Developmental Red Flags

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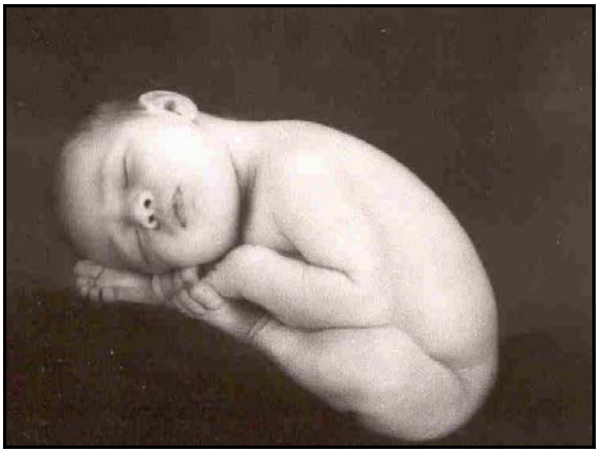
OBJECTIVES

"The most common presentation of a developmental disability is failure to achieve age-appropriate developmental milestones."

- *How common are developmental problems?*
- *How does one approach developmental delay?*
- *What are some "rules of thumb" for recognizing delay?*
- *What are the developmental implications of common newborn findings?*



DEFINITIONS

- Delay vs. Deviance
- Surveillance vs. Screening
- Streams vs. Domains

LOOK FOR PATTERNS!

Streams of Development

- Gross motor
- Fine motor
- Language
  - Expressive
  - Receptive
- Problem solving
- Social

Patterns in Development

	MOTOR	PS	RL/EL	SOCIAL
ID	V	D	D	D
CP	D	V-N	V-N	D
VI	D	D	N	D
HI	N	N	D	V-N
AUTISM	N	V-N	D	D

Prevalence of DD

Dx	per 1000	age (mo)	MD 1st
GDD	50	24	30%
ID (mild MR)	25	39	60%
ID (mod/severe MR)	5	12	90%
Learning disability	100	69	12%
ADHD	90	59	45%
CP	3	12-14	99%
AUTISM	8	40	30%
Visual impairment	.5	55	60%
Hearing Impairment	1-3	39	40%

What's the problem?

- 16% of preschool children have developmental or behavioral problems.
- The Federal Law mandates early identification.
- <30% of MDs screen.

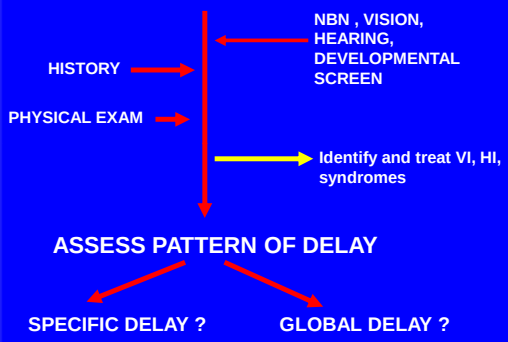
Use the parent!

Parent concern	PPV
Behavior-emotions	41%
Speech/language	55%
Social/self-help	NS
Fine motor	75%
School readiness	40%
Global concerns	80%

Ages for early diagnosis

0-12 mo.	1-2 yr.	2-3 yr.	3-4 yr.
ID, mod/sev.	ID, mod	ID, mild	ID, mild
VI/HI	HI	HI	
CP	CP	CP mild	
	Autism	Autism/LD	LD

CHILD WITH DEVELOPMENTAL DELAY



RED FLAG

Any time there is a history or exam consistent with

LOSS of SKILLS (regression)

THINK……metabolic, genetics w/u, neuroimaging, seizures, hydrocephalus, toxin exposure, autism

