Short Cases

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Case 1:

Beyond Bronchiolitis

Presentation

2 month old, term girl

Cough and rhinitis, increased work of breathing, wheeze and apnea x 1 day

Fatigue while feeding

Hypotonia

Hx: Entero/Rhino infection

Physical Exam

T36.4 RR46 SpO_2 94%

Noisy breathing

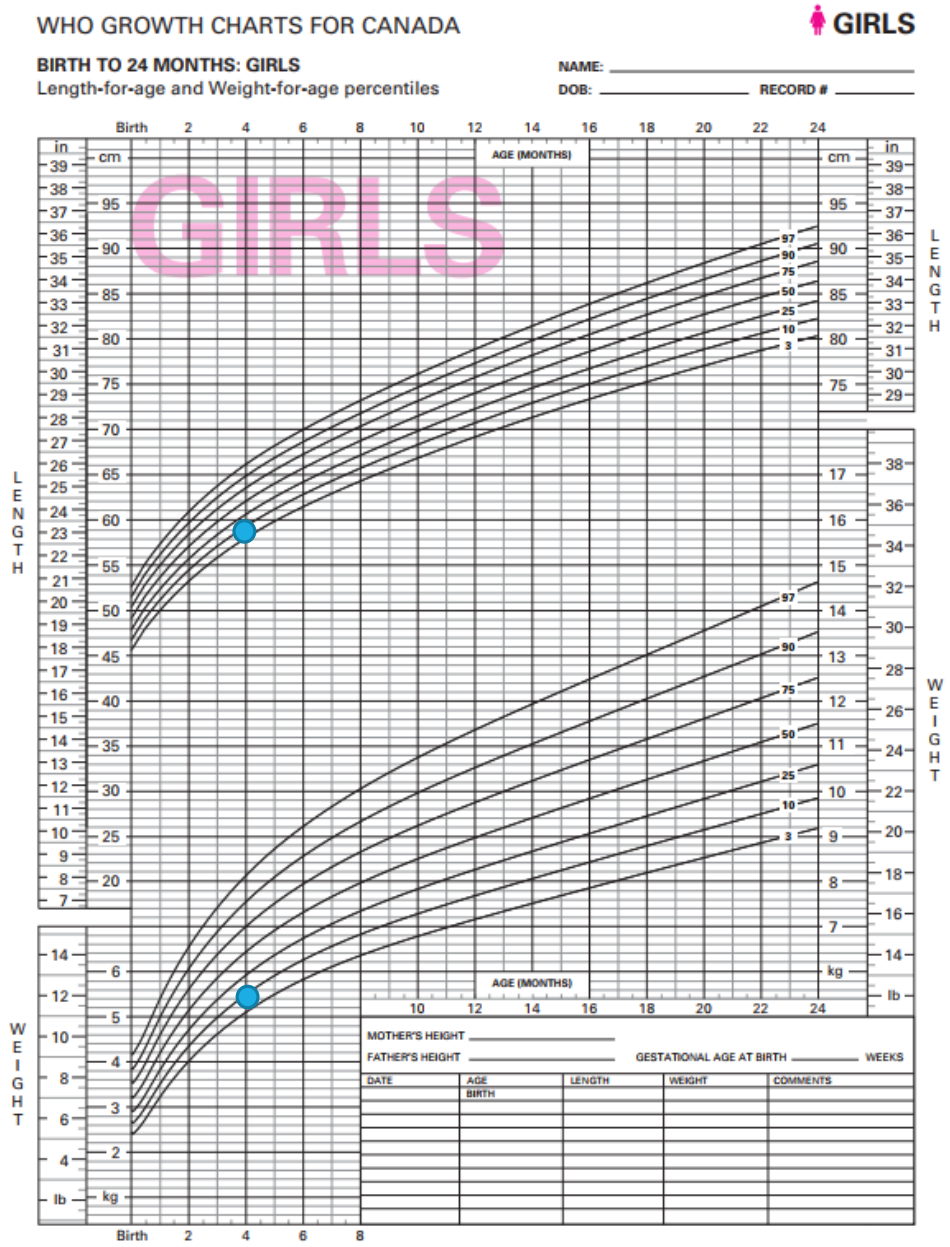
Not dysmorphic

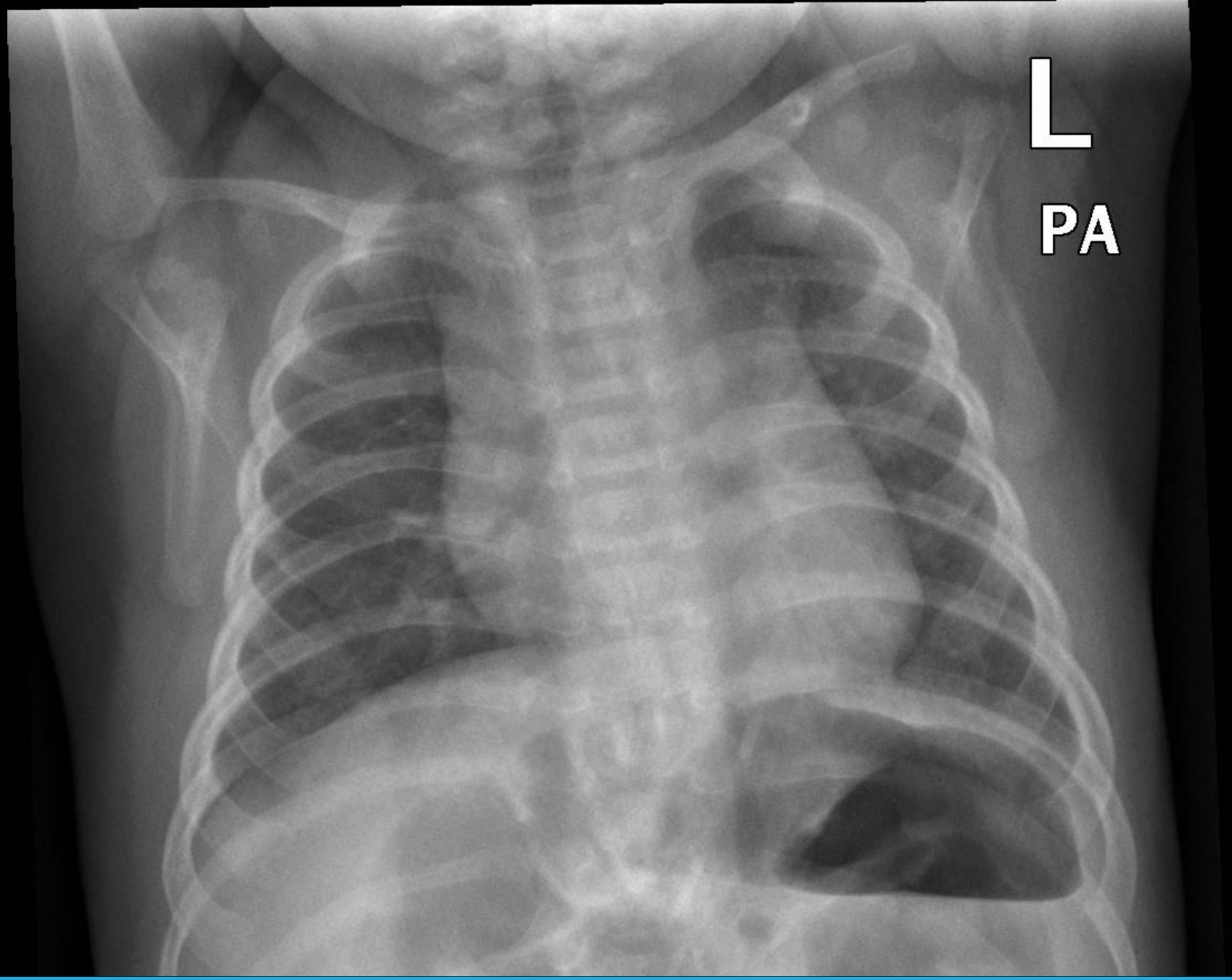
CVS normal

Air entry decreased on right

No wheezes, few scattered crackles

