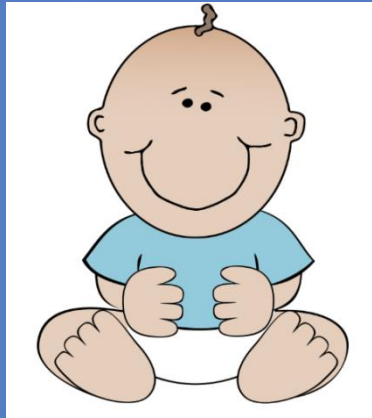
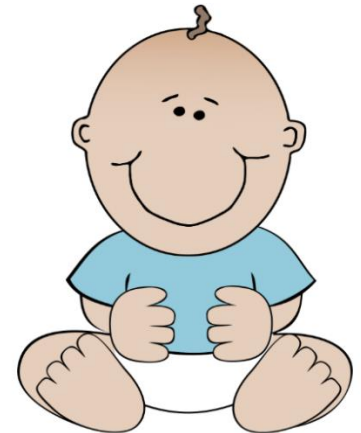




Newborn Screening

Why Do We Need Newborn Screening?

- Many disorders are not obvious or readily diagnosable in a newborn, leading to delay in instituting effective interventions that can prevent or ameliorate irreparable harm



Criteria for Newborn Screening

1. An important health problem whose natural history is understood.
2. Facilities for diagnosis and treatment are available
3. There should be a suitable and acceptable test and treatment.
4. A latent or early symptomatic stage exists during which intervention improves outcomes.
5. The cost of case-finding (including diagnosis and treatment) is economically balanced in relation to possible expenditure on medical care as a whole.

Newborn Screening is a Program, not a Test

Successful newborn screening requires:

- Acquiring the samples and performing the test,
- Having a system for notifying families whose infants test positive,
- Providing follow-up definitive testing and, if confirmed,
- Instituting appropriate management and services.

Newborn Screening – Example

Phenylketonuria (PKU)

- Deficiency of the enzyme phenylalanine hydroxylase prevents conversion of phenylalanine (an essential amino acid) to tyrosine
- If left untreated, blood and brain levels of phenylalanine soar and lead to intellectual and developmental disabilities (IDDs)
- Prior to screening, PKU was one of the leading causes of IDD in the U.S.
- Treating with phenylalanine-restricted diet from birth eliminates IDD
- Today, PKU has been virtually eliminated as a cause of IDD in this country

Newborn Screening is (mostly) “Phenotype Based” and not “Genotype Based”

