

# International Best Practices in Early Screening, Diagnosis & Intervention

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# Today's session will review:

- Rationale for and recommended practices in newborn infant screening
- Early developmental screening recommended practices and strategies
- Early intervention recommended practices and suggestions

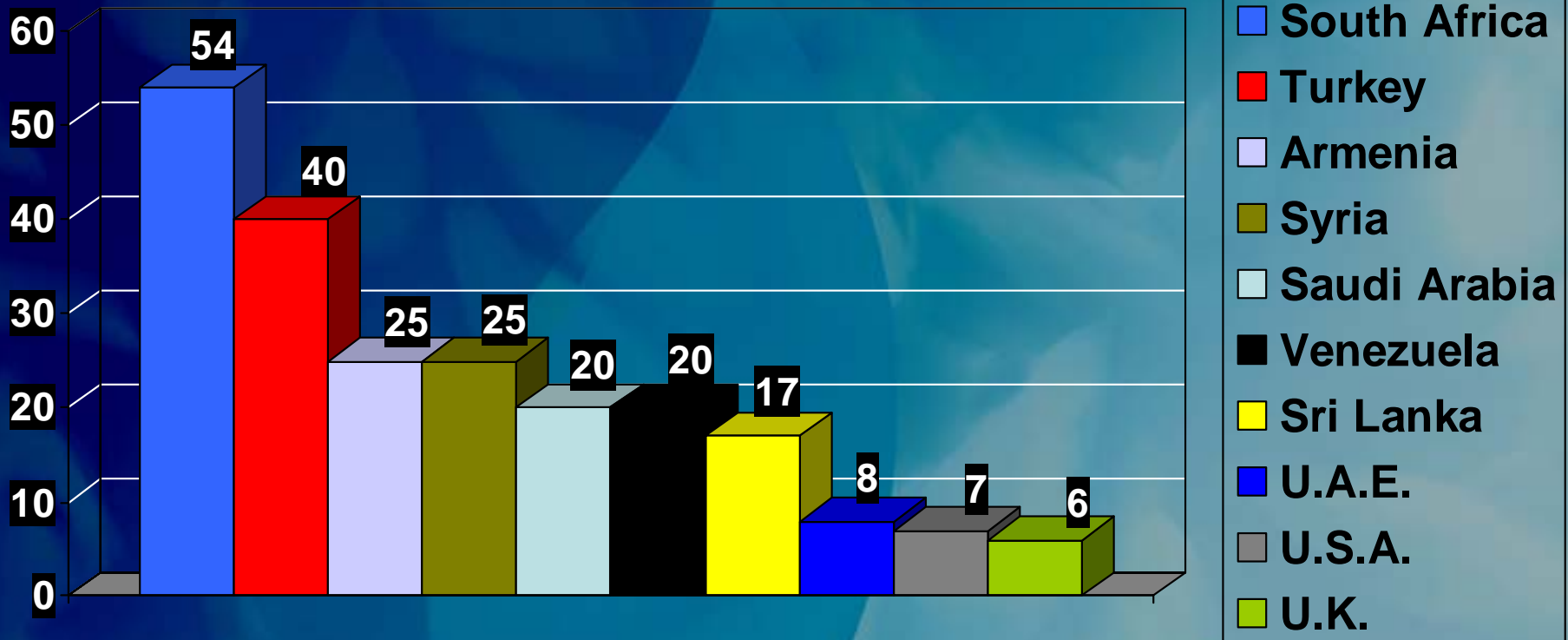


# Another way to look at these topics:

- Prevention
- Early Detection
- Early Intervention

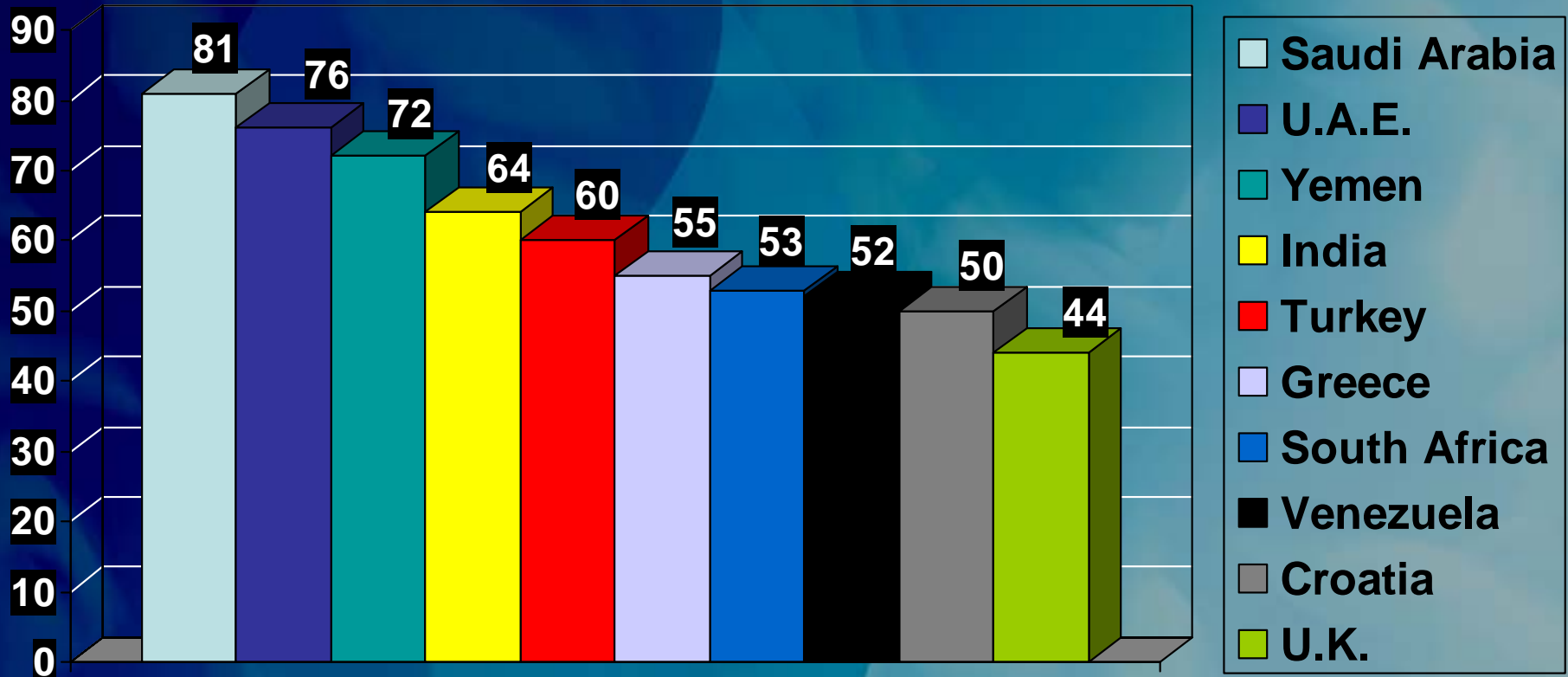


# Select Infant Mortality Rates Per 1,000 Births Worldwide (1999)



Source: UNICEF (2001).

## Birth Defect Prevalance Per 1,000 Live Births



Source: *March of Dimes Global Report on Birth Defects (2006).*



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# Benefits of Newborn Screening

- Early detection can significantly reduce the rate of infant mortality due to birth defects
- Early detection can ameliorate or cure up to 40% of infants born with a birth defect
- Early detection can significantly reduce the cost of services to children with disabilities attributable to birth defects



# Newborn Screening

- Screening procedures conducted at or very soon after the birth of a child
- Usually determining the presence of possible disorders related to:
  - Genetics
  - Metabolism/enzyme processing
  - Hormonal production
  - Function



# Methods of Newborn Screening

- Primary method is a blood stick screening for different disorders
- Hearing screening occurs in hospital
- In the U.S. in 2002 over 4.3 million infants were screened:
  - over 3,300 were found to have a metabolic disorder
  - over 12,000 infants were found to have a hearing loss



# Heel Stick Blood Testing

Methods include:

- Bacterial Inhibition Assay (BIA)
- Tandem Mass Spectrometry (MS/MS)
- Fluorometric Assay (FA)



# Heel Sticks

- Conducted shortly after birth
- Quick turn around time for receiving results
- Generally low false positive test results



Many thanks to Dr. Alaskary and the Kingdom of Saudi Arabia for the photos of the heel stick procedures.



