Developmental Delay

Last updated: April 21, 2019

PHYSICAL GROWTH - increase in size.
DEVELOPMENT - growth in function and capability.

Development is divided into specific domains (substantial overlap exists):

1) gross motor
2) fine motor
3) language (ability to understand language precedes ability to speak)
4) cognition
5) social/emotional growth

- progress within different domains varies.

Developmental delay - failure of child to achieve expected motor and cognitive milestones owing to *ENCEPHALOPATHY*.

Associated disorders:

1) **mental retardation**
2) **nonprogressive encephalopathy** - previous brain injury that is no longer active → *STATIC DISORDERS OF BRAIN DEVELOPMENT* (e.g. cerebral palsy)
3) **progressive encephalopathy** (expanding mass lesion, neurometabolic, neurodegenerative or chronic inflammatory diseases)
4) **spinal dysraphism**
5) **autism**.

Most children with developmental delay are mentally retarded, and most mentally retarded children have associated handicap such as cerebral palsy or epilepsy.

- range within which normal children reach different milestones is wide - it is difficult to diagnose developmental delay within first year of life (unless it is severe).
- *MOTOR DEVELOPMENT* cannot be significantly accelerated by applying increased stimulation.
- appropriate attachments and nurturing in infancy and early childhood are critical factors in *COGNITIVE* and *EMOTIONAL GROWTH*.
- it may be very difficult to discriminate between *motor delay* and *mental retardation*.

SPEECH DELAY

- delays in expressive speech are typically not accompanied by other developmental delays (vs. delays in both receptive and expressive speech - often additional developmental problems).
- evaluation should start with **hearing assessment**.
  Hearing deficits impair language development (hearing problems must be remedied as early as possible!!!)
- most children who experience speech delay have *normal intelligence* (vs. children with accelerated speech development are often of *above average intelligence*).

CEREBRAL PALSY (CP)

- nonprogressive motor disorder (abnormal control of movements or posture) due to intrauterine ÷ early postnatal nonprogressive (static) injury to DEVELOPING brain (cerebrum or cerebellum), i.e. due to nonprogressive [static] encephalopathy.

N.B. term "cerebral palsy" does not apply to disorders of spinal cord, peripheral nerves, or muscles!

N.B. nonprogressive is misnomer – child's nervous system has plasticity – some functions improve over time, others, if not treated, deteriorate

Timing of brain injury (most important factor determining resulting pathology):

a) **early fetal life** → arrested / altered development of immature brain → congenital malformation.
b) **second half of pregnancy** → destructive injury to already formed brain.
c) **close to term or postnatal** → morphology closer to adult pathology.

PREVALENCE – 2.5 children out of 1000 live births (40% are born prematurely).

ETIOLOGY

- cause could not be identified in most cases!
- **intrauterine** (90%) / **neonatal** (10%) factors that injure DEVELOPING brain:
  1. Ischemic / anoxic accidents
  2. Malformations
  3. Infections
  4. Kernicterus

CLINICAL FEATURES

- manifest before age 5 yr.; symptoms may be inapparent at birth.
- before specific syndrome develops, symptoms include lagging motor development and often persistent infantile reflexes, hyperreflexia, altered muscle tone.
  N.B. *patients do not lose skills once acquired*! (vs. progressive neurologic disorders!)
- clinical variants (depend on lesion location):
  1. **Mixed CP** – combination of dyskinetic CP and spastic or ataxic CP – most frequent form!!!! (perinatal insults only rarely are specific enough to affect ony one motor component)
  2. **Spastic diparesis (diplegia), LITTLE disease** – most common form (≈ 45%).
  3. **Spastic hemiparesis (hemiplegia)** – commonest form (≈ 34%) in term neonates.
  4. **Spastic quadriparesis (quadriplegia)** (≈ 7%)
  5. **Hypotonic** CP
  6. **Dyskinetic** CP (athetosis, choreoathetosis) (≈ 7-20%)

7. **Ataxic** CP – rarest form (< 5%)

- commonly (25%; esp. in spastic variants) *associated with spectrum of developmental disabilities* (mental retardation, epilepsy, visual, hearing, speech, cognitive, and behavioral abnormalities) - motor handicap may be least of child's problems.
- spastic CP → joint contractures → joints may become misaligned.