Developmental Quotient

DQ = Developmental Age/ Chronologic age X 100

DQ <70	Delay
DQ 70-85	Monitor
DQ >85	Typical range

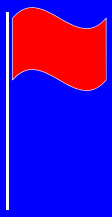
*Perform for each stream of development

Gross motor milestones

- Prone
  - Head up 1 mo
  - Chest up 2 mo
  - On elbows 3 mo
  - On hands 4 mo
- Roll
  - Front to back 4-5 mo
  - Back to front 5-6 mo
- Sit
  - With support 5 mo
  - Without support 6 mo
- Crawl (1/3 don't!) 8-9 mo
- Pull to stand 9 mo
- Cruise 9-10 mo
- Walk alone 12 mo
- Run 15 mo
- Jump in place 24 mo
- Pedal trike 30-36 mo
- Gallops 48 mo(4)
- Skips 60 mo(5)
- Tandem
  - Forward 5 yr
  - Backward 7-8 yr

RED FLAG

RED FLAGS

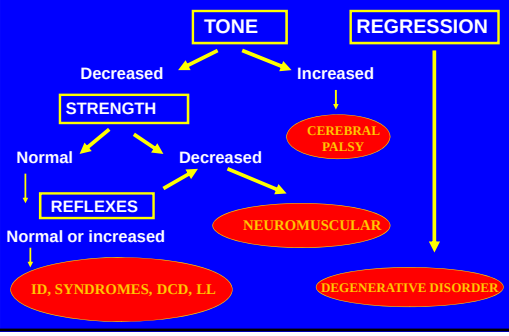


- Any boy not walking by 15 months should get a CPK to r/o MD
- The most common cause of delayed GM is global delay.

MD = muscular dystrophy

- 3/10,000 boys
- 30% have no Fam Hx
- Absent dystrophin
- Xp21.2
- CPK is >20x normal!
- DNA analysis of blood can make dx in 2/3's.

Motor delay



Motor delay: Evaluation

- Central (CNS)
  - MRI
  - CHROMOSOMES
  - FRAGILE X
  - ? METABOLIC EVAL
  - ? TFT
  - Get PT or OT eval
- Neuromuscular
  - CPK
  - LFT'S
  - ESR
  - LACTATE/PYRUVATE
  - TFT
  - CARNITINE
  - EMG/NCV
  - MUSCLE BX

Definitions

- Language- a system of verbal, written, or gestured symbols used to communicate information or feelings.
  - Components: phonology, morphology, syntax, semantics, pragmatics
- Speech- The physical production of spoken language.
  - Components: articulation, phonology, voice quality, pitch, loudness, resonance, fluency, rate, rhythm

Identify early!

- 5-10% of all children have developmental language disorder (that is 2 / day for PCM)
- Language is the BEST predictor of later cognitive function
- Delayed language may be a marker of other developmental disorders
- Early intervention yields best outcomes

Normal Language Development

- Expressive: two phases

Social smile 5 wk Coos 6-8 wk Laughs 3-4 mo Rasberry 4-5 mo Squeals 5 mo Babbles 6 mo UNTIL 6-8 mo!	1st word 11 mo Immature jargon 12 mo 4-6 words 15 mo 2 words 21 mo Pronouns indiscrim 2 y Tells stories 4 years, 100% intelligible

Normal Language Development

- Receptive
  - Infant

Alerts to voice 1 mo
Regards speaker 3 mo
Listen then vocalizes 5 mo
Enjoys gesture games 9 mo
Orients to name 10 mo
Understands "no" 11 mo
Command, with gesture 12mo

- Receptive
  - Toddler

1 step command without gesture	14 mo
1 body part	15 mo
Fetches on command	16 mo
Points to picture	18 mo
6 body parts	20 mo
2 step command	24 mo

Language delay

Developmental Language Disorder	5-10%
ID	3%
Hearing Impairment	.5-1%
Autism	.6-1%

RED FLAG

- All children with language delay should be referred for hearing assessment.

Infants who are deaf may have normal pre-linguistic expressive language until 6-9 months of age.

RED FLAG

- Normal Pattern is for RL>EL.
- Expressive language that significantly exceeds receptive language is deviant.

