Public Health Genetics Week

Public Health Screening

Public health screening, within the field of genetics, uses biomarkers to screen for genetic conditions in certain populations. Examples include newborn screening, population screening, expanded carrier screening, and non-invasive prenatal screening.

Learn more at:
https://bit.ly/PHGScreening

Newborn Screening

Each state screens newborn babies, a total of nearly 4 million a year across the US, for certain genetic conditions. The newborn screening process includes:

1. Blood test or heel stick
2. Hearing screen
3. Pulse oximetry

Additional Resources:

Newborn Screening Information Center

Navigate Newborn Screening

Population Screening

To identify individuals at higher risk for certain genetic conditions, the CDC recommends screening for three Tier 1 applications that affect nearly 2 million individuals in the United States. Those conditions are:

- Familial hypercholesterolemia
- Hereditary Breast and Ovarian Cancer
- Lynch Syndrome

Additional Resources:

CDC Tier 1 Applications Toolkit

Expanded Carrier Screening

During pregnancy, parents of the fetus can be screened to see if they are carriers for particular genetic conditions that can be passed to their children. Expanded carrier screening offers screening for a number of conditions regardless of the parents' ethnic background or family history. Conditions screened include:

- Cystic fibrosis
- Ehlers-Danlos syndrome
- Spinal Muscular Atrophy (SMA)

Additional Resources:

Genetic Support Foundation

Non-Invasive Prenatal Screening

Non-invasive prenatal screening (NIPS), otherwise known as prenatal cell-free DNA screening, examines the risk of the fetus having certain genetic conditions by taking a sample of blood from the pregnant individual. Conditions screened include:

- Trisomy 13
- Trisomy 18
- Trisomy 21

Additional Resources:

ACMG ACTion Sheets for Healthcare Providers

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