Diagnostic Approach to Developmental Delay

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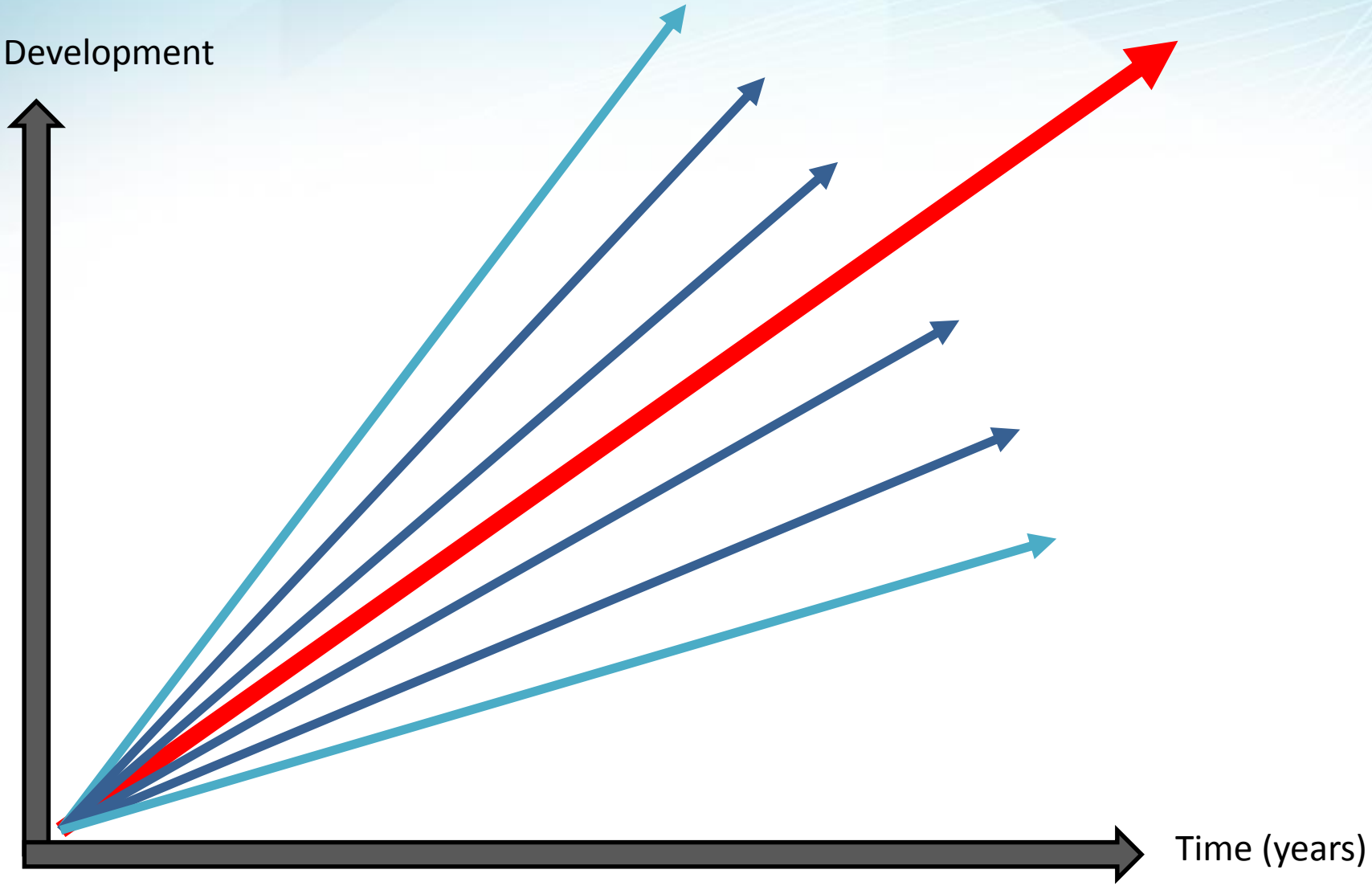
What is development?

