Using Evidence-Based Medicine and Genetics

Evidence-based medicine refers to clinical decision making that is based on the best available evidence about health care outcomes.

Scales for evaluating the quality of evidence have been developed:

- The best evidence for evaluating medical interventions comes from well-designed randomized clinical trials (RCTs).
- The best evidence for prevalence and risk assessment comes from large population-based samples.
- The best evidence for diagnostic accuracy of a test comes from studies comparing the test in representative affected and unaffected populations.

Learn More

Applying the Principles of Evidence-Based Medicine to Genetics

- **Prevalence.** Many genetic diseases are rare and, as a result, difficult to study.
- **Genotype-phenotype correlation.** The correlation between a person's genotype (the specific variant(s) present in a gene) and phenotype (the clinical manifestations) is not always predictable.
- **Genetic test characteristics.** Genetic tests may use DNA-based technology to assess genotype, or may use other laboratory techniques to identify findings indicating an inherited condition. Evidence about genetic tests can be assessed in terms of analytic validity, clinical validity, and clinical utility.
- **Outcomes.** Information about the outcome of treatment for genetic diseases is often limited, and RCTs are often not feasible. For many genetic conditions, treatment options are limited or absent. For these conditions, testing may be used primarily to confirm a diagnosis or provide genetic risk information to family members.