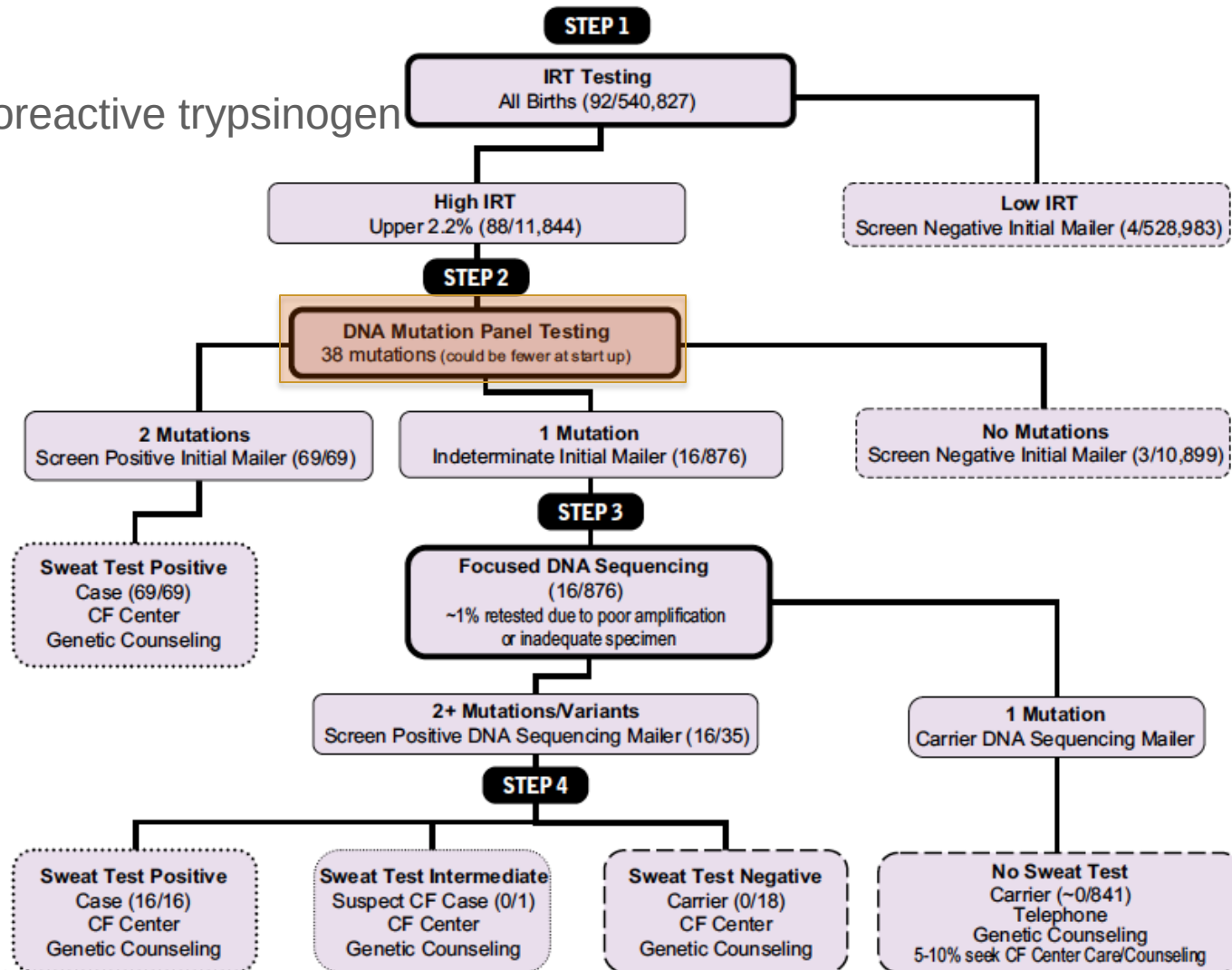
- Deafness
- Cystic Fibrosis
- Hypothyroidism
- Galactosemia
- Hemoglobinopathies
- Fatty Acid Disorders
- Organic Acid Disorders
- Amino Acid Disorders
(>40 disorders by Tandem Mass spectroscopy)
- Congenital Adrenal Hyperplasia
- Biotinidase deficiency
- Severe Combined Immunodeficiency
(T-cell lymphopenia)

Newborn Screening – Scope

- Varies by state
- Uniform panel of 31 core disorders and 26 secondary disorders currently recommended by the US Secretary's Advisory Committee on Heritable Disorders in Newborns and Children
- 2003: all but 4 states screening for only 6 conditions
- 2013: all states screening for more than 30

Example: California Cystic Fibrosis Newborn Screening Includes Gene Testing

IRT-Immunoreactive trypsinogen



Newborn Screening – A Success Story

- Virtually all of the 4,000,000 babies born in the United States receive some degree of screening but which tests are done differs between states.
- 12,000 babies in the United States screen positive and are confirmed to have one of the disorders for which screening is done
- Every \$1 spent on screening results in savings of \$10-20 by reducing medical costs, averting expenses for special services, and increased productivity
- Screening is available in many other countries, but not all, with a varying menu of conditions for which screening is provided

Future of Newborn Screening – Many Contentious Issues

- Should we be adding more disorders and do we need to better define, refine, or expand criteria for utility?
- Should we doing screening to find parents at risk for disease in future children early enough to inform their reproductive decision making?
- Introduction of DNA sequencing – adjunct to or replacement for some phenotypic screening?