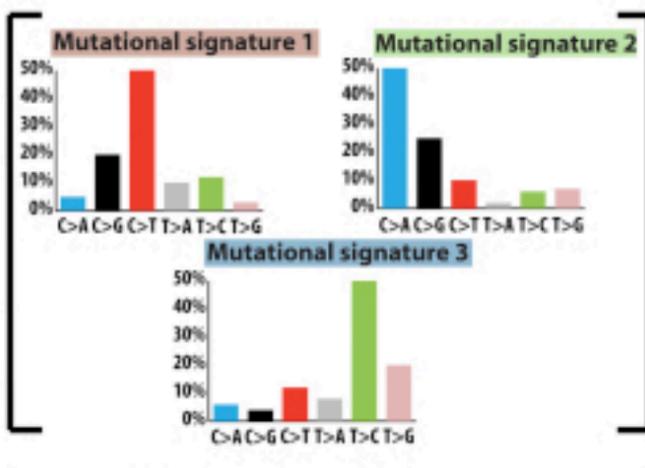
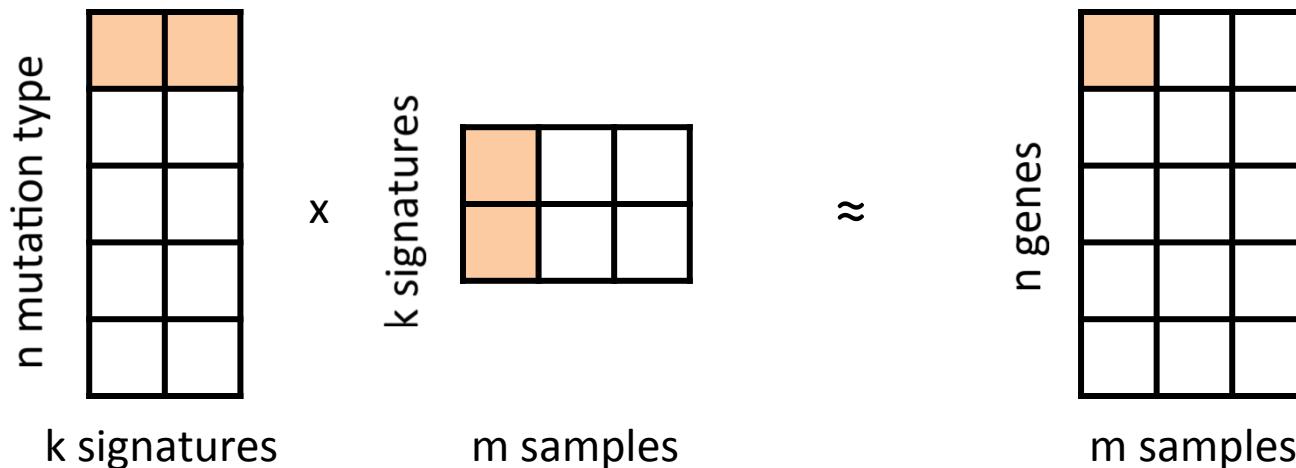
Mutational processes and the genome



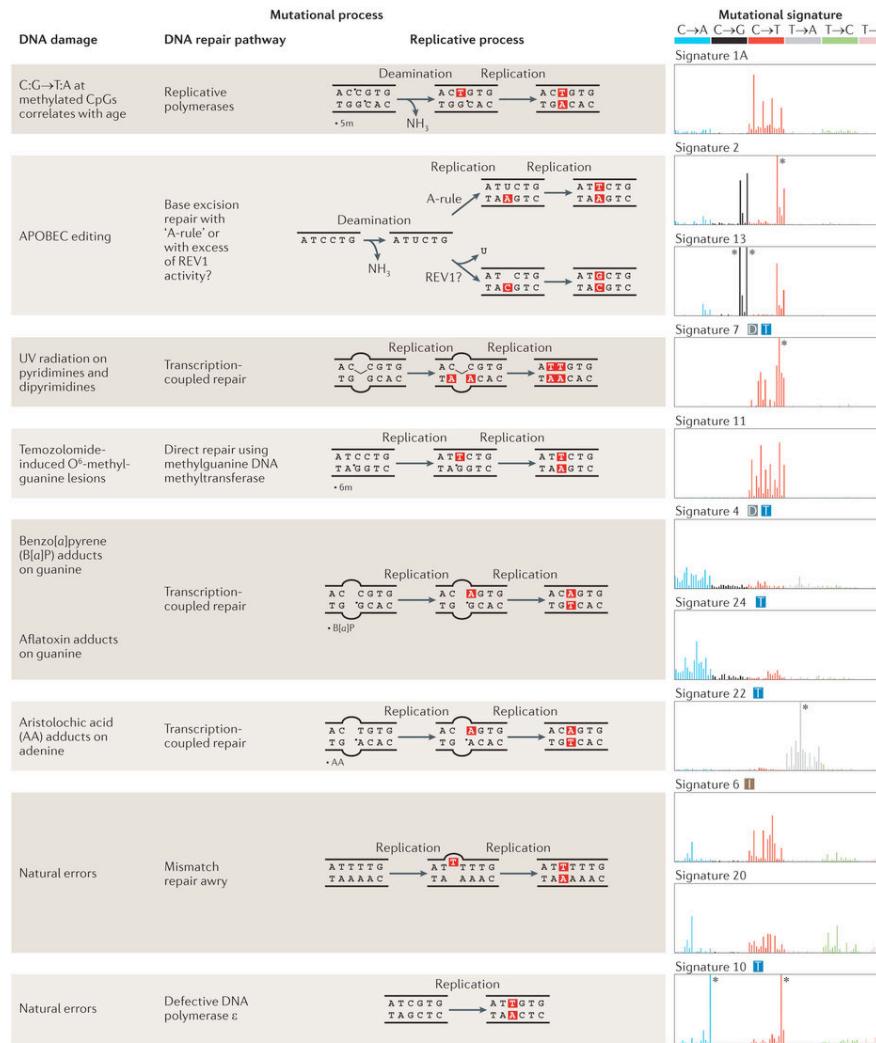
Non-negative matrix factorization



Non-negative matrix factorization



Some known mutational processes



Nature Reviews | Genetics

See also: <http://cancer.sanger.ac.uk/cosmic/signatures>