Central Nervous System Congenital Abnormalities

Eva Brichtova, M.D., Ph.D.,
Department of Pediatric Surgery, Orthopaedics and Traumatology, University Hospital Brno
Neural tube defects

- Dysraphism
  - uncomplete neural tube closure with possible herniation

1. Cranial dysraphism
2. Spinal dysraphism

- Total dysraphism - craniorachischisis
  - non developed calvarium with complete spinal canal splitting (mostly abortus)
1. Cranial dysraphism

- uncomplete neural tube closure with „cranial bifidum“ (middleline calvaria defect) with possible cephalocele

- Cephalocele
  a) cranial meningocele: dura mater and CSF herniation
  b) encephalocele: cerebral tissue herniation
  c) anencephaly: open dysraphism, without calvaria bones

- Localisation
  - middle line, frontal, parietal or occipital

- Diagnosis
  - X-rays skull and spine - skeleton defects
  - Ultrasound - hernia content
  - CT or MRI - detailed information
Cranial dysraphism

- Cranial meningocele mostly has a good prognosis
- Encephalocele is mostly accompanied by hydrocephalus, mikrocephaly, mental retardation, epileptic seizures
Cranial dysraphism

a) Microcephalia
b) Hydranencephalia - a loss of almost all cerebral tissue
c) Holoprosencephalia - hemispheral development disturbance
d) Lissencephalia - severe disturbance of neural tissue migration

- **agryia:** completely smooth cerebral surface
- **pachygyria:** few flat gyruses
- **polymicrogyria:** small gyruses, shallow sulci (similar to pachygyria)

e) Porencephalia
f) Agenesis of corpus callosum
g) Dandy-Walker syndrom (cerebellar hypoplasia)
h) Macroencephaly - megalencephaly
i) Schizencefaly
Cranial dysraphism

- Schizencefalia
2. Spinal dysraphism - spina bifida

a) Spina bifida occulta
   - congenital absence of processus spinosi and vertebral arches changes

- Cutaneous changes in middle lumbosacral region:
  - hypertrichosis
  - lipoma
  - dyscoloration
  - dermal sinus (cave infectious complications)

- Serious conditions associated
  - Syringomyelia
  - Diastematomyelia
  - Tethred cord syndrom

- X-rays diagnosis
  - L5-S1 level - dorsal part of spinal canal - closure defect
Spinal dysraphism

- Diastematomyelia in CT 3D
Spinal dysraphism

a) Spina bifida aperta seu spina bifida cystica
   - eningocele - vertebral arches defect, meningeal cyst, in 1/3 neurological deficit
   - myelomeningocele - vertebral arches defect, meningeal cyst, structural and functional nervous tissue abnormalities

- **Myelomeningocele epidemiology:** 1 from 1000 newborns

- **Clinical features**
  - lower extremities paresis proprioceptive reflexes disturbance incontinence

- **Associated conditions**
  - hydrocepalus (in 65-85 %)
  - Chiari malformation (in 80 %)
Spinal dysraphism

Myelomeningokéla

Meningokéla
Spinal dysraphism
Spinal dysraphism
Spinal dysraphism
Spinal dysraphism
Spinal dysraphism
Spinal dysraphism
Craniostenosis (caniosynostosis)

- Premature skull sutures synostosis
- 1852  Rudolf Virchow
- 1 from 2100 children
1. Skaphocephalia - dolichocephalia

- Premature sagittal suture synostosis
- Výskyt 40-60 %
Skaphocephalia - dolichocephalia
Skaphocephalia - dolichocephalia
Skaphocephalia - dolichocephalia
2. Brachycephalia

- Premature coronar suture synostosis
- 20–30 %

a) Frontal plagiocephalia
   - one side coronar suture

b) Occipital plagiocephalia
   - one side lambdoid suture
Brachycephalia
Brachycephalia
3. Trigonocephalia

- Premature sutura metopica synostosis
- 10%
Trigonocephalia
Trigonocephalia
Trigonocephalia
Trigonocephalia
Trigonocephalia
Trigonocephalia
4. Morbus Crouzon - dysostosis craniofacialis (1912)

- Turicephaly
- Shallow orbits
- Exophthalmos
- Hypertelorism
- Hypoplasia of middle facial skelet

- 1 from 25 000 children
Morbus Crouzon
Morbus Crouzon
Morbus Crouzon
Morbus Crouzon
Morbus Crouzon
Morbus Crouzon
Post surgery care

Preventive protective helmets