Molecular taxonomy of disease

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Disease classification

• “Phenotypic” appearance:
  – Cluster of symptoms

• Etiological pathways:
  – Infectious agent
  – Response to treatment
  – Clinical behavior
  – Inherited mutation
  – Molecular portraits
How to identify etiologically distinct disease subtypes?

Etiological pathways → Disease subtypes

Molecular classification

Gene discovery
Molecular classification of tumors

- Immunohistochemistry
- Somatic mutations
- Integrated analyses of chromosomal, genomic, epigenomic, transcriptomic, proteomic profiles
Molecular classification of breast cancer
Challenges

• Understanding biological pathways underlying genetic associations

• Associations between genetic variants and molecular portraits in tissues
  – Tissue heterogeneity

• Collection of tissue samples specimens in well-characterized populations

• Assay costs and throughput