

Molecular taxonomy of disease

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**Frontiers in Population Genomic Research Directions for
NHGRI**

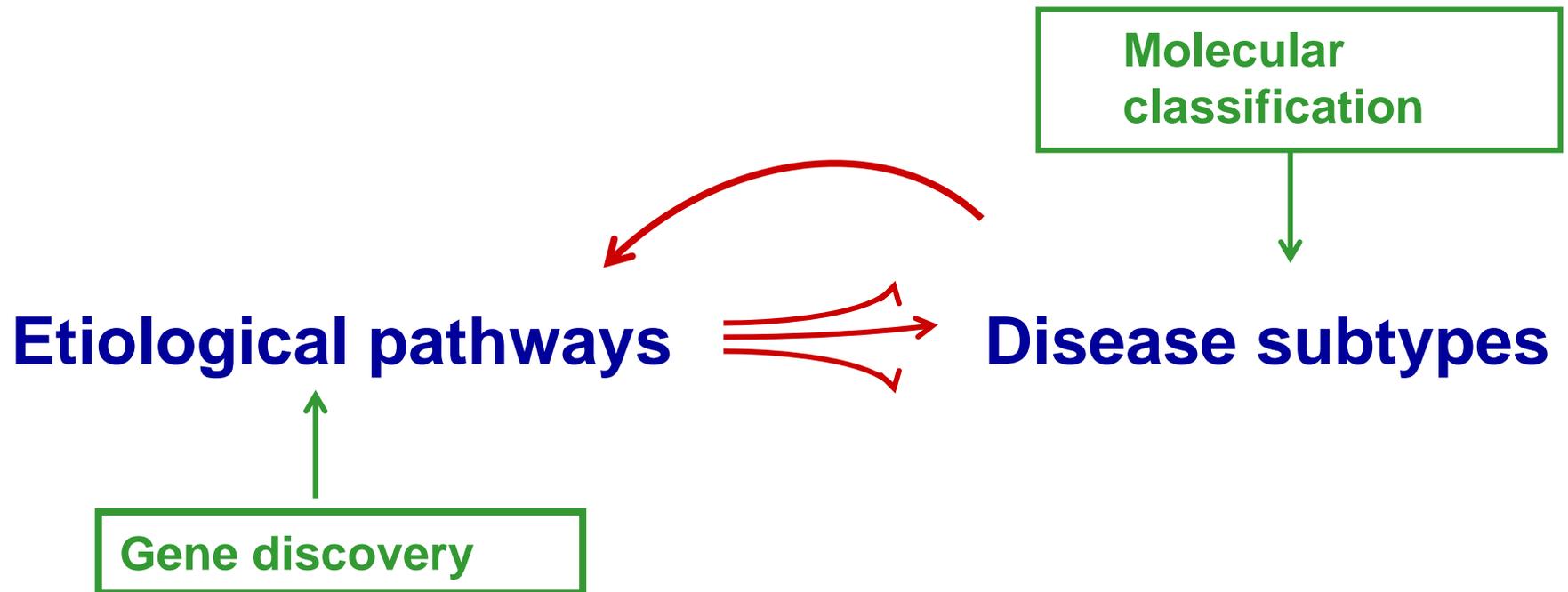
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Disease classification

- **“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 ?



Molecular classification of tumors

- **Inmunohistochemistry**
- **Somatic mutations**
- **Integrated analyses of chromosomal, genomic, epigenomic, transcriptomic, proteomic profiles**

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**