Young Baby → Adulthood

- Wide variation between children
- Variation between domains in same child

Rule: Variable and Progressive



What is Developmental Delay?

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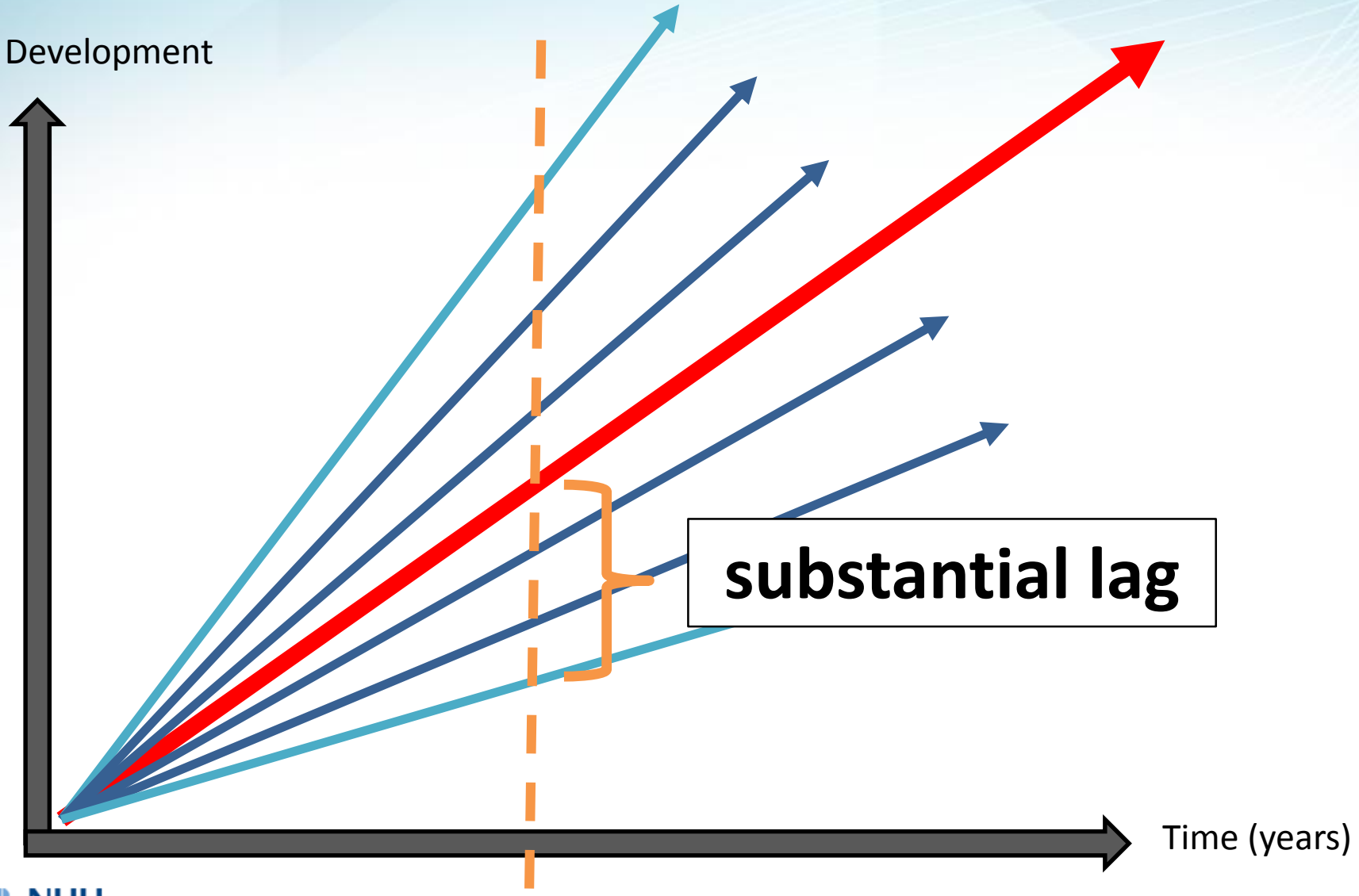