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# American College of Medical Genetics Recommended Screenings

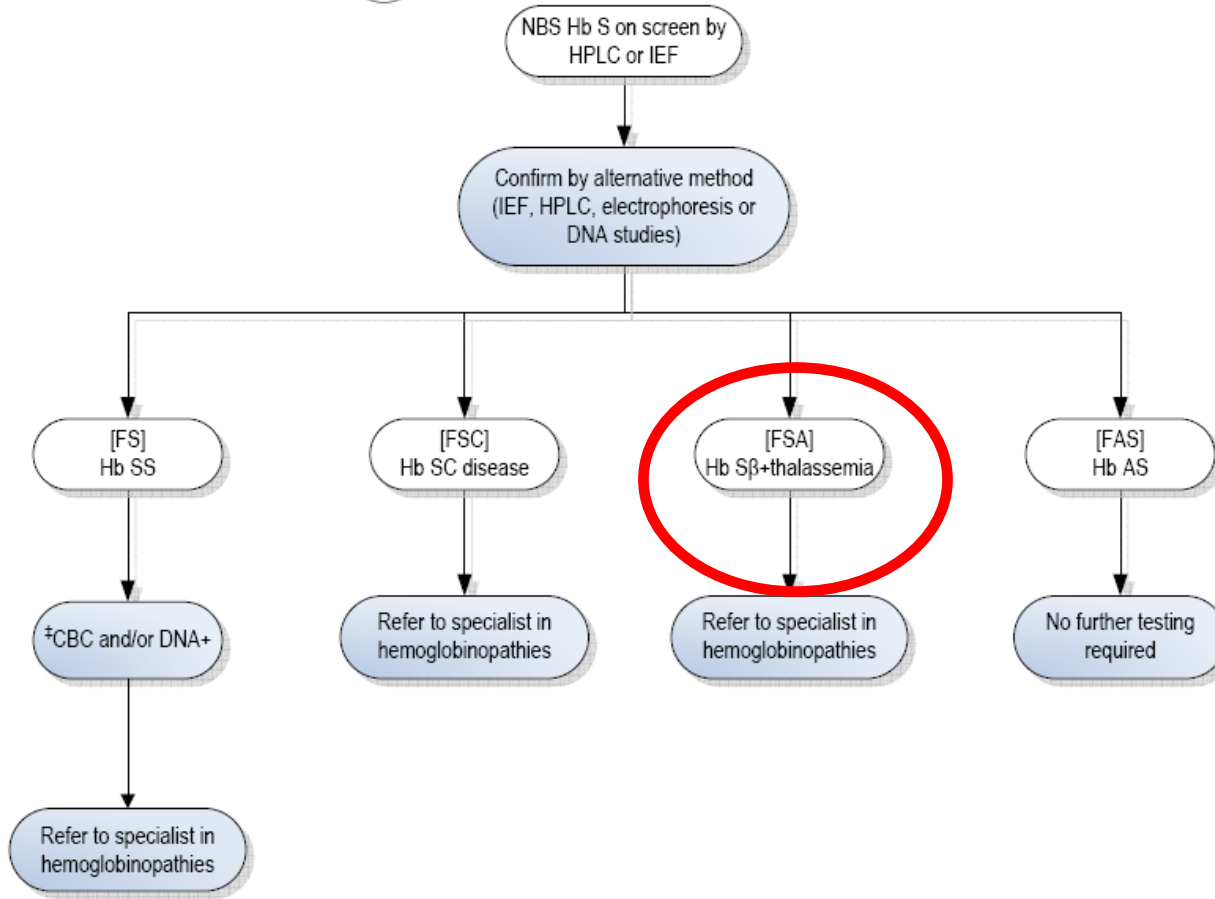
- Seven screening categories for 29 core disorders
  - *Endocrinopathies* (e.g., primary congenital hypothyroidism)
  - *Hemoglobinopathies* (e.g., sickle cell anemia)
  - *Genetic conditions* (e.g., cystic fibrosis)
  - *Galactosemias* (e.g., classic galactosemia)
  - *Fatty acid oxidation* (e.g., glutaric acidemia)
  - *Organic acidemias* (e.g., malonic aciduria)
  - *Amino acidemias* (e.g., phenylketonuria, maple syrup urine disease)