Hypotonic extremities and head lag

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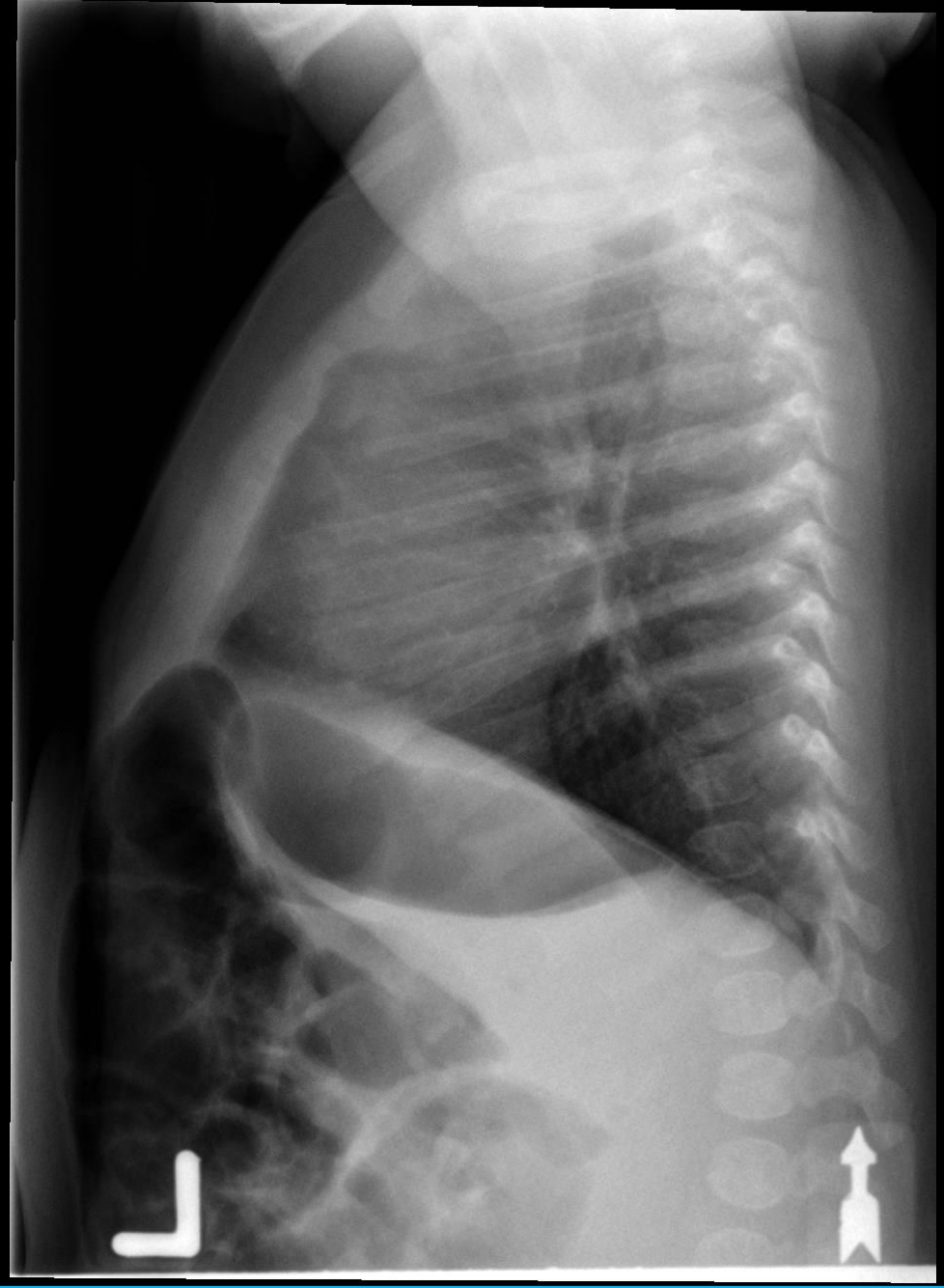
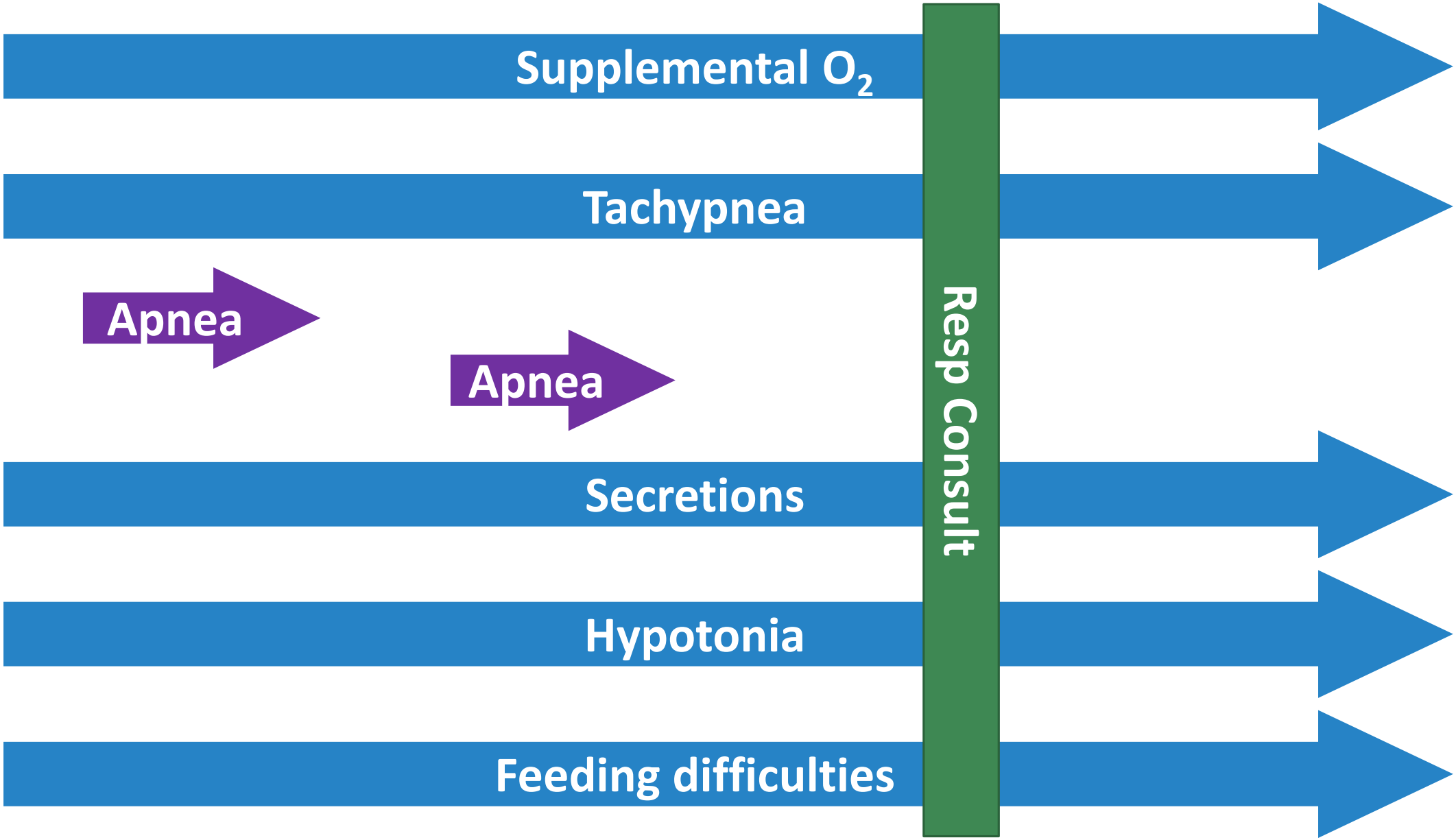
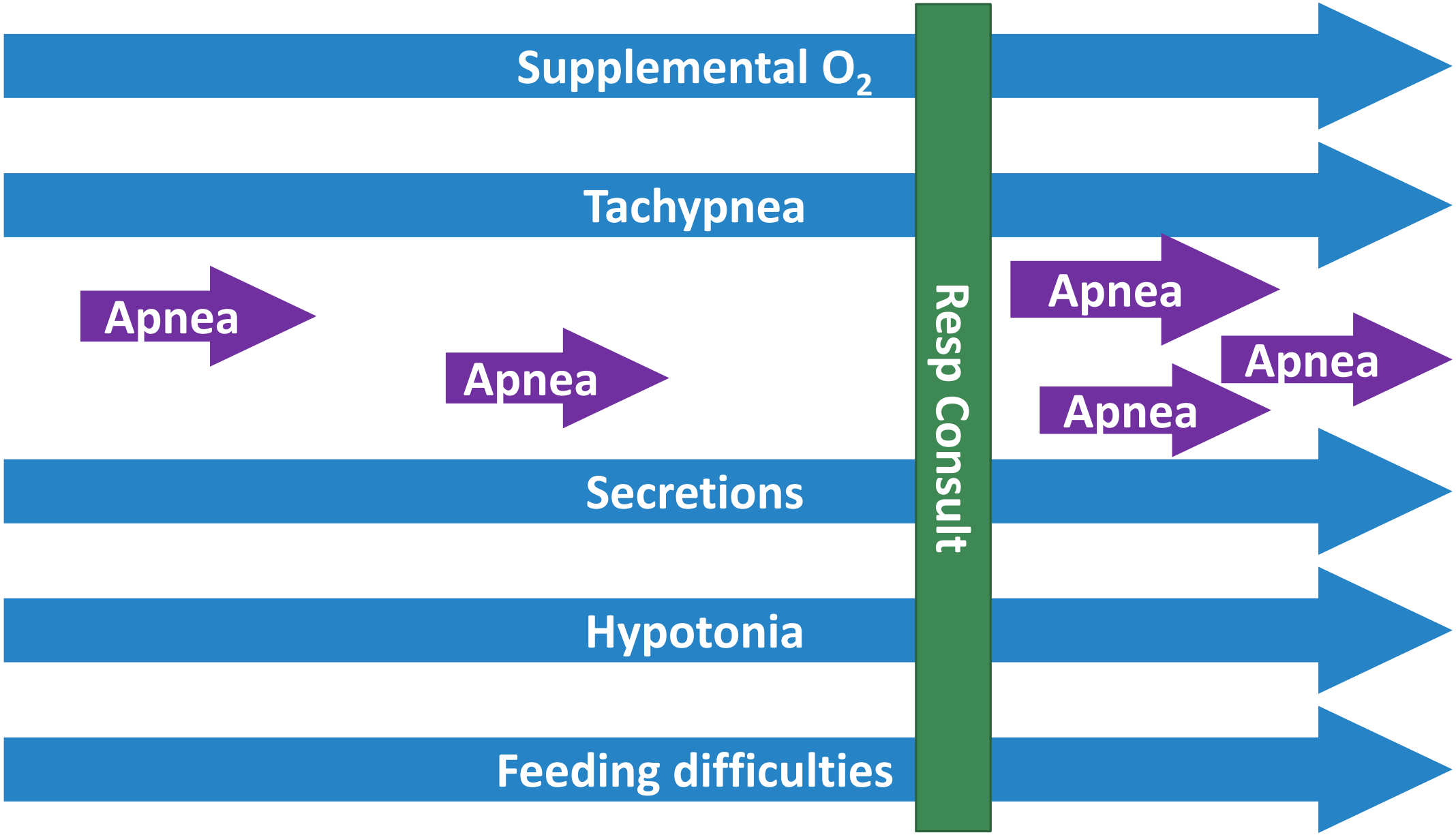


CBG	7.31 // 41 // 45 // 21
CBC	Hb 99 WBC 11.5, normal differential Plt 295
Electrolytes, Urea, Creatinine	Normal
CRP	52.3 ↑
Viral Panel	Entero/rhino positive


Supplemental O_2
Tachypnea
Apnea
Apnea
Resp Consult
Secretions
Hypotonia
Feeding difficulties