DIAGNOSIS

- **MRI** is indicated!
- diagnosis of underlying cause rarely influences therapy (advanced neuroimaging in CP has not been widely used until recently), but exact diagnosis is very important for parents!
- seek for rare treatable causes (e.g. hydrocephalus)
- vision & hearing must be tested early.

TREATMENT

- *parents should be taught how to handle* child in daily activities (feeding, carrying, dressing, bathing, playing).
- severe limitations in sucking and swallowing → feeding by gastrostomy tube.
- **physical therapy** is essential to train ambulation, stretch spastic muscles, and prevent deformities.
- **occupational therapy** - self-help skills and interpersonal communication.
- **education** tailored to intellectual abilities.
- **drugs / procedures** for spasticity. see p. Mov3 >>
  N.B. best age for spasticity surgery is 4-7 years – enough time for spontaneous improvement to occur; orthopedic problems should be fixed after spasticity is addressed!!!
- most survive to adulthood.

PREVENTION

- MAGNESIUM SULFATE IV before birth for women delivering *extremely premature babies*.

SPASTIC HEMIPARESIS (HEMIPLEGIA)

- lesion of corticospinal system of one cerebral hemisphere.

- common causes:
  1) intrauterine stroke (e.g. in twins, due to ischemia related to shared placental vessels); stroke can also occur during birth process and in infancy (*ACUTE INFANTILE HEMIPLEGIA*).
  2) intraventricular hemorrhage (in small premature infants) complicated by intraparenchymal hemorrhage.
- hemiparesis affects arm & hand more than leg.
- *all children walk*, albeit often later and on toes of affected foot (because of tight heel cord that may necessitate surgical lengthening).
- **growth "arrest" of arm and leg** is frequent (esp. with parietal lobe lesions) - arm and leg are shorter and thinner, compensatory scoliosis.
- hemiparesis may not be evident until child starts to grab for objects and shows precocious handedness or failure of hand use; this does not imply that lesion was acquired postnatally.
- spasticity tends to increase in first and second years and is more evident when child is erect.
- child *learns to speak & read competently* (speech acquisition may be delayed).
- *intelligence may be spared*, but subtle neuropsychologic differences between right and left lesions may be demonstrable; 25% have cognitive abnormalities (incl. mental retardation).
- 1/3 patients have **seizures** (when lesion affects cortex).
- neuroimaging - atrophic cerebral hemisphere with dilated lateral ventricle.
- treatment:
  Large unilateral lesion, intractable seizures, and severe behavior disorders → hemispherectomy or other excisional surgery.

SPASTIC DIPARESIS (DIPLEGIA), s. LITTLE disease

- most common causes:
  1) prematurity with bilateral germinal matrix hemorrhage ± intraventricular hemorrhage and hydrocephalus.
     Most patients are prematures!
  2) perinatal ischemia in watershed parasagittal zone between territories of ACA and PCA.
- first noted when infant begins to crawl - child uses arms in normal reciprocal fashion but drags legs behind more as rudder (commando crawl).
- ***adductor spasm*** is responsible for leg "scissoring";
  – application of diaper is difficult.
  – child walks on tiptoes; marked spasticity may preclude ambulation without walker and long-leg braces.
  – when child is suspended by axillae, scissoring posture of lower extremities is maintained.
- disuse atrophy and ***impaired growth of lower extremities*** (disproportionate growth with normal development of upper torso).
- variable clumsiness of hands.
- *intelligence* and *speech* unimpaired.
- likelihood of seizures is minimal.
- neuroimaging - periventricular leukomalacia.

SPASTIC QUADRIPLEGIA

- most severe variant of CP

- often associated with moderate-to-severe mental deficiency.
- rarely able to walk, and most are totally dependent.
- pseudobulbar manifestations (→ aspiration pneumonia).
- seizures are frequent.
- poor hand use precludes learning of all but most rudimentary signs (difficult to assess cognition).
- neuroimaging - extensive brain damage of both grey and white matter.

Spastic quadriplegia – "scissoring" of legs, pronated forearms, "fisted" hands:

HYPOTONIC CP

- floppy but with hyperactive tendon reflexes (vs. LMN or primary muscle diseases).

- pathophysiology is not understood.
- usually severe mental deficiency.