# Example of Screening Algorithm



## Hb S Screening

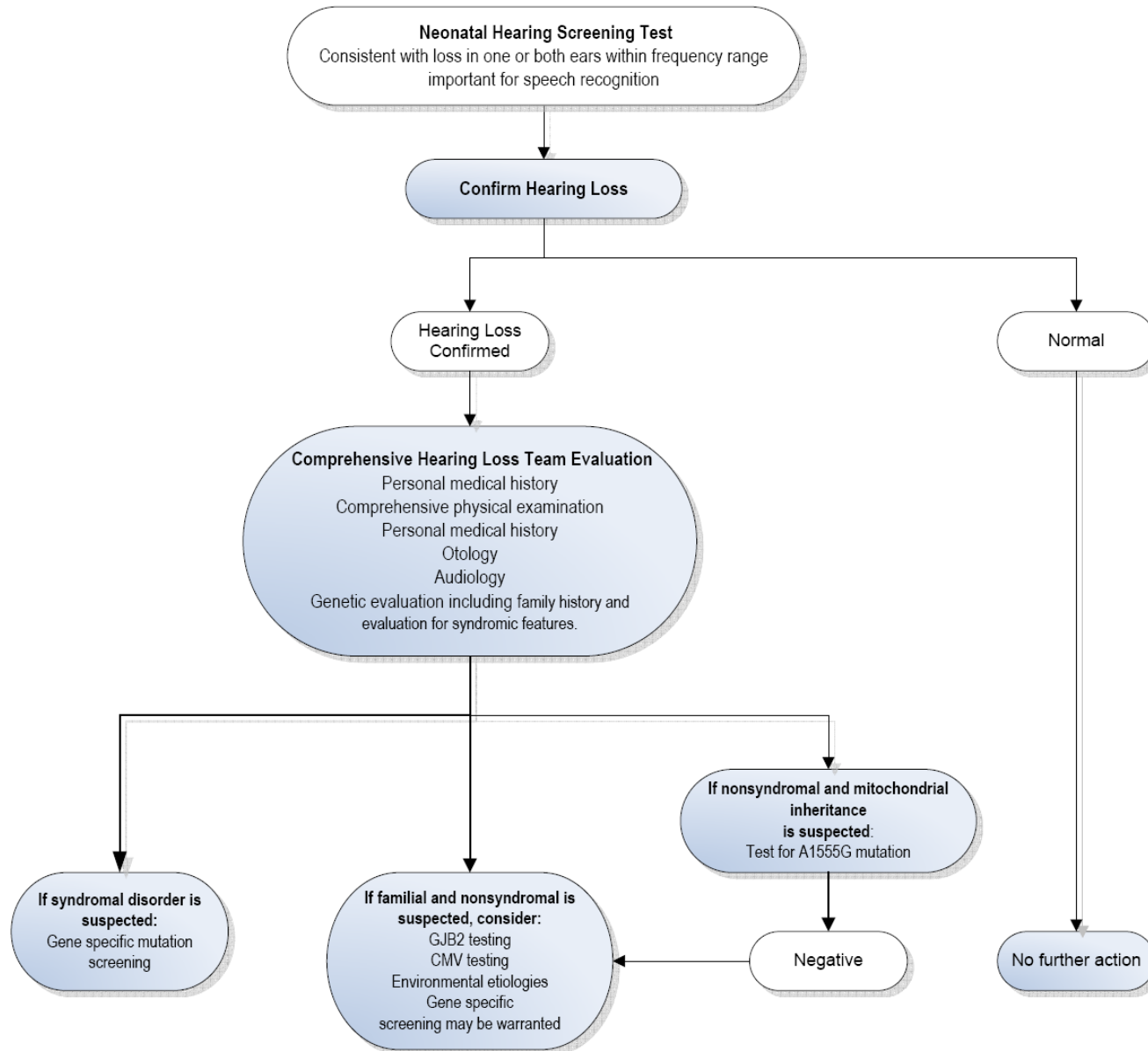


- Hb S present in screening; specific hemoglobin types identified to determine d