Investigations

Pertussis	Negative
Chlamydia	Negative
FEES	Not safe to orally feed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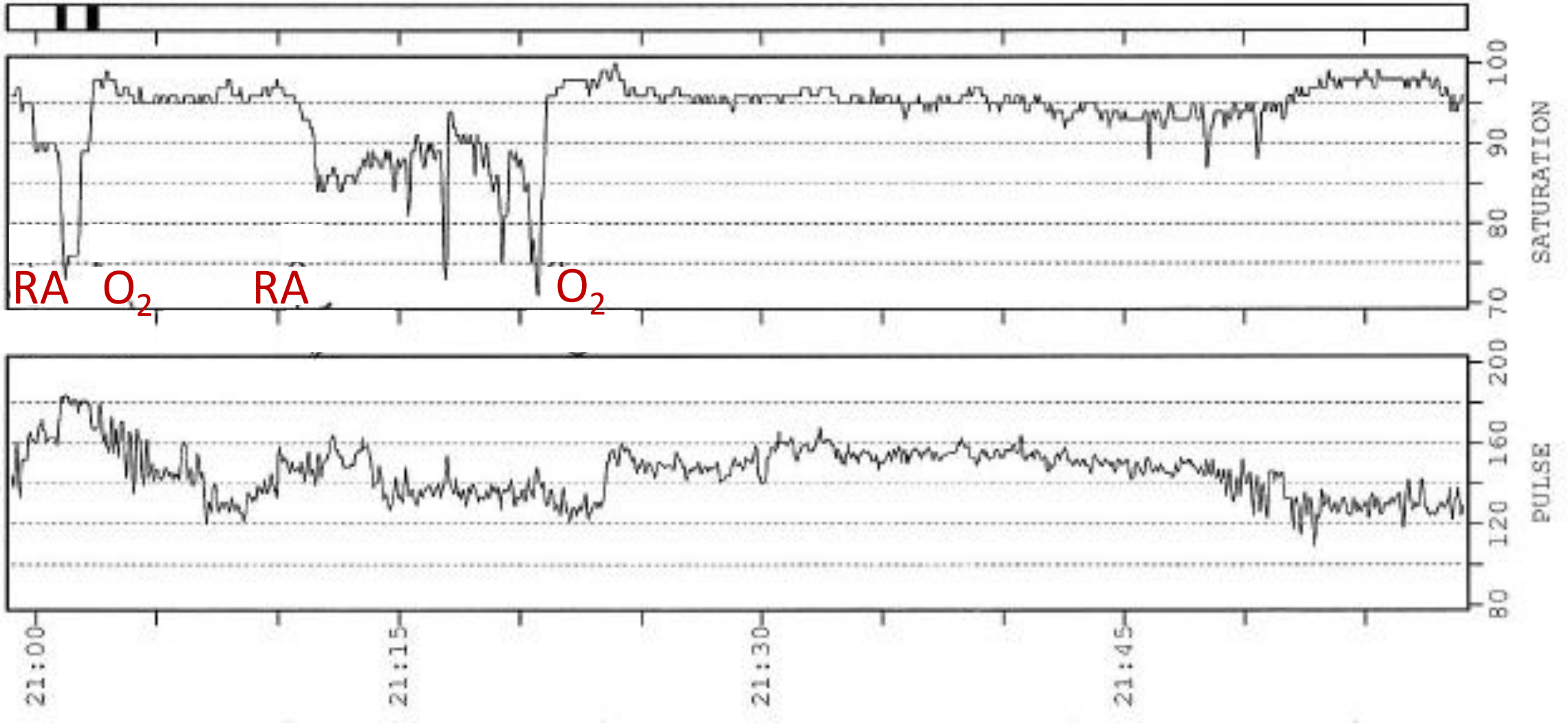
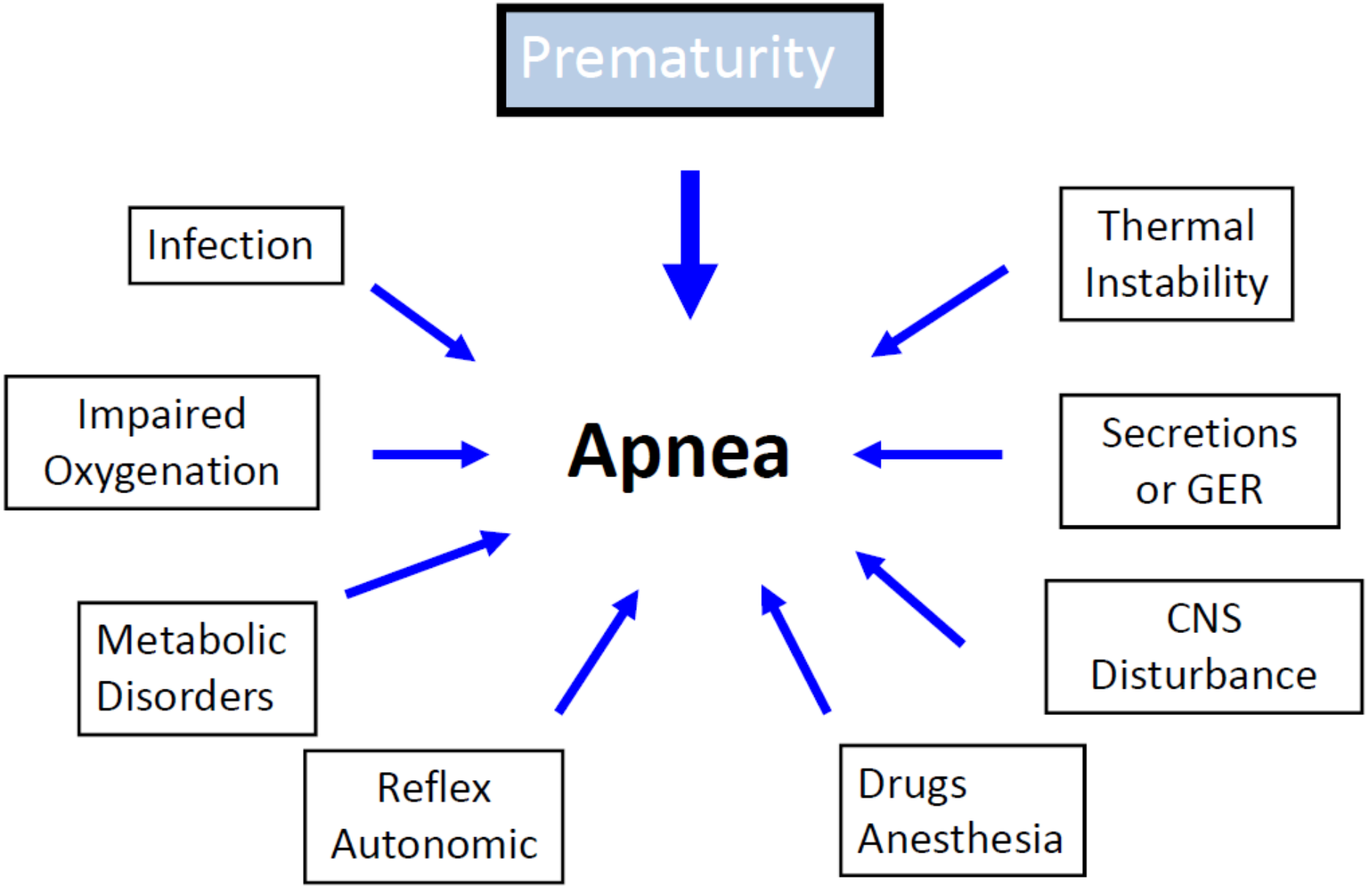
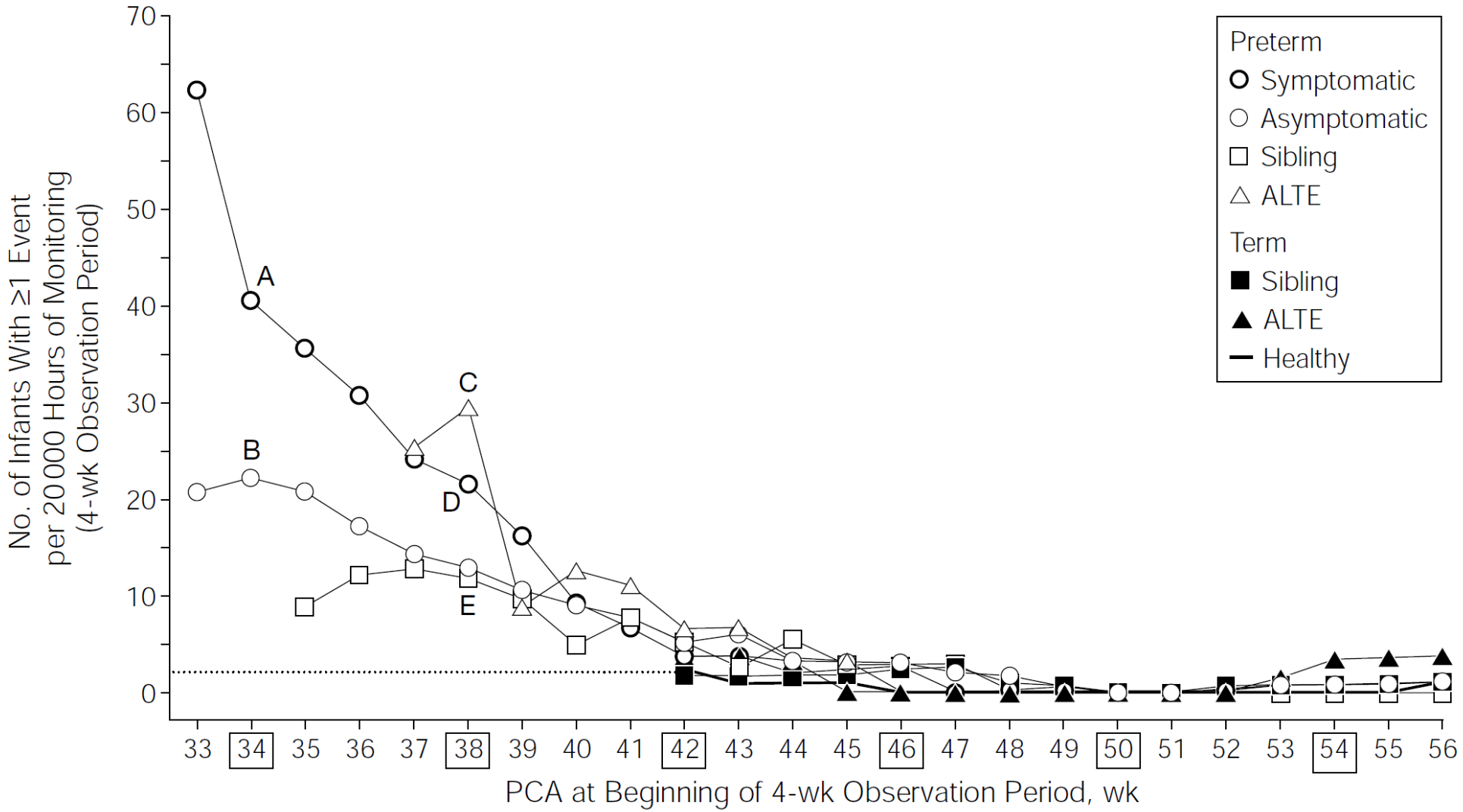