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Developmental Quotient (DQ)

$$\frac{\text{Developmental age}}{\text{Chronological age}} \times 100 = \text{DQ}$$

DQ < 70 indicates developmental delay

Red flags in evaluation of children with Neuromotor delay

TABLE 3 "Red Flags" in the Evaluation of a Child With Neuromotor Delay

Red Flags: Indications for Prompt Referral	Implications
Elevated CK to greater than 3× normal values (boys and girls)	Muscle destruction, such as in DMD, Becker muscular dystrophy, other disorders of muscles
Fasciculations (most often but not exclusively seen in the tongue)	Lower motor neuron disorders (spinal muscular atrophy; risk of rapid deterioration in acute illness)
Facial dysmorphism, organomegaly, signs of heart failure, and early joint contractures	Glycogen storage diseases (mucopolysaccharidosis, Pompe disease may improve with early enzyme therapy)
Abnormalities on brain MRI	Neurosurgical consultation if hydrocephalus or another surgical condition is suspected
Respiratory insufficiency with generalized weakness	Neuromuscular disorders with high risk of respiratory failure during acute illness (consider inpatient evaluation)
Loss of motor milestones	Suggestive of neurodegenerative process
Motor delays present during minor acute illness	Mitochondrial myopathies often present during metabolic stress

Referral Guidelines : Speech Delay

TABLE 8 Guidelines for Children with Abnormal Speech Development

Age, mo	Referral Guidelines for Children With "Speech" Delay
12	No differentiated babbling or vocal imitation
18	No use of single words
24	Single-word vocabulary of ≤10 words
30	Fewer than 100 words; no evidence of 2-word combinations; unintelligible
36	Fewer than 200 words; no use of telegraphic sentences; clarity <50%
48	Fewer than 600 words; no use of single sentences; clarity ≤80%

Source: Matkin ND. *Pediatr Rev.* 1984;6:151.

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Guidelines on Identification of Children with ASD

Prespeech Deficits Common in Children with Autism Spectrum Disorders
Decreased or absent use of prespeech gestures (e.g., waving, pointing)
Delayed onset of babbling past nine months of age
Disregard for vocalizations (i.e., lack of response to own name), yet awareness of environmental sounds
Lack of appropriate gaze
Lack of expressions such as “oh-oh” or “huh”
Lack of interest or response to neutral statements
Lack of recognition of mother's (or father's or consistent caregiver's) voice
Lack of the alternating pattern of vocalizations between infant and parent that usually occurs at approximately six months of age
Lack of warm, joyful expressions with gaze

Identification and Evaluation of Children With Autism Spectrum Disorders Paediatrics November 2007

Developmental Trajectory

Development

Time (years)

Developmental Trajectory

Developmental Delay

↓

Transient Developmental delay → **Typical Developmental**

↓

Persistent Developmental delay → **Developmental Disability**

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1. Environmental causes of DD are real

