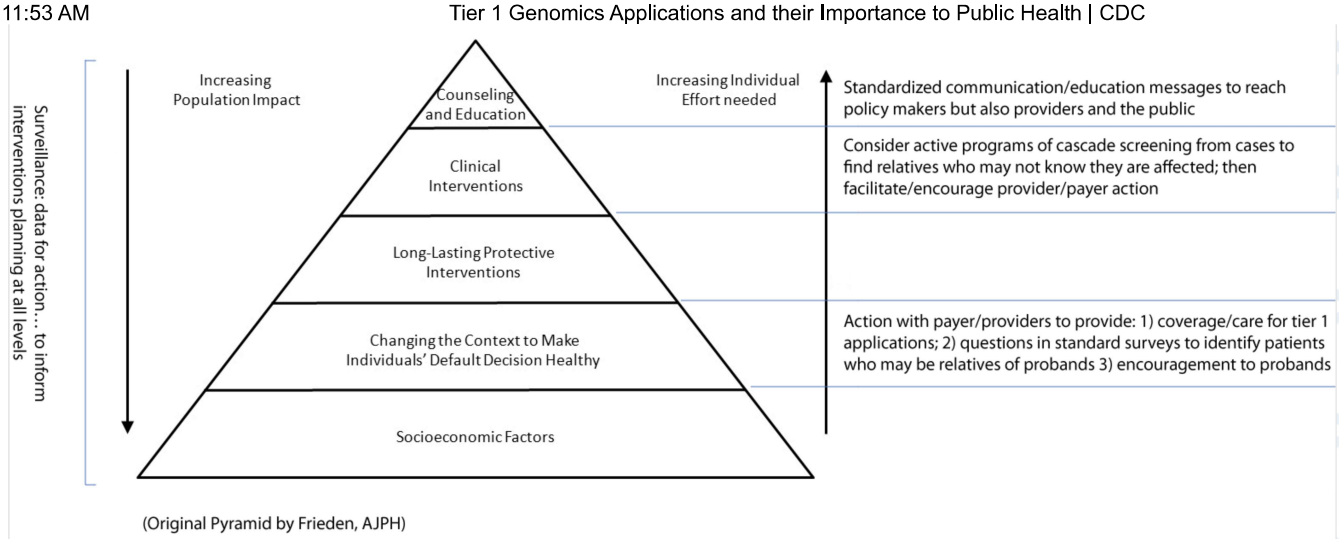
Tier 1 Genomics Applications and their Importance to Public Health

Tier 1 genomic applications are defined by CDC's Office of Public Health Genomics (OPHG) as those having significant potential for positive impact on public health based on available evidence-based guidelines and recommendations. Tier 1 applications are listed in the Genomic Tests and Family History by Levels of Evidence Table which includes a growing number of genomics tests and family health history applications.
Presently, this toolkit focuses on three Tier 1 applications. Nearly 2 million people in the United States are at increased risk for adverse health outcomes because they have genetic mutations which predispose them to one of the following conditions:

- Hereditary Breast and Ovarian Cancer Syndrome (HBOC) – increased risk for breast, ovarian, tubal, peritoneal, and other cancers due to mutations in *BRCA1* or *BRCA2 genes*;
- Lynch syndrome (LS) – increased risk for colorectal, endometrial, ovarian, and other cancers associated with mutations in mismatch-repair genes; or
- Familial hypercholesterolemia (FH) – increased risk for heart disease or stroke due to mutations leading to very high cholesterol levels from an early age

Because, at present, these conditions are poorly ascertained by the healthcare system, many individuals and families affected by them are not aware that they are at risk; however, early detection and intervention could significantly reduce morbidity and mortality. This toolkit currently focuses on these three applications because of the public health burden of the diseases associated with them and because there are clear steps that can be followed to improve health and prevent disease. Public health programs are encouraged to consider examples from states that have initiated programs to date, to work with partners to create or modify programs to fit their own states, and to report on their progress so that others may benefit.

The need for this toolkit and many of the action steps within it were discussed as a result of a working meeting of 80 experts and stakeholders representing federal, state, and local public health agencies, clinicians, key advocates and community leaders who came together in 2012 to develop Tier 1 implementation plans using the Health Impact Pyramid as a guide. The pyramid (see below) describes different health interventions with those at the base providing the greatest potential improvement for a given resource level and the critical importance that policy actions play in the practice of public health genomics at the state level. A report of the meeting is posted on the APHA Genomics Forum webpage .



(Original Pyramid by Frieden, AJPH)

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