RA
O_2
RA
O_2
100
90
80
70
SATURATION
200
160
120
80
PULSE
21:00
21:15
21:30
21:45

“We would always watch her sleep…”


Prematurity
Infection
Thermal Instability
Impaired Oxygenation
Apnea
Secretions or GER
Metabolic Disorders
CNS Disturbance
Reflex Autonomic
Drugs Anesthesia

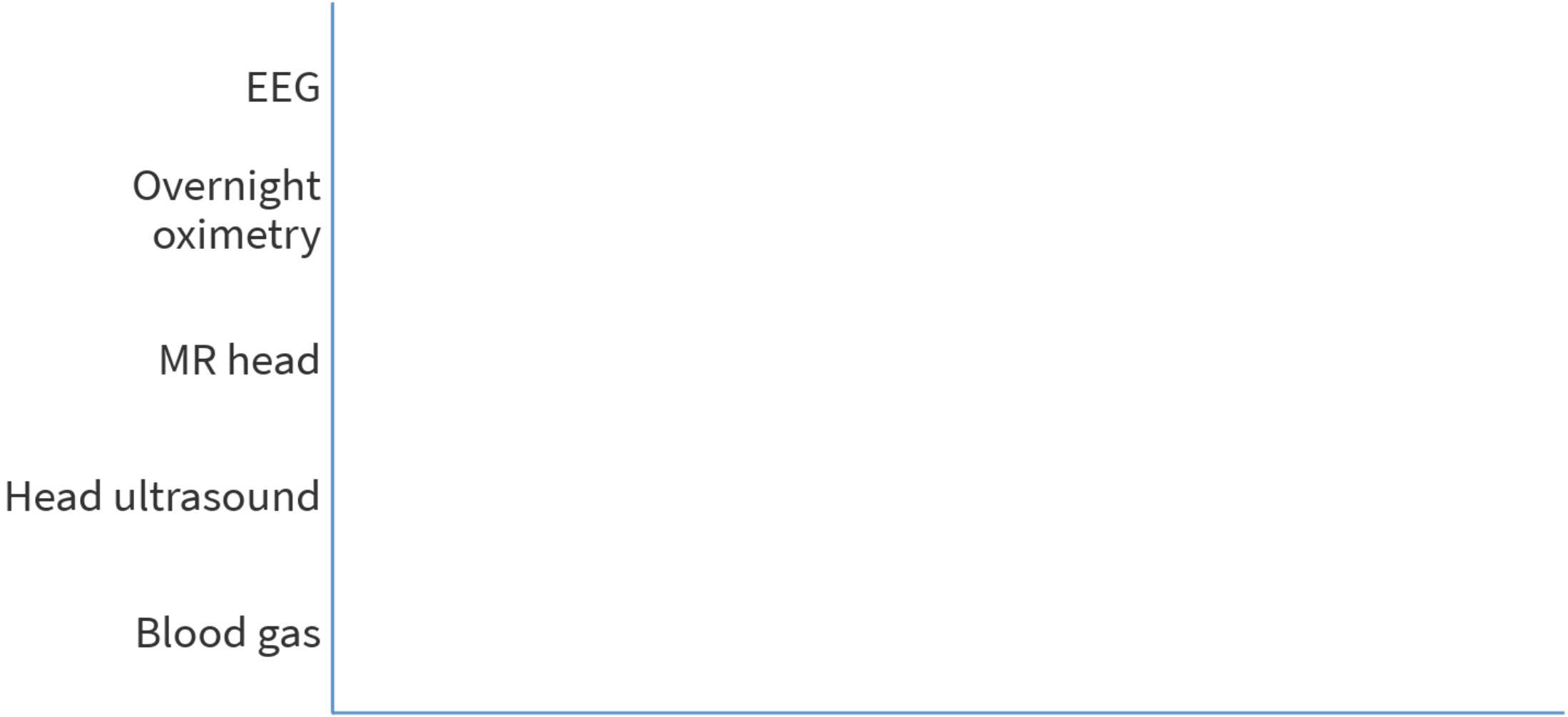
Rate of Infants with One or More “Extreme” Events



Ramanathan et al. JAMA 2001

Primary	Secondary
• Congenital • Late-onset central hypoventilation syndrome • Idiopathic hypothalamic dysfunction • Arnold-Chiari Malformation • Apnea of prematurity	• Trauma • Infection • Tumour • Central nervous system infarct • Asphyxia • Increase intracranial pressure • Metabolic • Drugs

Which investigation would you choose next?



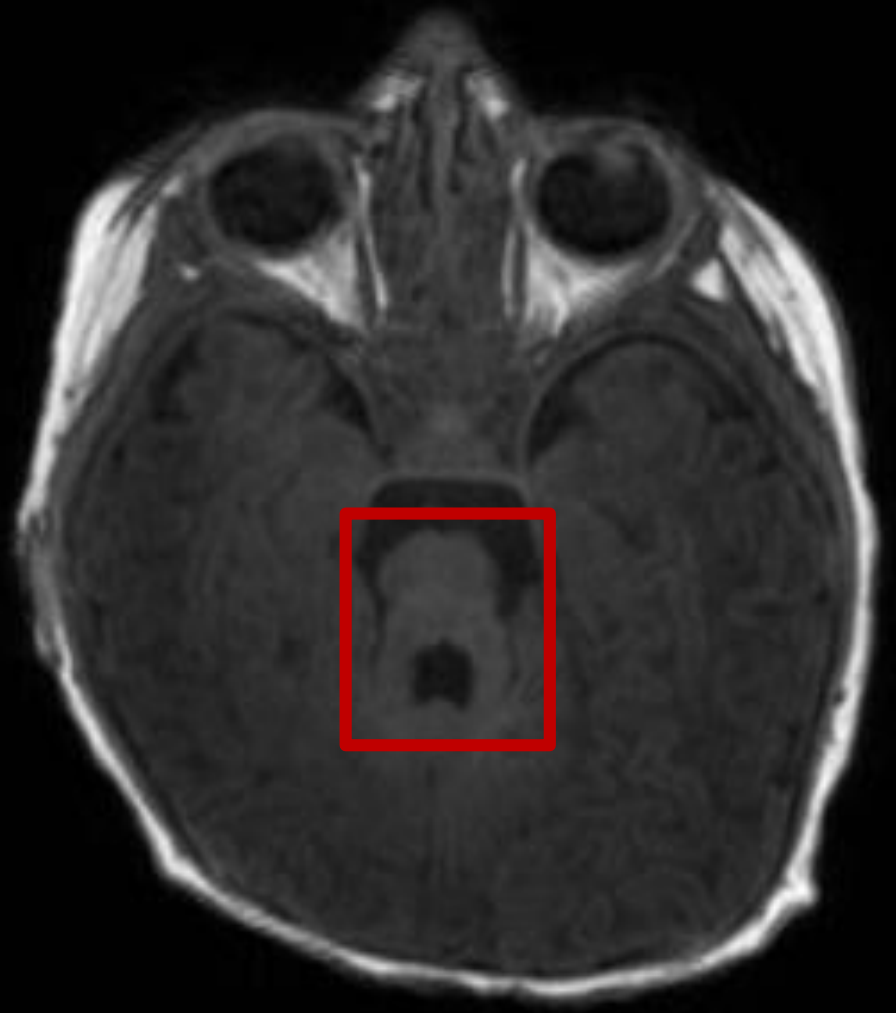
Limited guidance to direct evaluation of suspected central apneas

MR Head most routinely recommended investigation

Abnormal MR Findings

	OR (95% CI)
Abnormal neurologic exam	29.5 (1.2 - 702.0)
Clinical GERD	11.2 (1.6 – 78.4)