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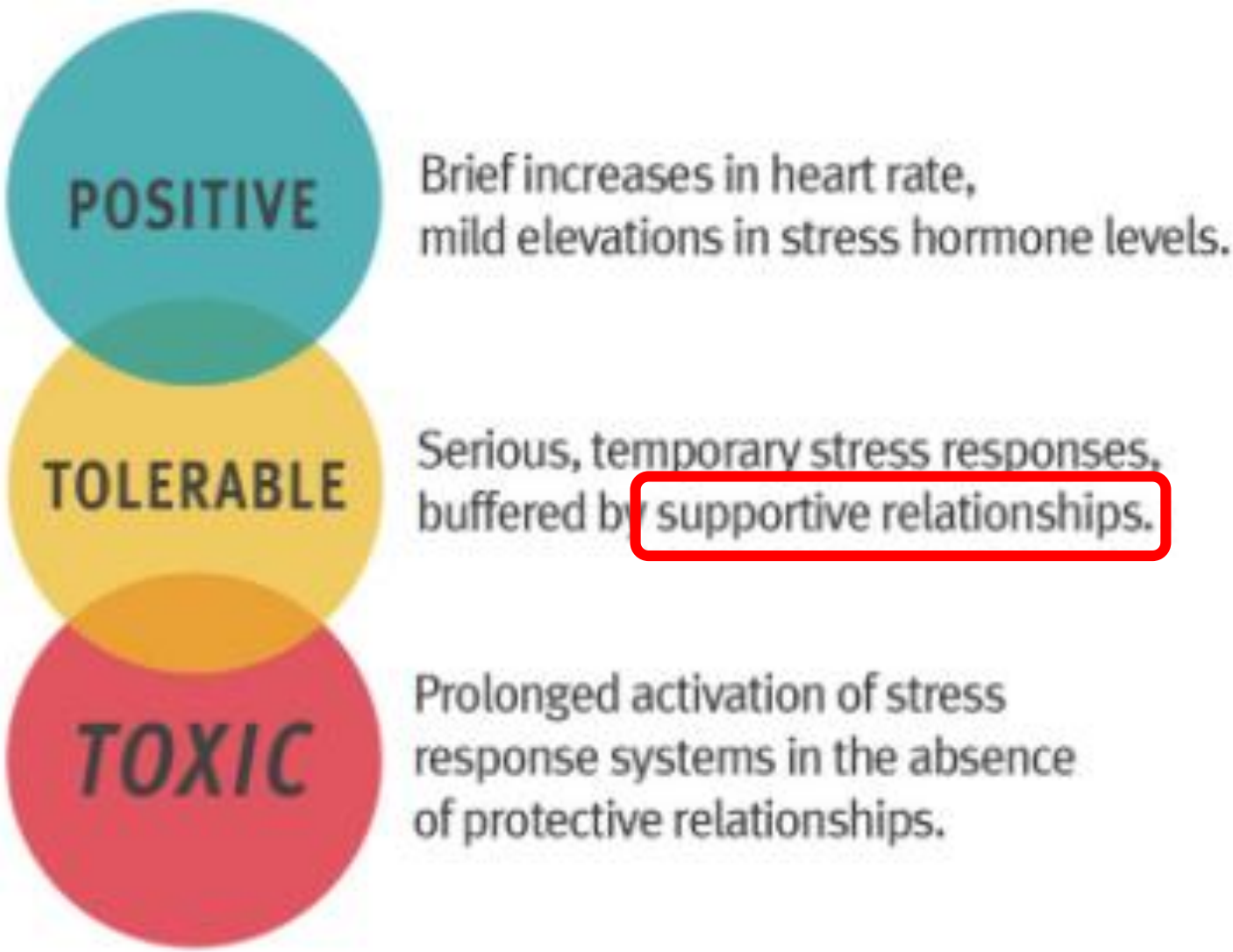
#1
Even infants and young children are affected adversely when significant stresses threaten their family and caregiving environments.

Stress



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Nurturing & Stable & Engaging relationships

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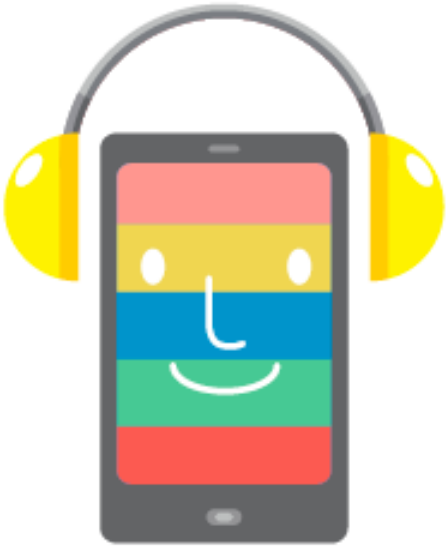
While attachments to their parents are primary, young children can also benefit significantly from relationships with other **responsive caregivers** both within and outside the family.

