OBSTETRICS & GYNECOLOGY

**Clinical Expert Series**

# Newborn Screening and the Obstetrician

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1. According to the authors, the ideal disorder for genetic screening is one in which:

   A. Intervention prevents later disabilities for infants who may appear normal at birth
   B. An abnormality is readily confirmed at birth
   C. In utero intervention prevents the disorder
   D. The invariable result is neonatal or infant death allowing for early termination
   E. There is a greater than 1 in 300 chance of finding the disease

2. The first newborn screening test to be widely accepted was:

   A. Phenylketonuria (PKU)
   B. Group B *streptococcus*
   C. Herpes simplex virus (HSV)
   D. Rh-negative blood type
   E. Type II diabetes mellitus

3. During this century, in many states newborn screening has been expanded to include:

   A. Fungal diseas
   B. Aneuploid
   C. Chromosom breaks
   D. Hearing disord s
   E. Vision disorders



4. Most newborn screening is perform d on:

   A. Urine
   B. Frozen serum
   C. Cord blood
   D. Stool samples
   E. Filter paper blood samples

5. The first step in the follow-up of an initial positive newborn screen ng test should be:

   A. Genetic counseling
   B. Referral to a specialist
   C. Diagnostic testing
   D. Long-term care planning
   E. Maternal testing

6. Exceptions to the usual protocol for newborn blood screening in the first 24 hours of life include:

A. Prematurity
B. Male circumcision
C. Breastfeeding
D. National holidays
E. Cesarean delivery

7. Fatty acid disorders, such as medium chain acyl-coA dehydrogenase deficiency (MCADD), generally manifest symptoms when children:

A. Reach puberty
B. Are fasting or have increased energy demands
C. Ingest fatty foods
D. Are fed commercial milk formula
E. Enter school

8. The most common inherited life-shortening disease of childhood onset in the United States is:

A. Sickle cell anemia
B. Cystic fibrosis
C. Phenylketonuria (PKU)
D. Duchenne muscular dystrophy
E. Fragile X syndrome

9. The definitive confirmatory test for cystic fibrosis is:

A. Immunoreactive trypsinogen concentration
B. Single mutation analysis
C. Serum sodium-to-chloride ratio
D. A sweat chloride test
E. White cell activation test

10. Though yet to be implemented, recommended newborn screening for critical congenital heart disease is based upon:

A. Three-dimensional ultrasonography
B. Congenital anemia
C. Pulse oximetry
D. Enzyme defect detection
E. Metabolite accumulation

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