# Hearing Loss



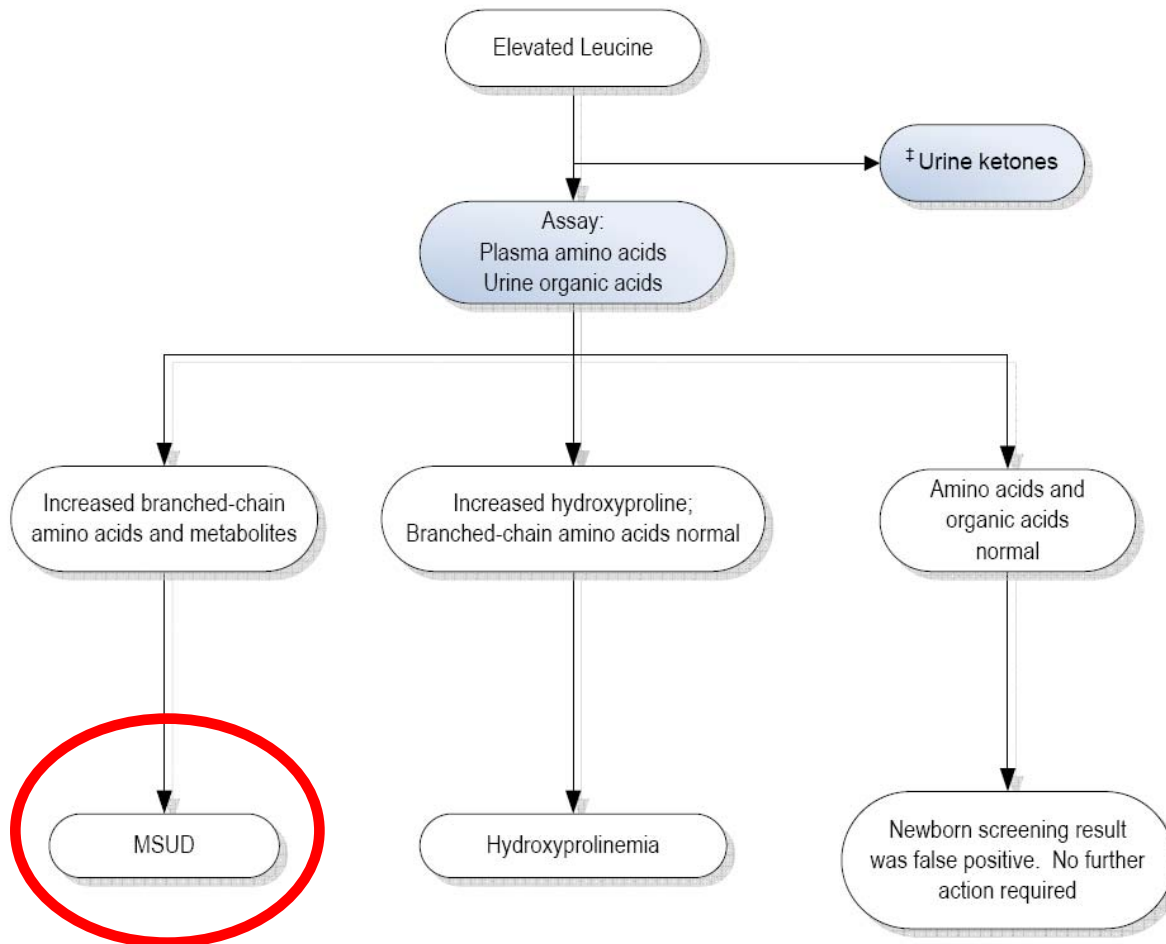
- Example of a screening algorithm leading to multiple investigations of etiology of the disorder