Unplug

SWAP SCREEN TIME FOR ACTIVE PLAY

MAKE SCREENTIME A REWARD—
not a routine



NO SCREENTIME BEFORE AGE 2
and no more than two hours of screentime for kids older than 2

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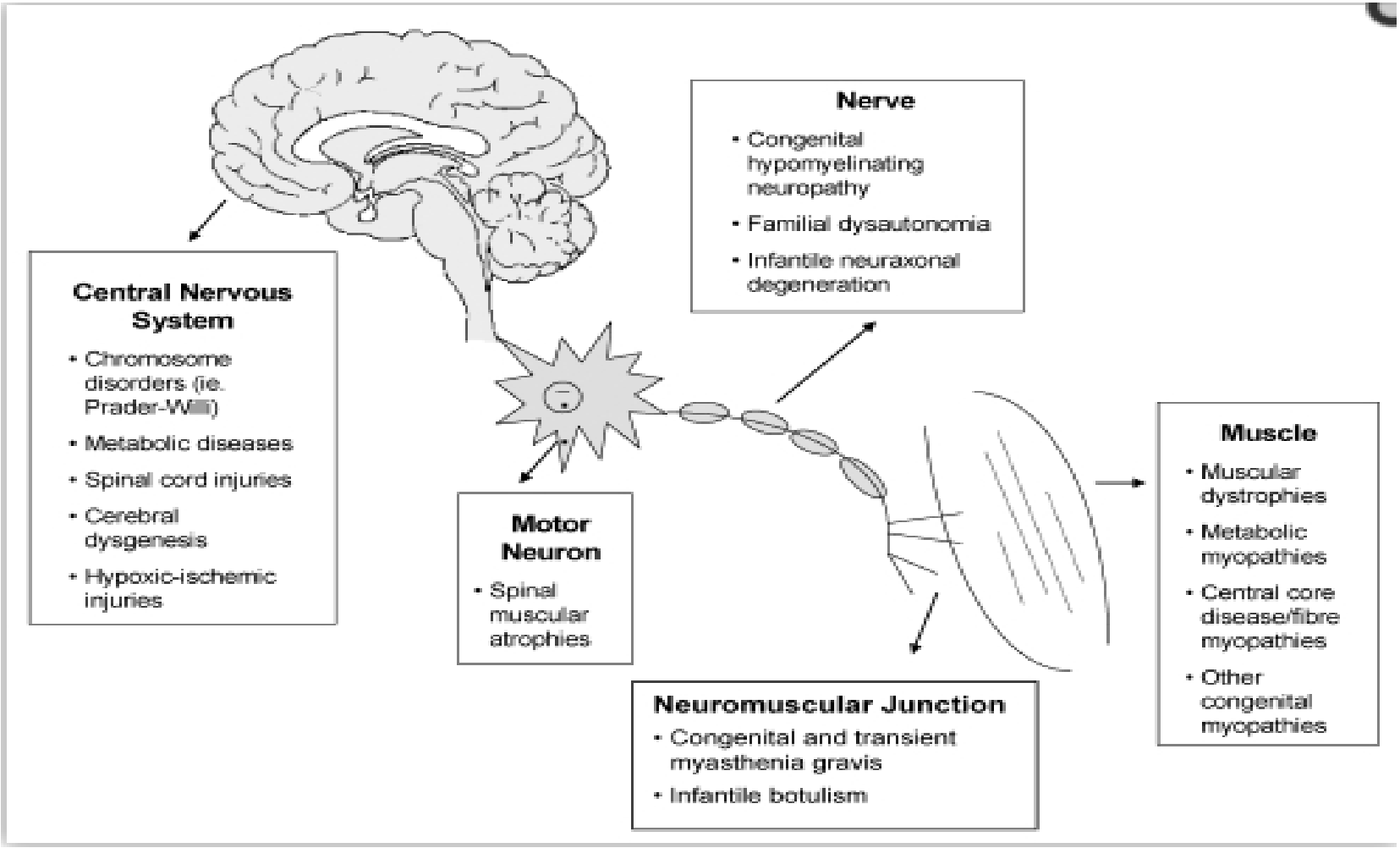
DDx of hypotonia in infancy



