Congenital anomalies in newborn

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Malformation

Internal or external morphological abnormality arising during intrauterine life and present at birth, regardless of its etiology, pathogenesis, and the moment of recognition.
Causes of malformation

1. Genetics
   - monogenic
     - multifactorial
     - somatic cell mutation

2. Environment (teratogens)
Types due to pathogenesis

- **Malformation** - primary developmental disorder organ or tissue in the early stages of development (e.g., Heart defects)
- **Disruptions** - interruption or disturbance initially normal developmental process (e.g., Amniotic cord syndrome)
- **Deformations** - irregularities in shape or position of parts of the body (club feet varus)
- **Dysplasia** - abnormal cells in tissue differentiation (ectodermal dysplasias skin, nails, teeth)
• **Sequences** - cascade irregularity each of which results from the initial single fault (from Potter)

• **Complexes** - association of defects resulting from disrupted development of the developing areas depending (malformation of the brain midline + narrow rear eyeballs)

• **Associations** - accidental occurrence of defects associated with a person (Vater, Vacterl)

• **Syndroms** – Down Syndrome
Two groups of defects due to life threatening
First group:

Disadvantages of damaging the basic functions of the system and requiring immediate surgical correction in the first hours or days after birth.
First group

1. Obstruction of the gastrointestinal tract:

→ congenital obstruction of esophagus
→ of duodenum
→ of small intestine
→ of large intestine;
→ of rectum and anus.
First group

- Opened defects
- Abdominal wall defects (umbilical cord hernia, gastroschisis)
- Diaphragm defects
- Heart Defects
First group

Acute dyspnea

Pierre – Robin syndrome;
giant congenital lung cysts;
congenital lung lobe emphysema;
vascular clamping rings esophagus and trachea.
First group

Infections of the peritoneal cavity:
perforation of the stomach and intestines;
turn mesentery;
necrotizing colitis;
obstruction of meconium
Second group

Not so urgent – treatment after some days, weeks
Second group

1. orofacial cleft.

2. Some birth defects of the skeletal system
   hip dysplasia,
   club feet - varus
Congenital anomalies of the gastrointestinal tract
1. Congenital obstruction of the esophagus.
classification:

6 – level division depending on the connections and coexistence obstruction esophageal fistula - tracheoesophageal by Gross
A. Obstruction without fistula.
B. - with upper fistula
C. - with lower fistula
D. - with two fistulas – upper and lower.
E. Isolated H – esophageal-tracheal fistula
F. Atresis of esophagus

without fistula
Rycina 100. Klasyfikacja postaci wrodzonej niedrożności przelyku (W.Gross): A - niedrożność bez przetoki przelykowo-tchawicznej, B - niedrożność z górną przetoką, C - niedrożność z dolną przetoką, D - niedrożność z górą i dolną przetoką, E - izolowana przetoka typu H, F - zwężenie przelyku bez...
→ 82 – 86% obstruction of upper part with connection of lower with trachea
Diagnosis prenatal

→ visible since 23 HBD:

- USG,
Symptoms

→ polyhydramnion (A);
→ lack of gas in the gastrointestinal tract (A, B);
→ mucuse of the mouth and nose;
→ cyanosis (when the fistula into the trachea);
→ birth weight - about 1800g;
→ the upper fistula - a catheter stops 6 - 10 inches from her Cavity;
→ the lower fistula - breath sounds in abdomen (symptom Drescher);
Diagnosis

X ray
Can’t be fed!!!