# Example of a Newborn Screening Algorithm



## Leucine Elevated



- Elevated *Leucine* as a marker for Maple Syrup Urine Disease

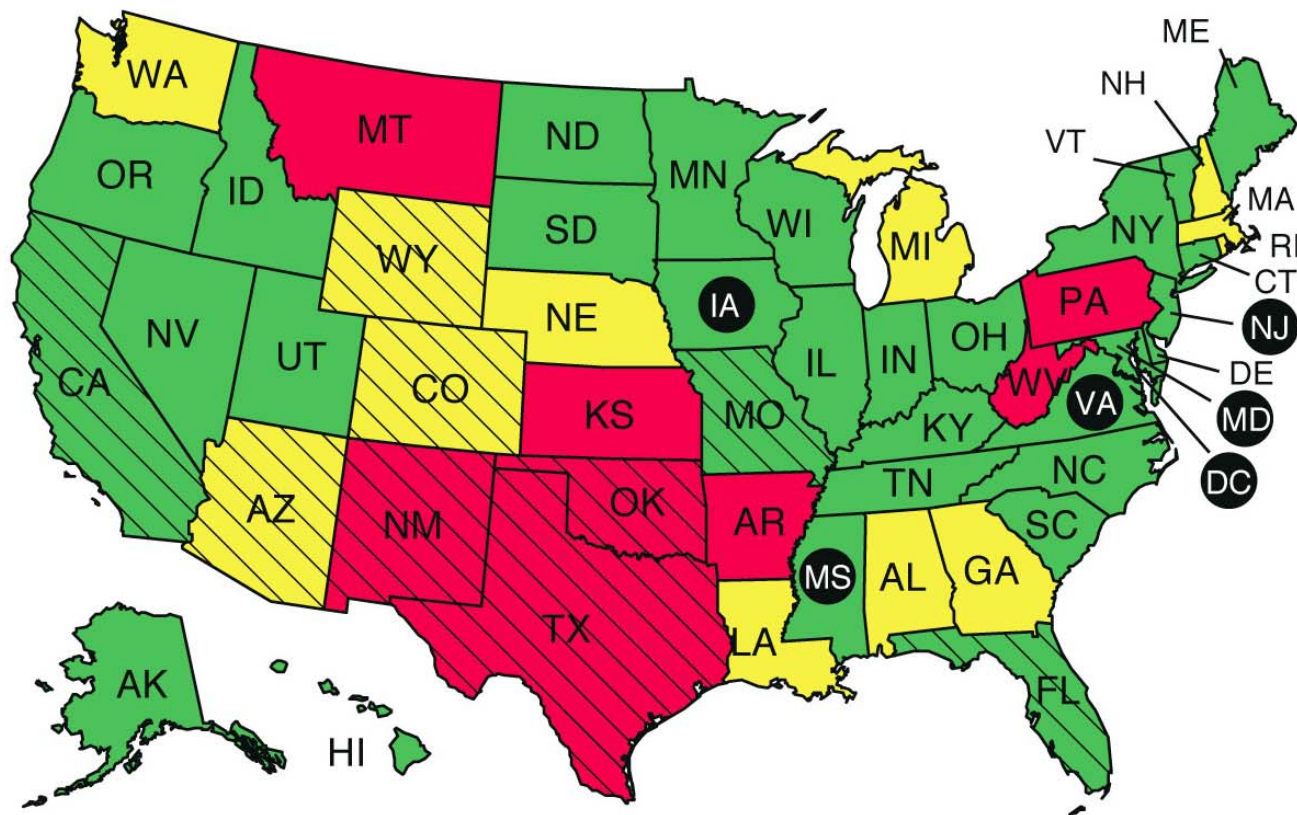


# Increased *Leucine*

- Clinical intervention leads to primarily typical development of the infant
- Non-diagnosis results in:
  - Irreversible cognitive disorders
  - Feeding intolerance
  - Failure to thrive
  - Seizures
  - Cerebral edema
  - Possible death



# Newborn Screening Tests by U.S. States, 2006



- More than 20 core conditions (31)
- 10–20 core conditions (12)
- Fewer than 10 core conditions (8)
- Hatch marks indicate testing for some conditions required but not yet implemented.