A schematic approach to hypotonia in infancy JoAnna Leyenaar, MD MPH, Peter Camfield, MD FRCPC, and Carol Camfield, MD FRCPC

Approach to Investigations

- *General Investigations*: TSH, free T4, electrolytes (& Ca)
- *CNS Dysfunction* : CT/MRI head, consider EEG, consult neurology and consider karyotype
- *Metabolic Disease* : urine and serum amino acids, urine organic acids, ammonia, liver function tests
- *Lower motor neuron disease* : creatine kinase, referral to neurology for specialised test

2. Motor Delay warrants closer attention

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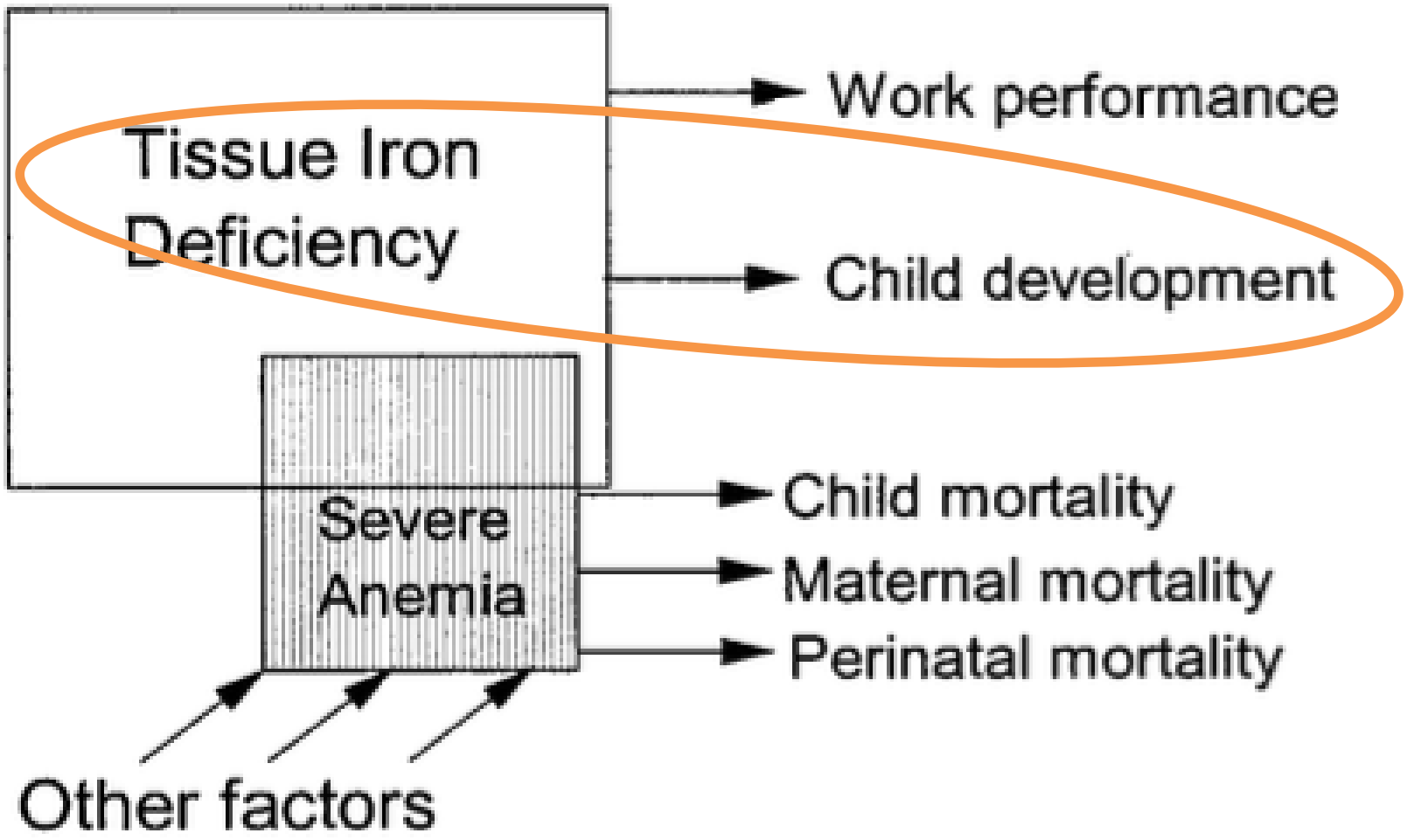
Fe def anemia affects development