Very high risks of

aspiration pneumonia;
chemical damage to the epithelium.
Surgery

- when the distance stretches 2cm - connection end to end and closing the fistula (80 %) ;
- when the distance stretches 2cm - Primary postponement of esophageal anastomosis (20 %) :
  • the spontaneous / mechanical extension sections of the esophagus
Surgery

→ If there’s no long enough part - steps
  • gastrostomy and selecting the top section on the neck
Post surgery care

stay in the ICU

The early extubation;

parenteral nutrition

antibiotic;

inhalation;

physiotherapy;

enteric feeding through a tube or gastrostomy after the return of peristalsis (about 36h);

oral feeding in 8 - 10 days after control test of the esophagus.
complications

- anastomosis dehiscence - pleural empyema ;
- anastomotic stenosis ;
- esophageal dysfunction ;

These newborns need to conduct post-operative for a year and periodic expansion of the esophagus.
2. Atresio ventriculi
Stomach atresia

→ concerns pyloric / parts antrum;

represents 1% of the total occlusion.
3. Atresia of duodenum
Etiology

→ disorder of proliferation and neural tube recanalization intestinal;
  family history?
  environmental factors (infection in pregnancy, drugs, radiation, hormonal disorders);

1: 4000 - 5000 births.
Types

1. Internal obstacle:
   atresia the membranous septum;
narrowing the membranous septum
Types

2. External Obstacle

- Annular pancreas: the oppression of one of the pancreatic primordia descending part
- Lada band - band of connective between undescended cecum and the upper right back wall of the abdomen.
Prenatal diagnosis

→ USG „double bubble”

  – double bubble fluid amniotic fluid in the proximal intestine ;
Symptoms

vomiting in the first day of life, with a touch of bile;
high retention in the stomach;
epigastric bloating – should be sucked
X ray
Before surgery

- Intragastric probe
Surgery

→ membranous atresia - excision of obstacles ;
  atresia total - anastomosis duodeno - duodenal (side to side, diamonds ) ;

  Annular pancreas - duodenojejunalostomia

Never cut the pancreas circinate !!!!!!
4. Congenital atresia of small intestine
→ type I
- continuity of the bowel wall preserved;
- closed septum;
- expansion of the proximal part of the intestine
- empty, narrow distal part of the colon.
→ **type II**
- blindly completed sections ;
- fiber bandwidth connection ;
- proximal : expanded , overgrown , atonic , necrosis , perforations ;
- mesentery - normal,sometimes the loss of the "V"
- the length of the intestines correct
→ **type IIIa**
- blindly completed sections
- always separated ;
- frill - always defect " V " ;
- the length of the intestines - shorter
→ **type IIIb**

- pagodas syndrome = peel apples ;
- blindly completed, overgrown sections ;
- always separated ;
- frill - significantly large loss ;
- 50 % - short bowel syndrome.
→ **type IV**

- multiple atresia = Character string of sausages
- the length of the intestine largely reduced.
Rycina 134. Klasyfikacja niedrożności jelita cienkiego wg Bland-Suttona.
Surgery

- end to end anastomosis with cutout section which closer excessively bloated;
- membranous septum resection and putting a probe to a circumferential section;
- if the perforation and peritonitis - the emergence of both ends of the intestine.
6. Malrotations
Rycina 146. Postaci nieprawidłowego zwrotu jelita.
Abdominal wall defects

- **Umbilical hernia (omphalocoele)** - displacement of abdominal organs through a defect centrally located anterior abdominal wall covered with a hernia sac is not vascularized.

- **Evisceration (gastroschisis)** - displacement of the abdominal organs through the abdominal cavity mostly located to the right of the umbilicus. Eviscerated organs are covered with a bag.
Umbilical hernia

- Hernia umbilical cord - diameter of the cavity to 3 cm
- Small - loss of less than 5 cm
- Large - loss > 5 cm
Gastroshisis

• Frequently loops of the small intestine and colon, less stomach, liver and spleen exceptionally.
• Guts damaged, inflamed infection
• Often, intestinal malrotation
Surgical treatment

• Closure of the wall
• Shuster bag
Congenital diaphragmatic hernia

- At 7 weeks of gestation separating the chest from the abdomen
- This is a displacement of the abdominal cavity through the diaphragm into the chest
classification

- Latero-back diaphragmatic hernia back (Bochdaleka)
- Right side diaphragmatic hernia
- Sterno-rib diaphragmatic hernia (Morgagni)
- Pleuro-peritoneal canal
- One or bilateral absence of the diaphragm Hiatal hernia of the diaphragm
Symptoms

• respiratory failure
• sunken belly
• Peristalsis heard over the lung fields
Treatment

