Paediatric Pathology

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Paediatric vs ‘adult’ pathology

Scope-defined by the age
Children vs adults

Disorders are unique to children

Susceptibility to injury varies

Diagnosis requires knowledge of each stage of development
Children vs adults

Therapeutic considerations

Variable outlook for recovery from the same disease
Infant mortality 2002, England & Wales, main causes

- Immaturity: 45%
- Congenital anomalies: 28%
- Others: 27%
Childhood deaths

- Accident / injury: 33%
- Cancer: 22%
- Neurological: 12%
- Congenital Abnormalities: 11%
- Others: 33%
Role of Paediatric Pathologist

- Diagnosis
- Prognosis
Case 1

10-day old former 27 week male

Abdominal distension

Rapid clinical deterioration
Complications of preterm birth

- Respiratory distress syndrome
- Necrotising enterocolitis
- Intraventricular haemorrhage
- Patent ductus arteriosus
At laparotomy
MICROSCOPIC EXAMINATION

Normal small bowel

NEC
Case 2

2-day-old boy

Failure to pass meconium

Abdominal distension

Bilious vomits
Differential diagnosis

Hirschsprung disease

Meconium ileus

Atresia
Normal

Hirschsprung’s disease

GANGLION CELLS

Nerve trunks - abnormal
Hirschsprung’s disease - definition

Congenital absence of ganglion cells
Hirschsprung’s disease

- 1:5000
- M:F 4:1
- 10% familial
- 10% Downs syndrome
Hirschsprung’s disease—Complications

Enterocolitis

Perforation
normal vs aganglionic
Cystic fibrosis

Meconium ileus
Cystic fibrosis

1:2000 live births
most common lethal genetic disease
Cystic fibrosis

Thick tenacious secretions
A

Colonic (small)

Proximal ileum

Mid ileum contains thick and tenacious meconium

Distal ileum contains concretions
Bowel-cystic fibrosis
Bronchiectasis
Atresia

complete absence of a part of intestine

- Oesophageal
- Small intestine
- Biliary
Small intestinal atresia

Congenital malformation

Incidence 1 in 1500
Case 3

3-week old well baby
Jaundice
Persistent conjugated hyperbilirubinaemia
Biliary atresia

1:10,000 live births

Liver biopsy is the most reliable means of establishing the diagnosis

Surgery by 12 weeks
Normal

Biliary atresia
Liver cirrhosis
Case 4

2-yr-old boy
Failure to thrive
Diarrhoea
Coeliac disease - diagnosis

Clinical
Serology
Pathology
Clinical remission on gluten-free diet
Case 5

Newborn

Difficulty breathing
Respiratory tract malformations

Congenital diaphragmatic hernia
Respiratory tract malformations
congenital cystic adenomatoid malformation
Case 6

2-year old boy

Recurrent UTIs
Normal vs cystic kidney
Cystic kidneys

- Multicystic dysplastic kidney
- Autosomal recessive PKD
Dysplasia - definition

abnormal development and differentiation
Terms

Hamartoma vs Choristoma
Benign tumours

Haemangioma
Lymphatic malformation
Case 7

2 year old boy

Swelling in neck & shoulder
Diagnosis

Lymphatic malformation
Paediatric vs adult cancers

- Incidence
- Improved survival
- Type of tumour
Paediatric cancers - types

- Leukaemia: 35%
- Central Nervous System: 7%
- Lymphoma: 6%
- Neuroblastoma: 5%
- Renal: 5%
- Soft Tissue: 5%
- Bone: 3%
- Retinoblastoma: 3%
- Germ Cell: 3%
- Hepatic: 1%
% still alive

9 y | 51
6 y | 61
6 y | 70
5 y | 77

% still alive
Tumour specimens

- Biopsies
- Resections eg
  - Nephrectomy
  - Neuroblastic tumour
  - Partial heptectomy
Case 8

3-week old boy

Rapidly growing mass on the back
Differential diagnosis

- ✔ Neuroblastoma
- ✔ Rhabdomyosarcoma
- ✔ PNET/Ewings family
- ✗ Lymphoma
- ✗ Wilms tumour
The tumour handling kit

EM

TCM

-80°C

FFPE
Small Round Cell Tumours

- Undifferentiated, monotonous population of tumour cells
- Need special techniques to determine the histogenesis
Differential diagnosis

× Neuroblastoma
× Rhabdomyosarcoma
✓ PNET/Ewings family
× Lymphoma
× Wilms tumour
Case 9

2 year old boy

Huge abdominal mass
Wilms tumour
Teratoma

- tissue derived from all the three germ cell layers

- Benign, immature or malignant