



Public Health Screening

Category 1: Newborn Screening

Newborn screening is a nationwide public health program that screens nearly 4 million babies a year for a variety of conditions, as well as hearing screening and Critical Congenital Heart Disease (CCHD). Each state runs a newborn screening program that utilizes dried blood spots, collected soon after the birth of the baby, to screen for genetic conditions. By screening soon after birth, treatment can begin for individuals identified with a newborn screening condition.

Category 2: Expanded Carrier Screening

Carrier screening is defined by the American College of Obstetricians and Gynecologists (ACOG) as “a type of genetic test that can tell you whether you carry a gene for certain genetic disorders. When it is done before or during pregnancy, it allows you to find out your chances of having a child with a genetic disorder.” Carrier screening offered during pregnancy can be offered as a targeted screening or expanded screening.

Category 3: Population Screening

Tier 1 genomic applications [or population screening] 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.”

Category 4: Prenatal Cell-free DNA Screening

Prenatal cell-free DNA screening, examines the risk of a fetus having certain genetic conditions by taking a sample of blood from the pregnant individual. A small amount of the fetal DNA circulates in the pregnant individual’s blood and that DNA can be tested for particular chromosomal disorders.

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