Screening 29 Core Conditions

- District of Columbia
- Iowa
- Maryland
- Mississippi
- New Jersey
- Virginia

# U.A.E. Tracking of Congenital Abnormalities

- U.A.E. is an associate member of the Congenital Abnormality Study Group
- Started continuous monitoring of births in 1994
- The program covers ~8,000 births/year in three hospitals in the Al Ain Medical District of Abu Dhabi
- Still births >500 gm are included in the tracking
- The program is funded by the Medicine and Health Sciences Department of the UAE University
- A neonatologist in each hospital examines, identifies abnormalities and records information at birth; diagnoses are further assisted by a clinical geneticist and pediatricians
- Some basic information on exposure (e.g., maternal disease) is collected
- General epidemiological data for all births are available.



# Developmental Screening

- Approximately 16% of all children under five have a developmental delay ranging from very mild to severe (e.g., speech, language, cognition, social-communication)
- Approximately 25% of these children are identified prior to five years of age
- The earlier children are identified, the more likely the delay will be ameliorated



# Early Developmental Screening

- Developmental screening is “the application of a simple, accurate method for determining which children in the population are likely to be in need of special services in order to develop optimally.”

--Dumar, Duran-Flores, Foster, & Stills, 1987



# Criteria for Developmental Screening Procedures

- Simple (easy to complete, quick)
- Accurate (sensitive, specific, clearly defined criteria)
- Comprehensive (physical, cognitive, sensory, communication, social, and behavioral)
- Cost-effective
- Partnership with families



# Information Necessary to Conduct Accurate Developmental Screenings

- Child developmental information (via observation, checklist, screening tool)
- Child health information (via observation, records, parental report)
- Parent information about child (development, interaction patterns, temperament)
- Environmental information



# Recommended Guidelines for Developmental Screening

1. Screening is a service; it is a component of intervention; it is not merely a means for identification and measurement
2. Processes, procedures, and instruments intended for screening should only be used for their specified purposes
3. Multiple sources of information must be used to make screening decisions; single source decisions are highly inaccurate
4. Screening should occur on a recurrent basis; screen young children every six months for the first five years



# Recommended Guidelines for Developmental Screening (continued)

5. Screening instruments must be reliable and valid
6. Family members should be an integral part of the screening process
7. Screening procedures should be relevant and familiar to the child to increase validity
8. All tests, procedures, and processes intended for screening must be culturally valid and unbiased
9. Screeners must have extensive and comprehensive training in order to screen very young children

Source: *Zero to Three* ([www.zerotothree.org/](http://www.zerotothree.org/))



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# American Academy of Pediatrics Developmental Screening Recommendation:

- “Developmental surveillance should be performed at all well-child visits from infancy through school-age, and at any age thereafter if concerns are raised about social acceptance, learning, or behavior.”



## PEDS RESPONSE FORM

Child's Name Roger J. Parent's Name Malinda J  
Child's Birthday 8/8/03 Child's Age 2 Today's Date 8/10/05

Please list any concerns about your child's learning, development, and behavior.

*I'm worried about how my child talks and relates to us. He says things that don't have anything to do with what's going on. He's oblivious to anything but what he is doing. He's not doing as well as other kids in many ways.*

Do you have any concerns about how your child talks and makes speech sounds?

Circle one: No  Yes A little COMMENTS:

*He repeats odd things like "Wheel of Fortune"*

Do you have any concerns about how your child understands what you say?

Circle one: No  Yes A little COMMENTS:

*I can't tell if he doesn't understand, doesn't hear well or just ignores us*

Do you have any concerns about how your child uses his or her hands and fingers to do things?