- Stabilization
- HFOV, NO
- Surfactant?????
- Cardio – vasculare system support
- Anti infection
HYDROCEPHALUS
CEREBROSPINAL FLUID CIRCULATION

Sites of CSF formation: 20 ml/h
- Choroidal plexus: 60%
- Ependyma of the ventricles: 40%

Flow due to:
- Pressure difference between intracerebral and extracerebral fluid spaces,
- Vasogenic pulsation,

Absorption in:
- Perineural and perivasal spaces in children,
- Pacchionian bodies.
HYDROCEPHALUS DEFINITION

Dilatation of the brain fluid spaces caused by intraventricular hypertension because of the imballanced formation and absorption of the csf.

The cause of intracranial hypertension:
- impacted flow of the csf,
- distaubyancy of the csf absorption,
- overproduction of the csf.

Infantile hydrocephalus descendings:
- brain tissue ischemia - neurodegeneration,
- delayed and limited brain neuronalisation.

COMMUNICATING HYDROCEPHALUS

NONCOMMUNICATING HYDROCEPHALUS
CAUSES OF THE HYDROCEPHALUS

CONGENITAL:
• isolated CNS malformations: e.g. cerebral aqueduct stenosis,
• Syndrom: Dandy – Walker, Chiari

AQUIRED:
• tumor,
• cyst,
• inflammation,
• bleeding.
DIAGNOSIS OF THE ACUTE HYDROCEPHALUS

INFANTS:
• distended fontanelle,
• suture dilatation,
• accelerated head circumferency grow up,
• psychomotor developmental delay,
• neurological symptoms:
  • sun set syndrom,
  • squizz,
  • bulbar syndrom,
  • quadriparesis.

ELEDERLY CHILDREN:
• headache,
• morning vomiting,
• somnolency, dysconcinous,
• suture dilatation till the 5th year of life,
• papiledema,
• risk of stem compression
DIAGNOSIS

OF THE CHRONIC HYDROCEPHALUS

HYDROCEPHALUS ACTIVITY ASSESSMENT:

• head circumferency measurement,
• repeated USG or CT scan,
• neuropsychological assessment,
• fundus oculi examination,
• persistent 24 h ICP measurement,
• lambda infusion test.
Methods:

- Continuous or intermittent CSF external amelioration,
- Internal drainage of the CSF = the IIIth ventriculostomy, cyst fenestration, pelucidotomy,
- Persistent internal diversion of the CSF: valve system method = shunt method,
- Causative treatment of the hydrocephalus.
TREATMENT OF THE HYDROCEPHALUS

TYPES OF THE SHUNTS

VENTRICULO-PERITONEAL SHUNT

VENTRICULO – AMNIOTIC SHUNT
24-31 WEEK OF GESTATION

VENTRICULO – ATRIAL SHUNT
TREATMENT OF THE HYDROCEPHALUS

Do not treat stable, nonactive hydrocephalus

- The CNS and other body parts inflammations,
- Active bleeding in the CNS,
- Protein level in csf over 1.5 g/l.
Symptoms depend on shunt type:

• First symptoms:
  – Mostly: fever without any cause,
  – Shunt malfunction,
  – Dyspepsia.

• Following symptoms:
  – Sepsis,
  – Elevated ICP, disconcinous,
  – Cariopulmonary insufficiency,
  – Shunt nephritis,
  – Abdominal symptoms – ileus, peritonitis, palpable cyst.
COMPLICATIONS OF THE VALVE SYSTEM METHOD

TECHNICAL COMPLICATIONS

Symptoms of the elevated ICP, cause:
• Shunt occlusion:
  clot, choroid plexus, ependyma,
• Shunt disconnection/displacement.
Periferal symptoms, cause:
• Peritoneum: volvulus, ileus, perforation
• Heart perforation.
COMPLICATIONS OF THE VALVE SYSTEM METHOD

OVERDRAINAGE SYNDROM

Symptoms:

- Fall in of the fontanelle,
- Deceleration of the growth of the head,
- Headache and vomits disappering in lying position,
- Somnolency,
- Cerebellar symptoms,
- Cranium deformites,
- Slow filling of the valve antechamber,
- Diagnosis: CT scan

Slite ventricular system  Epidural hematoma
SPINAL DYSRAPHIC DEFECT

MENINGOCOELE

MENINGOMYEOLOCOELE

MYELOSCHISIS
THE TREATMENT OF THE NEURAL TUBE DEFECT

THE TARGET: DEVELOPMENT FOR LIFE

- PRENATAL TREATMENT OF HYDROCEPHALUS AND MENINGOCOELE,
- DEFECT POSTNATAL CLOSURE,
- SHUNT IMPLANTATION IN ACTIVE HYDROCEPHALUS,
- NEUROLOGIC, ORTHOPEDIC, UROLOGIC TREATMENT,
- SYSTEMS OF THE PSYCHOMOTOR REVALIDATION, SOCIAL ACCEPTANCE.
MENINGOCOELE
SURGICAL TREATMENT

TARGET:
MENINGITIS AND
NEURODEGRADATION
PROTECTION.

METHOD: 
DEFECT CLOSURE.

TIMING: 
OPEN: 24 – 48 HOURS AFTER 
DELIVERY 
CLOSURE: 72 HOURS AFTER 
DELIVERY.

CONTRAINDICATION: 
CONTROVERSIAL
• LORBER-ZACCHARY SCALE
• ALL CHILDREN ARE 
TREATED
MENINGOCOELE
COEXISTING AND DESCENDING CONDITIONS

- Neurogenic anorectal channel 90%
- Hydrocephalus 95%
- Lower extermititates pareses and deformities 80%
- Neurogenic bladder 90%
MENINGOCOELE
VERTEBRAL DEFORMITIES: NEUROGENIC, OSTEOGENIC

LUMBAR SCOLIOSIS

LUMBAR KYPHOSIS IN THE SITE OF MENINGOCOELE
MENINGOCOELE
DECUBITUS

CAUSES:
• SENSORY SKIN DISTURBANCES,
• ISCHEMIA IN THE SITE OF SENSORY INNERVATION.

PROPHYLAXIS:
• BODY POSIOTION CHANGES EVERY 30 MIN.,
• EXACT BODY HYGIENE,
• COMFORTABLE CLOTHS,
• CAREFUL WEARING OF ORTHESSES.

TYPICAL SITES OF DECUBITEST IN DYSRAPHIC CHILDREN

CONTAMINATED DECUBITUS = HIGH RISK OF SEPSIS
MENINGOCOELE
SECONDARY TETHERING AFTER RECONSTRUCTIVE SURGERY

CAUSES:

• PLACODA TO BROAD,
• PLACODA ADHESIONES,
• KYPHOSIS,
• OTHERS NOT CORRECTED SPINE MALFORMATIONS

SPINAL CORD DOES NOT MOVE UP IN THE TIME OF BODY GROWTH
DISTENDING OF THE VESSEL AND SPINAL ROOTS

MRI - TETHERING OF THE THORACAL MENINGOCELE
NEUROTERATOGENESIS
OPENED GROOVE DROPS INTO MESENCHYMA

SPINAL CORD LIPOSOMAS OF:
A) CONUS MEDULLARIS,
B) FILUM TERMINALE.

SYMPTOMS:
• SACRAL SUBCUTANEOUS TUMOR,
• TETHERING OF SPINE.

TREATMENT:
• SUBTOTAL LIPOMA REMOVAL,
• DURA MATRIX PLASTIC OP.
NEUROTERATOGENESIS

NEUROECTODERMA NOT COMPLETELY SEPARATED FROM SUPERFICIAL ECTODERMA

SYMPTOMS:
• MOSTLY CONTAMINATED FISTULAS IN THE BODY MIDLINE,
• MENINGITIS, BRAIN ABSCESS.

TREATMENT:
• TOTAL REMOVAL OF THE FISTULAS AND DG TUMORS

A) NEURO-DERMAL FISTULA,
B) DYSONTOGENETIC TUMORS