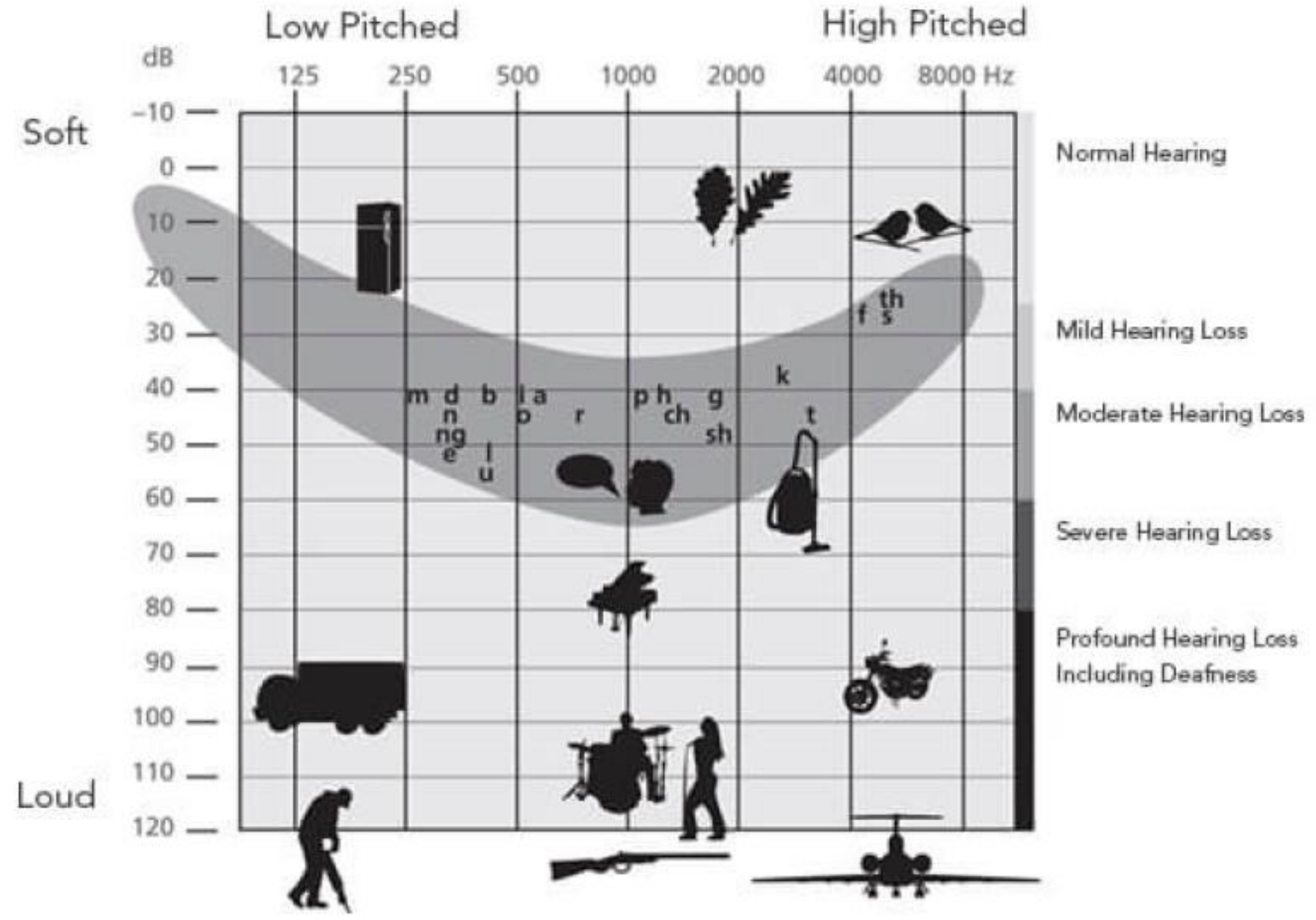
Summary: Implications for Research and Programs Rebecca J. Stoltzfus Center for Human Nutrition, Department of International Health, School of Hygiene and Public Health, The Johns Hopkins University, Baltim