Woughter et al. Otolaryngol Head Neck Surg 2015



https://radiopaedia.org/cases/joubert-syndrome-2

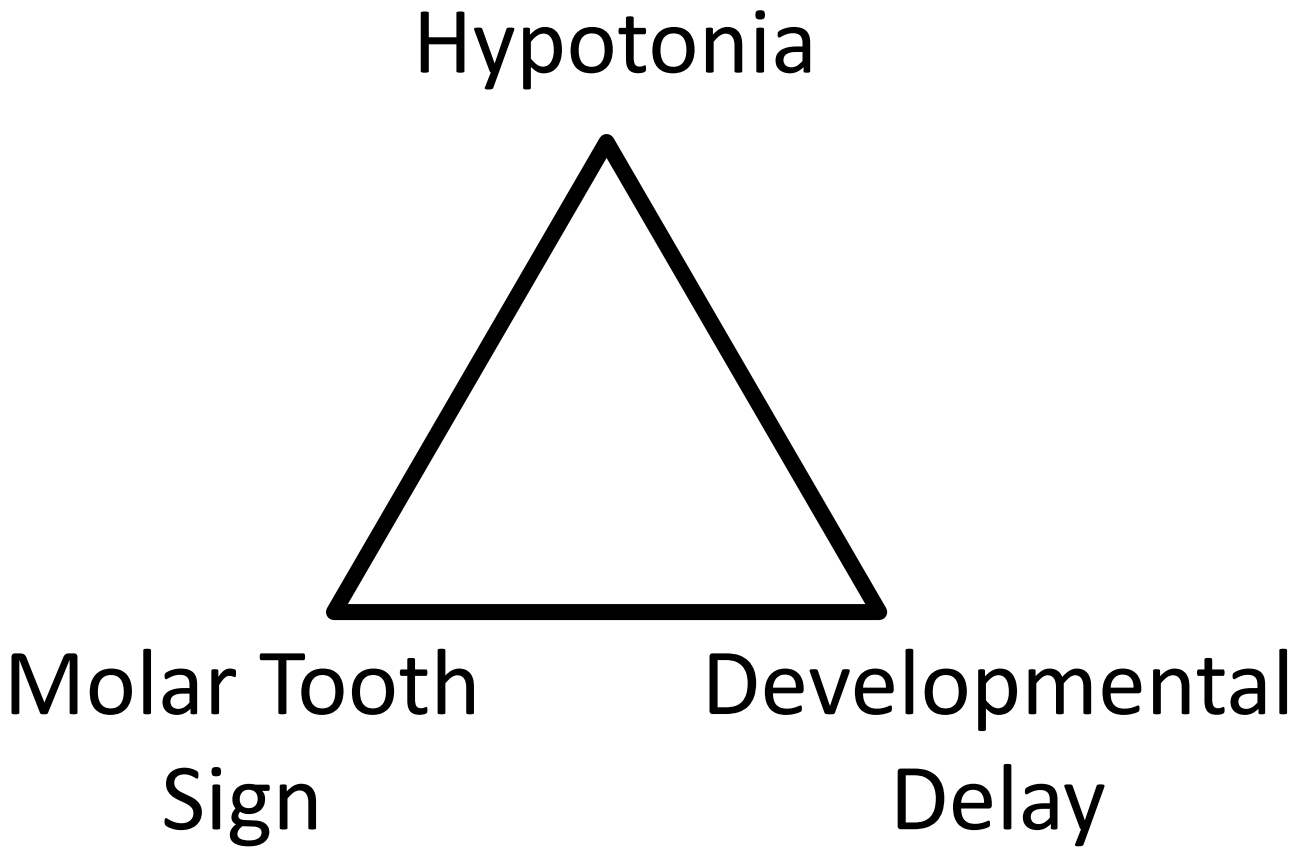
Joubert Syndrome

Prevalence 1: 100 000

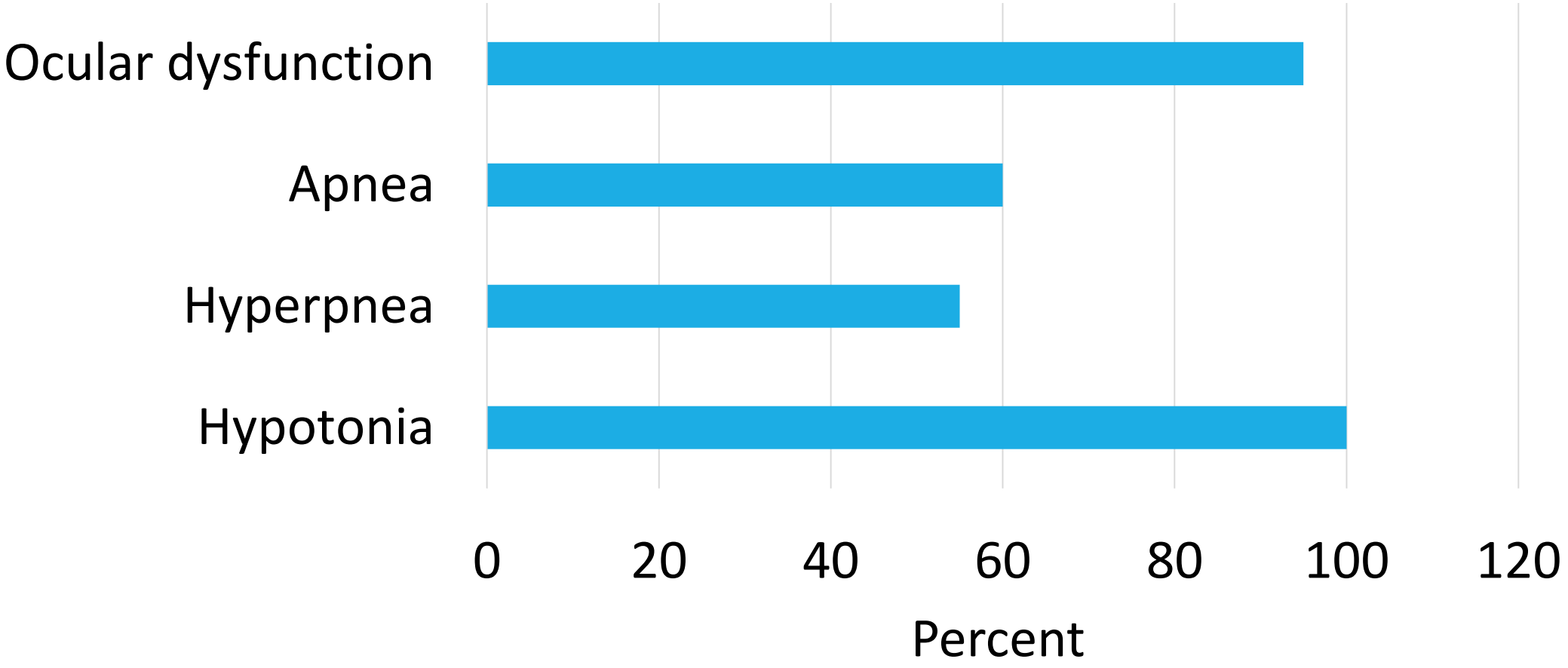
Autosomal recessive

- 34 genes

Ciliopathy

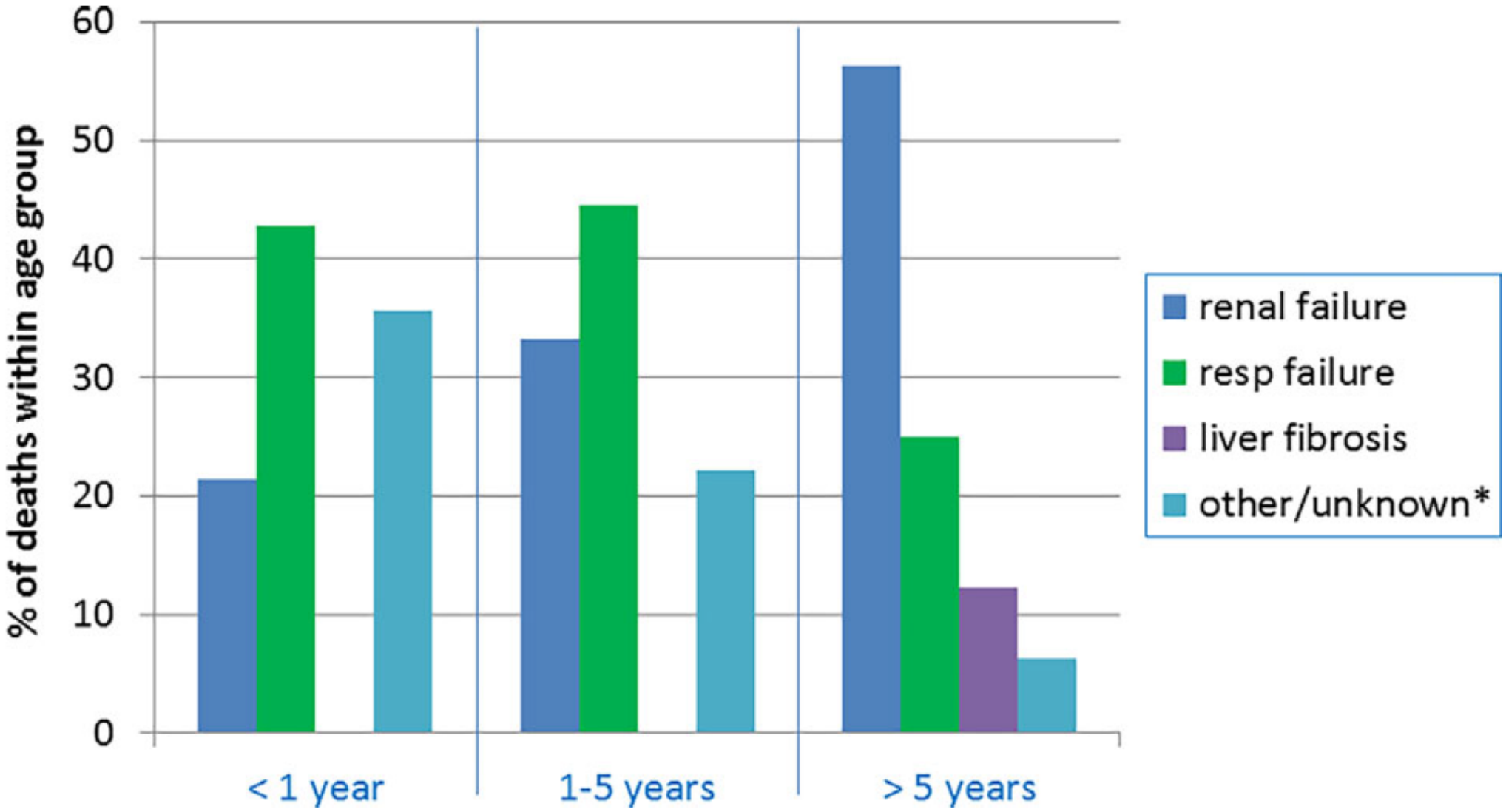


Additional Features



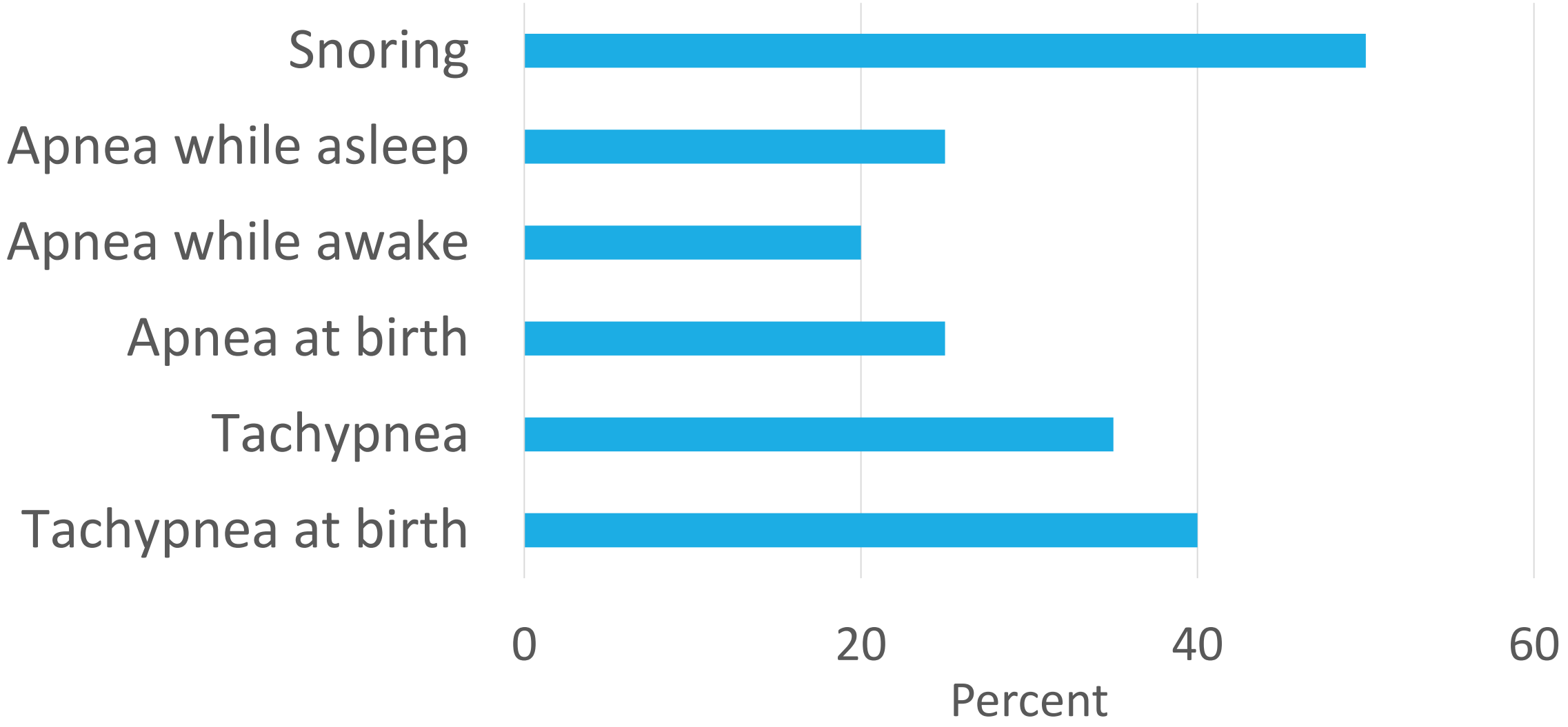
Maria et al. J Child Neurol 1999

Respiratory Prognosis



Dempsey et al. Am J Med Genet A 2017

Childhood Breathing Irregularities



Kamdar et al. Pediatr Neurol 2011

Respiratory Management

Spectrum of respiratory support:

- Low flow oxygen most common
- CPAP/BiPAP/tracheostomy much less common

Caffeine

Role unclear

Other considerations

Consider:

- Home monitoring
- Avoid opioids in anesthesia
- Use regional anesthesia

Disposition