THINK :

1. AUTISM, with echolalia
2. Syndromes with “cocktail personalities”
3. Parent mis-interpretation

Joint Attention Skills*

	TYPICAL	ASD
8-10 months	Gaze monitoring	No eye contact
10-12 months	Following a point	Does not respond to request "oh look!"
12-14 months	PIP	Develops advanced self help skills
14-16 months	PDP	* Consistently absent
14-18 months	Show and tell	Often brings to parent to obtain repeated action

*Joint attention deficits appear to be specific to ASD and reliably differentiate children with ASD from other developmental disabilities.

RED FLAG

A BIFID UVULA is evidence of a submucous cleft in the palate and warrants evaluation if associated with recurrent OM, speech delay, or VPI.

CONSIDER VCFS (del 22q.11)

Problem solving

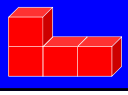
- Problem solving milestones are evidence of cognitive abilities, or intelligence, without the use of language.

…Patterns…..

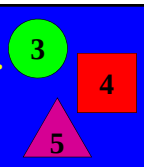
- Normal PS=RL>EL..COMMON, often resolves
- Normal PS>RL>EL…less common, often LD
- Low PS, Low RL, Low EL = ID

Normal development

- Blocks
  - Regards — 3 mo
  - Attains — 6 mo
  - Takes 2nd — 7-8 mo
  - Releases into cup — 12 mo
  - Takes a 3rd — 14 mo
  - Builds a tower of 2 — 15 months
  - Builds a tower of 4 — 18 months
  - Builds a tower of 6 — 24 months
  - Train — 30 months

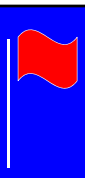


Normal development



- Pencil
  - Imitates scribble — 15 mo
  - Scribbles spontaneously — 18 mo
  - Imitates vertical line — 24 mo
  - Imitates horizontal line — 28 mo
  - Circle — 3 yr
  - Square — 4 yr
  - Triangle — 5 yr
  - Goodenough-Harris DAP — see HLH

RED FLAG



- Early Handedness

Children with handedness before age 15 months usually have an abnormally weak upper extremity on the other side.

"Global developmental delay"

- A significant delay in 2 or more streams
- NOT a diagnosis
- NOT regression or loss of skills
- Can't be used for services after age 6-9.

Intellectual Disabilities (Mental retardation)

- 2-3% of population
- Male 1.6 : Female 1
- 85% of ID is MILD category

Most common genetic cause: Down Syndrome
Most common inherited cause: Fragile X
Most common preventable cause: Fetal alcohol

MR =
ID =
Cognitive /Adaptive Disability

- Significantly sub-average general intellectual functioning (IQ <70-75)
- Limited adaptive functioning in at least 2 of 10 skill areas
- Onset before 18 years of age

Levels of ID (CAD)

- Mild (Intermittent Support) IQ~ 55-69
  - Vast majority 85%
  - More common in boys
- Moderate (Limited Support) IQ ~ 40-54
- Severe (Extensive Support) IQ~ 25-39
  - Rare .5%
  - Ratio of boys to girls is equal
  - Think about Rett Syndrome in girls
- Profound (Pervasive Support) IQ < 24

Intellectual Disability: Known causes

- Prenatal (60-75%)
  - CNS malformation
  - Chromosomal abnormality
  - Syndrome
  - Genetic
  - Toxins
  - Infection
  - Neurocutaneous syndrome
  - Malnutrition
- Perinatal (10%)
  - Hypoxia
  - Neonatal seizures
- Postnatal (1-10%)
  - CNS infection
  - Stroke/Hemorrhage
  - Trauma/Abuse
  - Hypoxia
  - Degenerative
  - Epileptic encephalopathy
  - Metabolic
  - Complications of prematurity

ID: The Search

The more severe the ID,
the more likely to find etiology.