Iron deficiency and impaired child development The relation may be causal, but it may not be a priority for intervention. Haroon Saloojee, senior lecturer and John M Pettifor, professor

Low vitamin D status & development

In utero → post natal → early childhood

Vitamin D and Autism Spectrum Disorder: A Literature Review Hajar Mazahery, Carlos A. Camargo, Jr. and Pamela R. von Hurst
The effects of vitamin D on brain development and adult brain function Molecular and Cellular Endocrinology December 2011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Normal Hearing
Mild Hearing Loss
Moderate Hearing Loss
Severe Hearing Loss
Profound Hearing Loss
Including Deafness

Alstrom Syndrome

Feature	Age of Onset Range (Mean)	Incidence
Cone-rod dystrophy	**Birth - 15 mos (5 mos)**	**100%**
Obesity	**Birth - 5 years (2.5 yrs)**	**98%**
Progressive sensorineural hearing loss	**2-25 yrs (9 yrs)**	**88%**
Dilated cardiomyopathy	2 wks - 4 mos	42%
Restrictive cardiomyopathy	**Juvenile - late 30s**	**18%**
Insulin resistance / type 2 diabetes mellitus	4-30 yrs / 8-40 yrs (16 yrs)	92% / 68%
Developmental delay	**Birth-adolescence**	**25%-30%**
Short stature	Puberty - adult	98%
Hypogonadotropic hypogonadism	10+ yrs	78% of males
Urologic disease	Adolescence - adult	48%
Renal disease	Adolescence - adult	Variably progressive with age in all individuals
Hepatic disease	**8-30 yrs**	**23%-92%**

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Based on a study of 182 patients by Marshall et al [2005]

3. Hearing screen is important

(despite newborn hearing screen)

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Hearing lost is difficult to diagnose clinically.

Use risk stratification instead.

AAP Clinical Report—Hearing Assessment in Infants and Children: Recommendations Beyond Neonatal Screening *Pediatrics* 2009

Risk Indicators Associated With Permanent Congenital, Delayed-Onset, and/or Progressive Hearing Loss in Childhood

- Caregiver concerns regarding hearing,
- speech, language delay
- developmental delay
- Warrants objective hearing assessment

AAP Clinical Report—Hearing Assessment in Infants and Children: Recommendations Beyond Neonatal Screening *Pediatrics* 2009

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Thank You.

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Questions?