Home oxygen
- 0.5 L/min for 24 hours daily

Home suction

Overnight caregivers

Exclusive NJ feeds

European Journal of Human Genetics (2007) 15, 511–521

www.nature.com/ejhg

PRACTICAL GENETICS

In association with orphanet

Joubert syndrome (and related disorders) (OMIM 213300)

Take Home Messages

Brain imaging useful with

- Neurologic findings
- Clinical GERD

Joubert syndrome

- Hypotonia, delay, and molar tooth sign
- Supportive management
- Breathing improves with time

Case 2:

It’s never too late

Presentation

9 year old girl, refugee

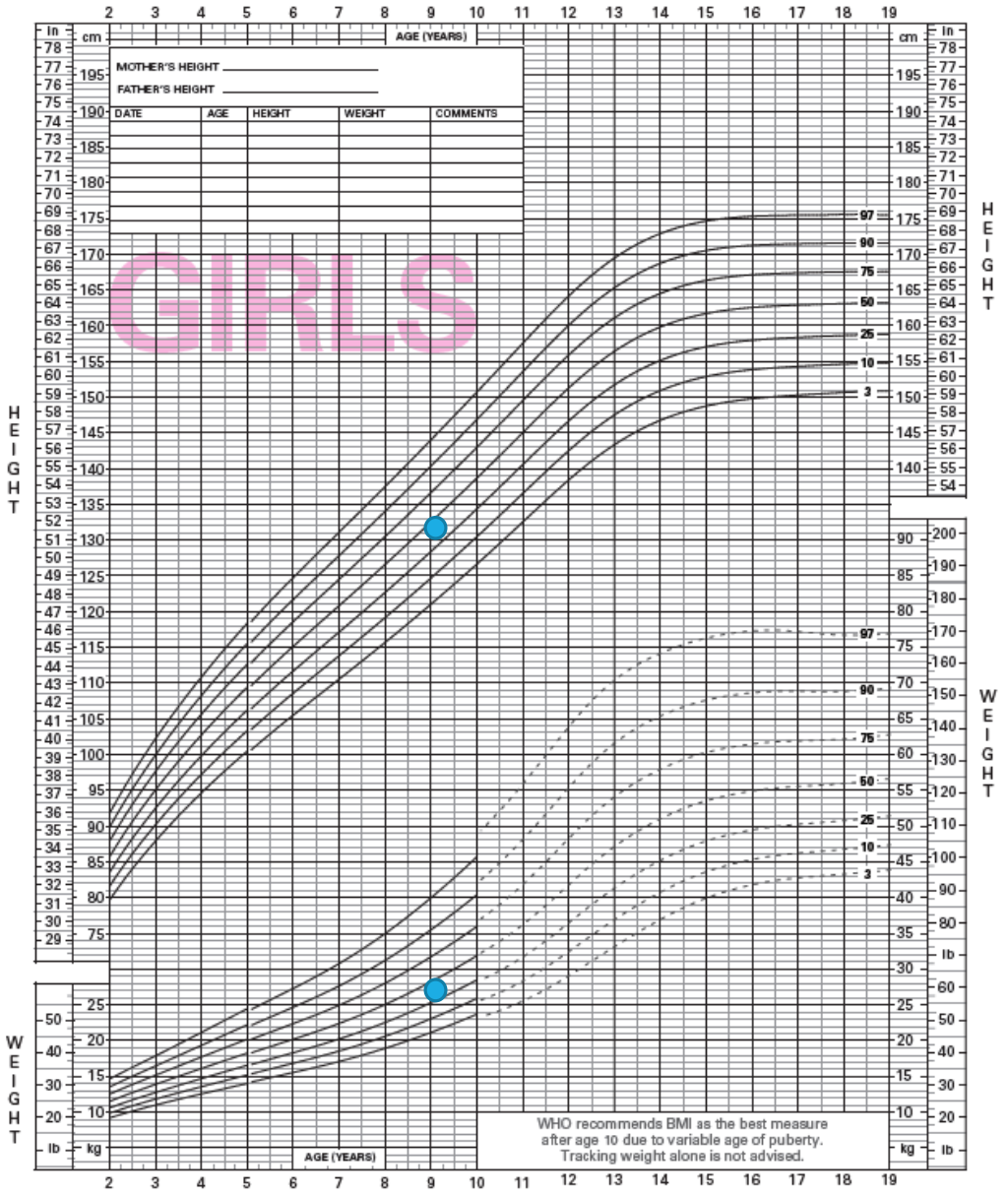
Cough x 16 days

Right chest pain

PMHx:
Normal pregnancy, delivery

Asthma (Ventolin prn)

Vitals	T 37.3°C RR 23 SpO_2 98% RA
CVS	Normal
Resp	Mild WOB Decreased on right No wheeze, no crackles
GI	Normal No organomegaly