- Chromosomes or CGH (40% + in SEVERE)
- DNA for Fragile X (2-6% +)
- ± Neuro-imaging (MRI study of choice)
  - IQ <50, micro/macrocephaly, abnormal neuro exam, seizures, loss of milestones
- ± Metabolic Studies

ID: Adult outcomes

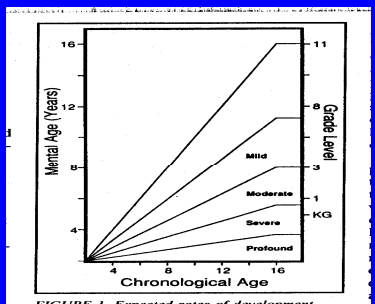


FIGURE 1. Expected rates of development with degrees of mental retardation. From Accardo PJ, Capute AJ. Mental retardation. In: Capute AJ, Accardo PJ, eds. Developmental Disabilities in Infancy and Childhood. *Baltimore, Md: Paul H. Brookes; 1991: 431 (used with permission).*

Predictive value of a good exam

The presence of three or more minor anomalies is highly predictive of a major malformation (19.6%)

- **Examples: bossing, absent hair whorl, anteverted nostrils, epicanthal folds, preauricular tags, pits, abnormal pinna of ears, bifid uvula, extra nipples, single umbilical artery, umbilical hernia, dimple over sacrum, single palmar creases, syndactyly, overlapping toes, recessed toes…..**

Head Circumference

- **Normal growth in full term infants:**
  - **2 cm/mo for 3 months**
  - **1 cm/mo for 3 months**
  - **1/2 cm/mo for 6 months**
- **12 cm in first 12 months!**

Head Circumference

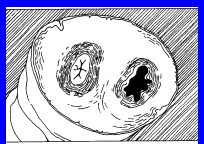
- Rule of 3's and 9's
  - Birth: 35 cm
  - 3 mo: 40 cm
  - 9 mo: 45 cm
  - 3 yrs: 50 cm
  - 9 yrs: 55 cm

Pits and tags

- 1/100 –common!
- ALL need Hearing
- Renal US if:
  - Other malformation
  - FHx of deafness, ear or kidney issue
  - Gestational DM

Single umbilical artery

- 6/1000
- More common in twins
- If exam normal, and mother did not have oligo/polyhydramnios, no further work up.
- Exam should r/o GI, renal, cardiac and CNS findings



A single umbilical artery with velamentous insertion of the cord. Most infants with SUA who survive do not need an extensive workup.

AJ Schuman, *Contemporary Peds*, Dec, 1991

Sacral lesions

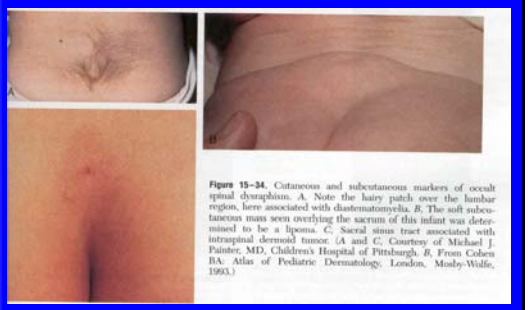


Figure 15–34. Cutaneous and subcutaneous markers of occult spinal dysraphism. *A*, Note the hairy patch over the lumbar region, here associated with diastematomyelia. *B*, The soft subcutaneous mass seen overlying the sacrum of this infant was determined to be a lipoma. *C*, Sacral sinus tract associated with intraspinal dermoid tumor. (*A* and *C*, Courtesy of Michael J. Painter, MD, Children's Hospital of Pittsburgh. *B*, From Cohen BA: Atlas of Pediatric Dermatology. London, Mosby-Wolfe, 1993.)

Sacral lesions

- **Dimple – need sacral neuro-imaging (US) if:**
  - – > 5 mm in size
  - – > 2 cm away from anus
  - – Associated with any other back lesion
    - Hemangioma, hairy patch, tail, masses
- **All hemangiomas, hairy patches, masses, tails need US (36% +)**

"Find the ability in disability"