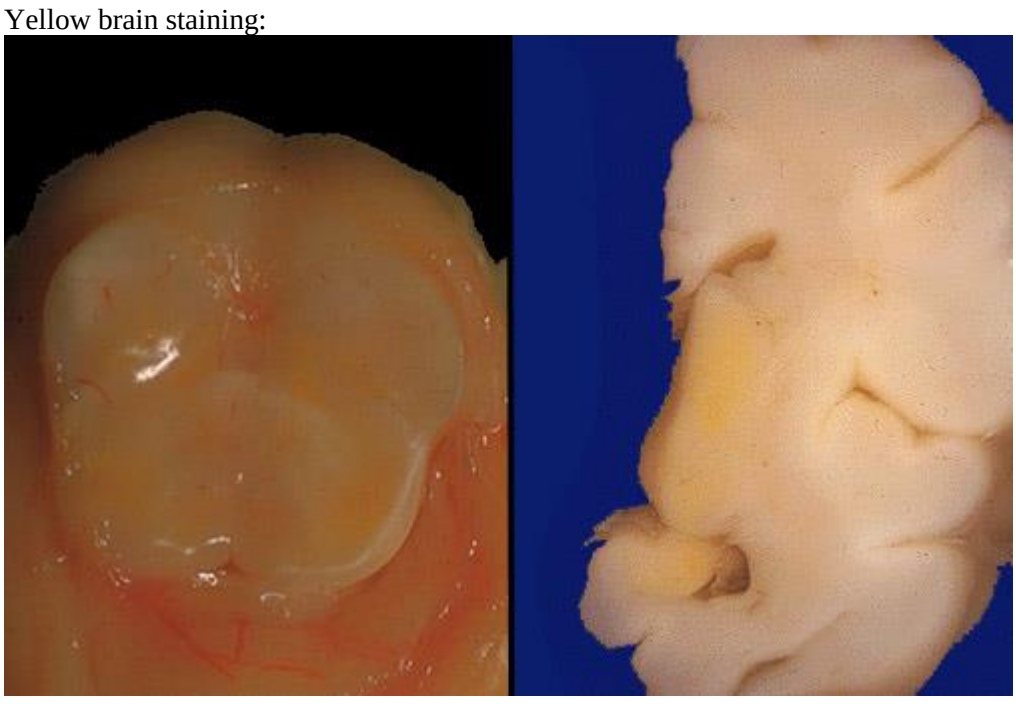
DYSKINETIC CP

- basal ganglia lesions lead to abnormal involuntary movements (athetosis, choreoathetosis, dystonia).

Chief causes:

1. **Kernicterus** (also see p. 1959 >>) - unconjugated bilirubin selectively damages basal ganglia, central auditory and vestibular pathways, and deep cerebellar nuclei (*cortex is not affected*!); may be unable to speak (because of facial dyskinesia) and hearing loss* and have little or no hand use, but may be normally intelligent; UMN signs not present, seizures uncommon.
   *hearing loss is typically in high tones; children are not deaf but cannot discriminate consonants that convey most of meaning of speech!

Yellow brain staining:



2. **Severe anoxia** - both cortical and subcortical damage (status marmoratus of basal ganglia) → intellectual as well as motor handicaps.

- movements emerge after age 1 year (in early infancy, children are hypotonic, with poor head and trunk control and little or no use of hands) - first sign may be ***tongue thrusting*** (makes spoon feeding difficult).
- some children walk but assume unusual postures and have stigmatizing facial grimaces, dysarthria, and dysphagia.

Treatment - high doses of TRIHEXYPHENIDYL, LEVODOPA, CARBAMAZEPINE may have modest effect; stereotaxic surgery on basal ganglia and thalamus runs risk of irreversible anarthria (if lesions impinge on internal capsule).

ATAXIC CP

- rarest form, due to maldevelopment of cerebellum or its pathways.

- truncal and gait ataxia are more striking than limb ataxia, but some children take long time to learn to feed themselves and have severe difficulty writing.
- eventually learn to walk but remain clumsy and fall frequently.
- nystagmus is uncommon.
- speech may be slow and scanning.
- neuroimaging - most have ***no abnormal findings*** (only 25% have detectable posterior fossa pathology; small number - supratentorial malformations).
- may improve with age.

BIBLIOGRAPHY for ch. "Pediatrics" → follow this LINK >>