Circle one:  No Yes A little COMMENTS:

*He's good with manipulatives but does a lot of the same things over and over: spinning wheels on cars, flicking light switches, flipping pages*

Do you have any concerns about how your child uses his or her arms and legs?

Circle one:  No Yes A little COMMENTS:

*He's very coordinated and very fast!*

Do you have any concerns about how your child behaves?

Circle one: No Yes  A little COMMENTS:

*still lots of tantrums but headbanging is almost gone. Behavior therapy has been helpful and his tantrums are less severe and shorter*

Do you have any concerns about how your child gets along with others?

Circle one: No Yes  A little COMMENTS:

*He doesn't seem interested in watching other kids, let alone playing with them*

Do you have any concerns about how your child is learning to do things for himself/herself?

Circle one:  No Yes A little COMMENTS:

*He's very independent*

Do you have any concerns about how your child is learning preschool or school skills?

Circle one:  No Yes A little COMMENTS:

*He's too young for any of that!*

Please list any other concerns.

*We spend lots of time playing with Roger and talking to him. This seems to be helping him be more engaged. I still wonder about his hearing.*

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# Screening Instrument Example

- Dr. Frances Glascoe's instrument—the Parent's Evaluation of Developmental Status (PEDS)
- Family-focused
- Developmentally anchored
- Language and culturally sensitive



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# Early Intervention Goals

- to promote child engagement, independence, and mastery
- to promote development in key domains
- to build and support children's social competence
- to promote generalized use of skills
- to provide and prepare for normalized life experiences
- to prevent the emergence of future problems or disabilities
- to support families in achieving their own goals related to their child with a disability



# Core Early Intervention Principles

- Services designed to promote development across the developmental domains
- Family-centered services
- Normalization and services in natural environments
- Service delivery models that fit family needs
- Interdisciplinary and transdisciplinary services
- Programming that addresses both functional and developmental needs of children
- Individualized programming



# Early Intervention Practices

- Inclusion
- A blending of special education and typical early childhood education approaches
- Naturalistic assessment
- Play-based intervention
- Individualized and small group instruction
- A focus on interactions and the processes of learning
- Activity-based intervention strategies
- Builds on children's preferences and interests
- Integrated therapy
- Collaborative/consultative roles for staff



# University of Delaware Early Intervention Model

- Child care center for 225 children birth to 12 years of age
- 20% children with disabilities
- 40% children living in poverty
- provides state of the art, evidence-based, education, prevention, and intervention services
- Currently conducts 18 research studies within the Center focusing on neurological development; PT, OT, and speech treatments strategies, and a host of other topics
- Currently provides training to over 2,000 undergraduate and graduate students through clinical and research experiences each semester
- Currently provides training and technical assistance early care and education providers throughout the state



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# Cost-benefit of Early Developmental Screening

- In the United States:
  - For every \$1 spent on screening and early intervention, \$7 in future services are saved
  - High quality early intervention programming results in higher graduation rates, better jobs for individuals with disabilities and lower rates of poverty
  - Families who are more cohesive and better able to care for their children



# Resources

- American College of Medical Genetics:  
<http://www.acmg.net/>
- March of Dimes: <http://www.marchofdimes.com/>
- International Clearinghouse for Birth Defects:  
<http://www.icbd.org/>
- International Birth Defects Information Systems:  
<http://www.ibis-birthdefects.org/>

# Resources (continued)

- Kingdom of Saudi Arabia Prince Salman Center for Disability Research:

[http://www.pscdr.org.sa/Arabic/Res\\_Programs.htm](http://www.pscdr.org.sa/Arabic/Res_Programs.htm)

- American Academy of Pediatrics:

<http://aappolicy.aappublications.org/cgi/content/full/pediatrics;108/1/192>

- National Early Childhood Technical Assistance Center (NEC\*TAC) [www.nectac.org/](http://www.nectac.org/)

# Resources (continued)

- Parents' Evaluation of Developmental Status:

<http://www.pedstest.com/content.php?content=brightfutures-qa.html>

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