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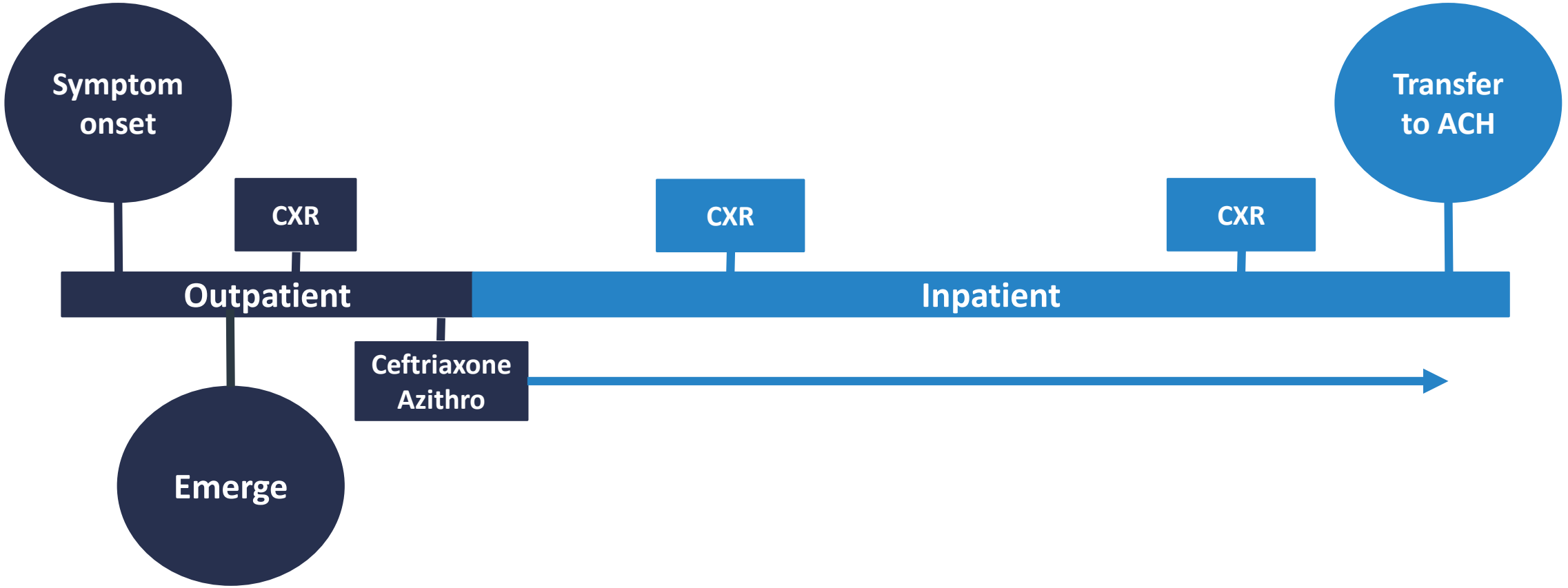
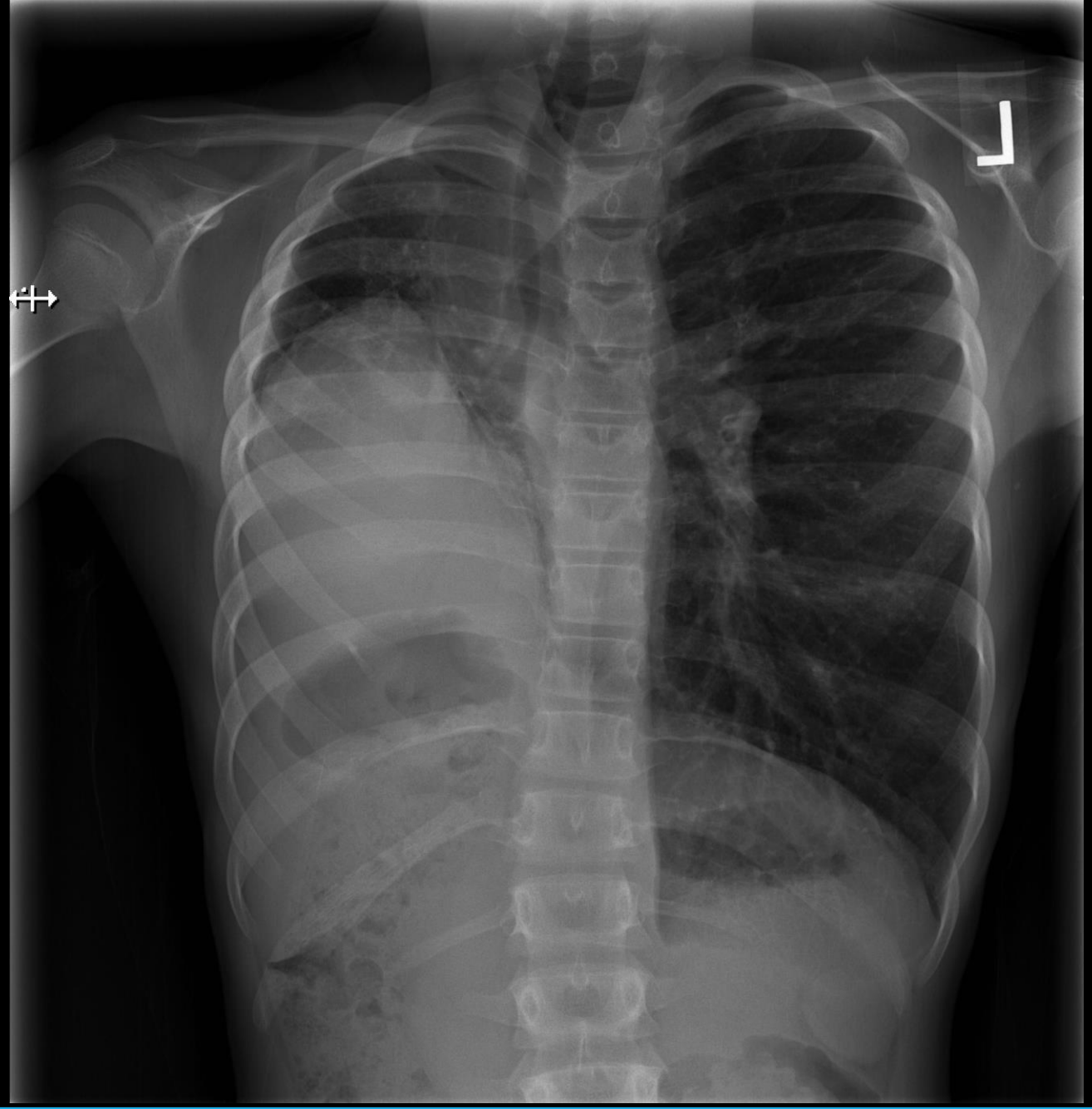
Timeline


Symptom onset
CXR
CXR
CXR
Transfer to ACH
Outpatient
Inpatient
Ceftriaxone Azithro
Emerge

