

## NEWBORN SCREENING

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- ▶ **Overview**
- Rationale for newborn screening
- Difference between a screening test and diagnostic test
- Types of screening tests available
- Limitations and pitfalls of screening



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### WHAT IS A SCREENING TEST?

A screening test

- 1
- 2
- 3
- 4

A diagnostic test

- ▶ Confirms or excludes the presence of a ...
- ▶ Should produce no false-positive or ...

Newborn Screening Tests 

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**WHY SCREEN NEWBORNS?**

Screening tests for conditions that are serious, identifiable, and treatable:

- 1
- 2
- 3
- 4
- 5
- 6



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**HOW IS NEWBORN SCREENING DONE?**


Blood samples taken during first few days after birth

Tandem mass spectrometer performs ...

Second-tier testing after initial abnormal...

- 1
- 2
- 3
- 4

Point of care testing at bedside for ...



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**CHILDREN WITH POSITIVE SCREEN**

States decide how to handle + screen results

Process should include

- 1
- 2
- 3
- 4
- 5

Families often experience...



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### CHILDREN WITH CONFIRMED DISEASE

Child seen by specialty physicians, including...

Additional testing to confirm specific diagnosis of...

Long-term care and therapy for...

Ongoing counseling to...


Ultimate goal: improve:

A.

B.

C.

Genetic testing may be recommended for...



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### DEVELOPMENTAL DISABILITY

Diseases can result in secondary neurodevelopmental & functional outcomes:

1.

2.


3.

4.

Children identified with disease in newborn screening instead of later Dx:

A.

B.



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### HOW CAN SCREENING FAIL?

Newborn not screened at hospital, transferred to...

Specimen lost or...

False-negative results based on...

Blood transfusion or other blood...

Infants "lost" to follow-up because of


1.

2.

3.

False-positive rates higher in...

Infant carriers who are unaffected



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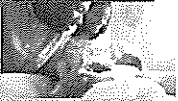
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### PAST, PRESENT, & FUTURE OF NEWBORN SCREENING

1969-1975:  
Development of successful...



Today more than 4,000,000 babies...

States have various requirements, most...

Consumer activism & new tech has driven further expansion of...

More D/O's will likely be added to...

Legal, ethical, & social concerns will have to be...

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### SUMMARY


Screening tests are important tools to define increased risk for medical and genetic conditions.

- 1.
- 2.
- 3.

Early screening often results in successful tx & outcomes.

Screening will continue to increase over time.

Screening tests for conditions that are serious, identifiable, and treatable.



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