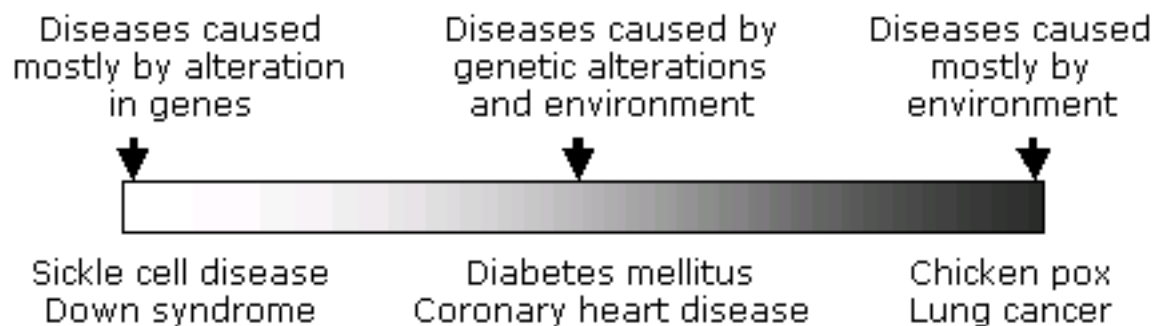


Genetics and Health Overview

Genes influence disease risk across a broad spectrum. At one end of the spectrum are the diseases commonly referred to as "genetic" — diseases like sickle cell disease, Duchenne muscular dystrophy, and Down syndrome, which are caused primarily by genetic differences. At the other end are diseases that are almost wholly determined by environmental exposure — diseases like chicken pox and lung cancer. In between are most diseases, including cancer, dementia, diabetes mellitus, and cardiovascular disease, to which both genetic and environmental influences contribute:



Genetic diseases, at the left of the spectrum, are categorized as **single gene** or **chromosomal** disorders, depending on the [specific genetic cause](#).

Diseases in the middle of the spectrum — including most common diseases — are **multifactorial**, and result from the interaction or additive effect of genetic and non-genetic factors.

Relevance to Primary Care

Most genetic diseases are rare, but as a group they are common enough that many will be seen in primary care.

Knowledge about basic genetic concepts and the more common genetic diseases helps a primary care provider in three areas:

- Critical evaluation of family history
- Appropriate consideration of genetic disorders in differential diagnosis
- Prudent use of genetic testing and medical genetics services

In addition, media coverage of genetics often involves overly simplistic interpretations of genetics as a contributor to disease etiology (e.g., "the

Crohn's disease gene") and unrealistic projections about medical cures. Knowledge about genetics allows primary care providers to help patients distinguish truth from fiction.