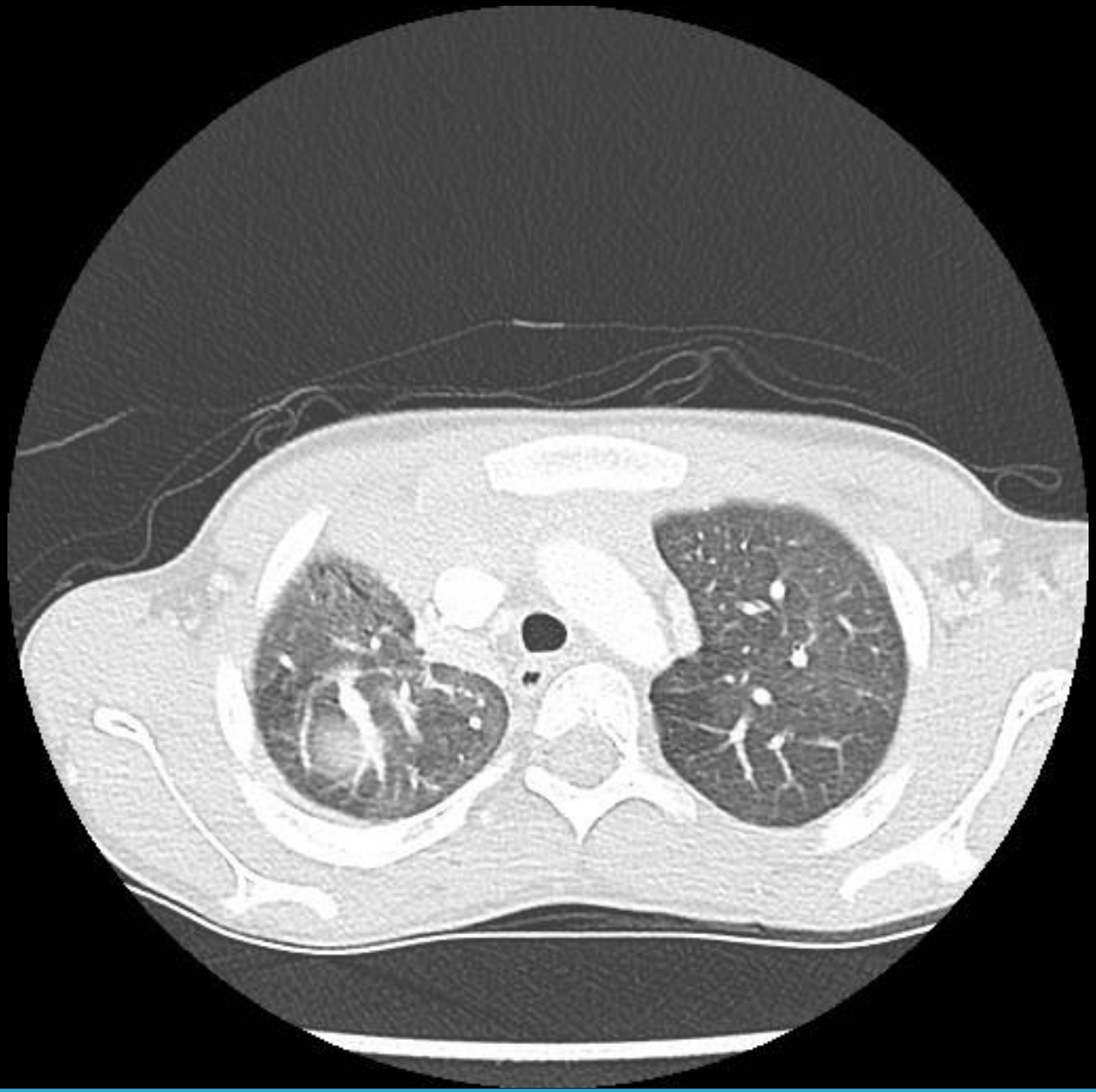


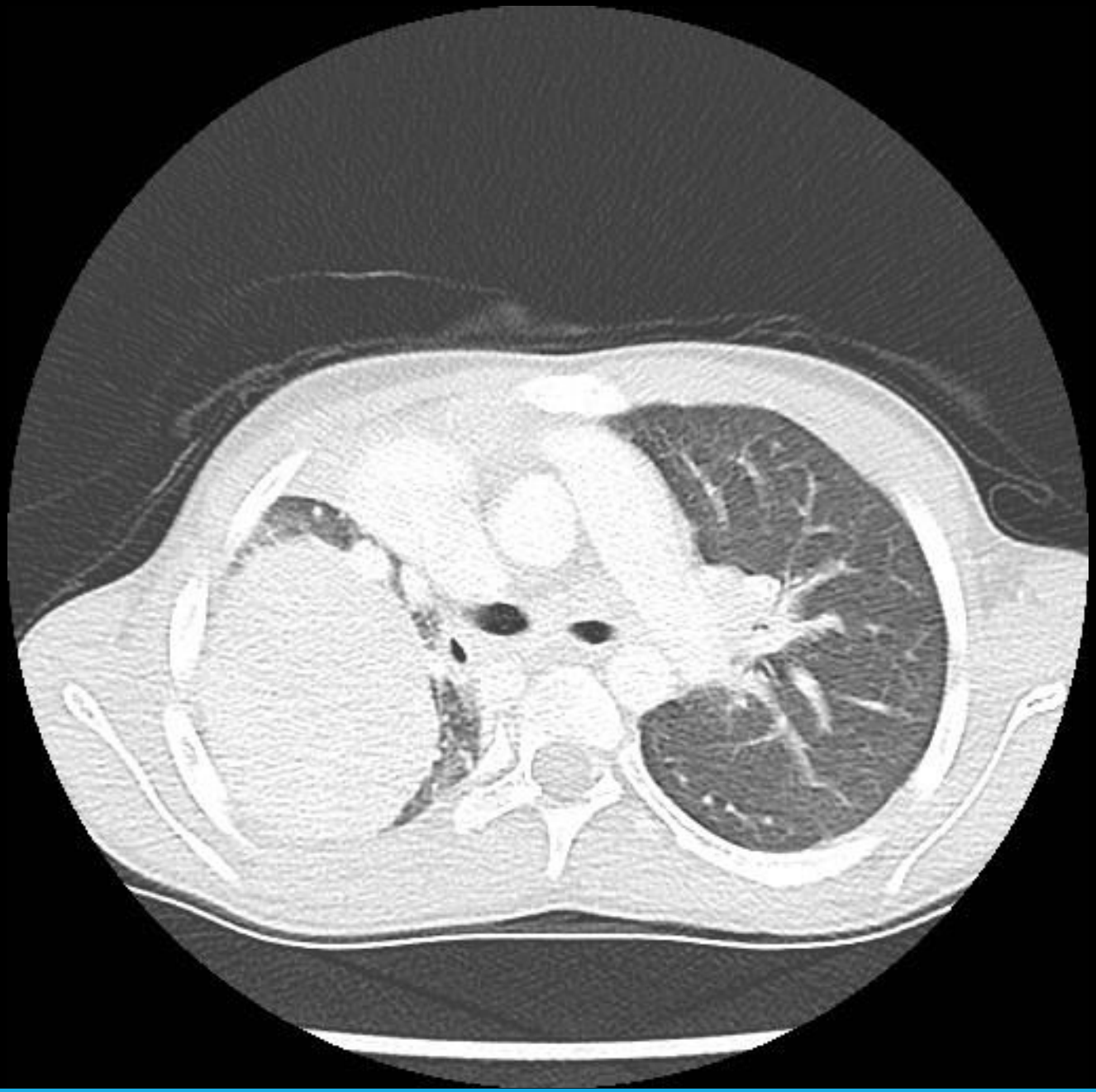
Day 7

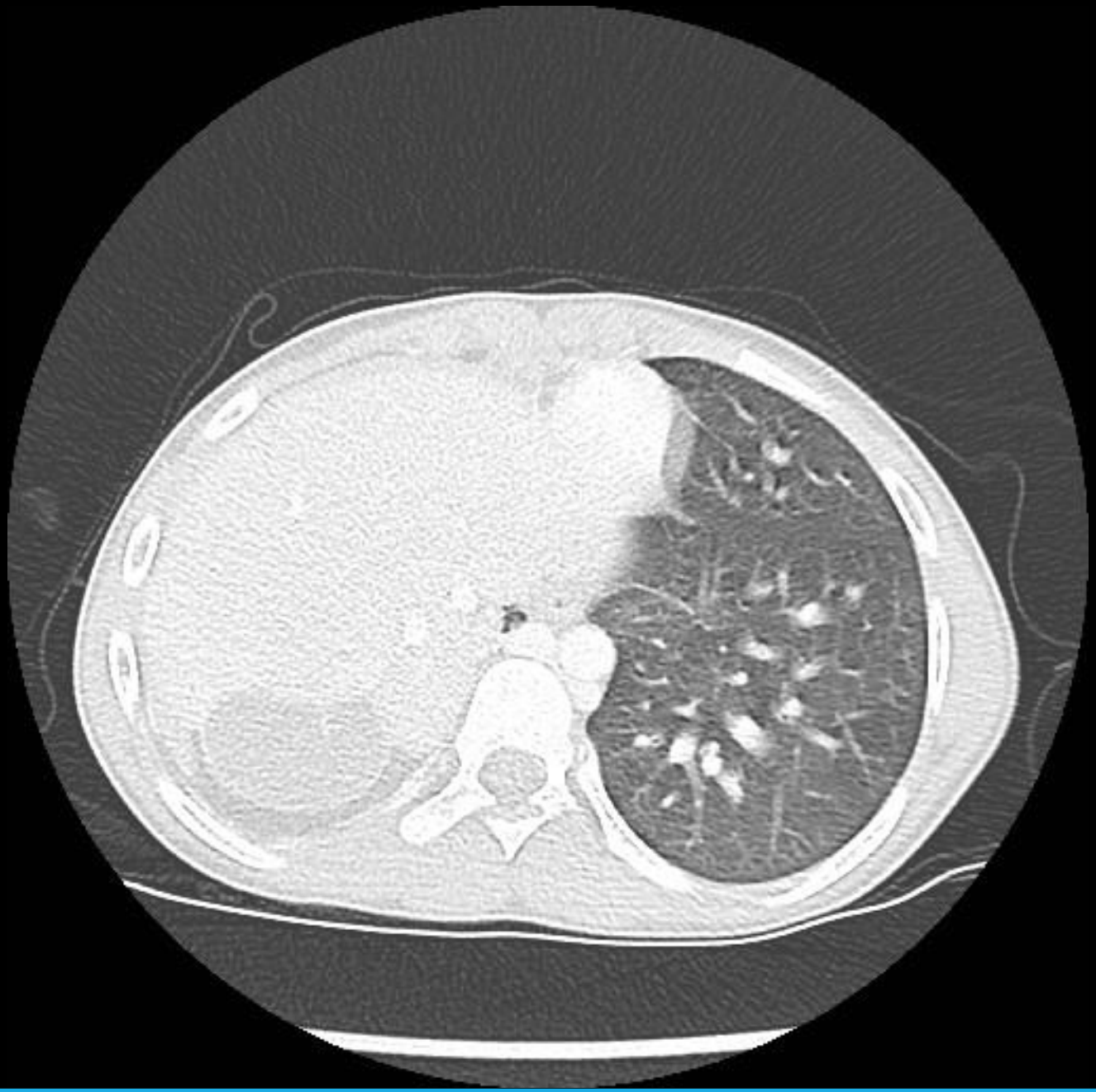
Viral panel	Negative
Pertussis	Negative
CBC	WBC 10.2, normal differential Hb 151 Plt 354
Electrolytes, Urea, Creatinine	Normal
Liver transaminases	Normal
Lipase	Normal
Bilirubin	Normal

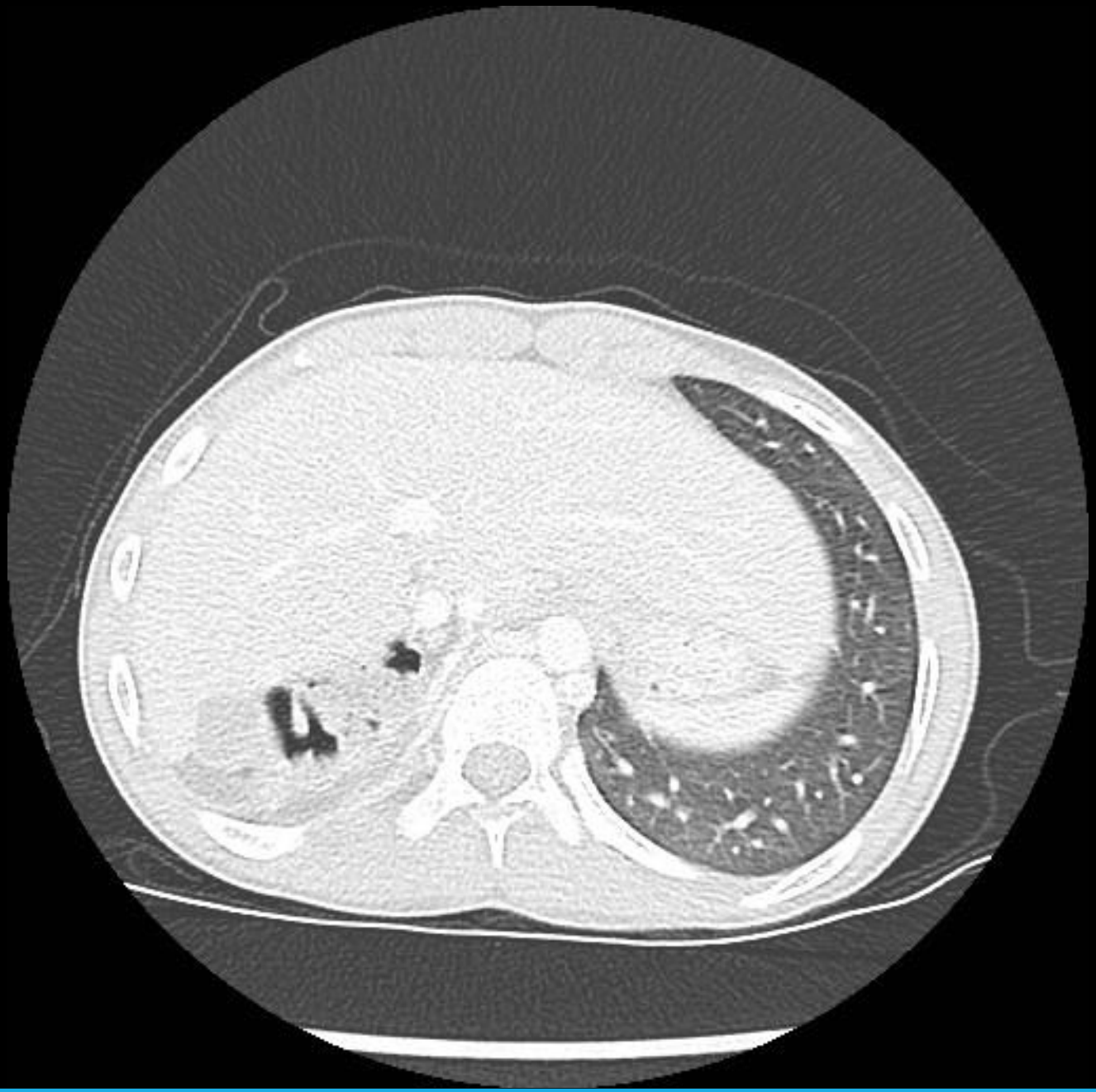
What is your differential diagnosis?

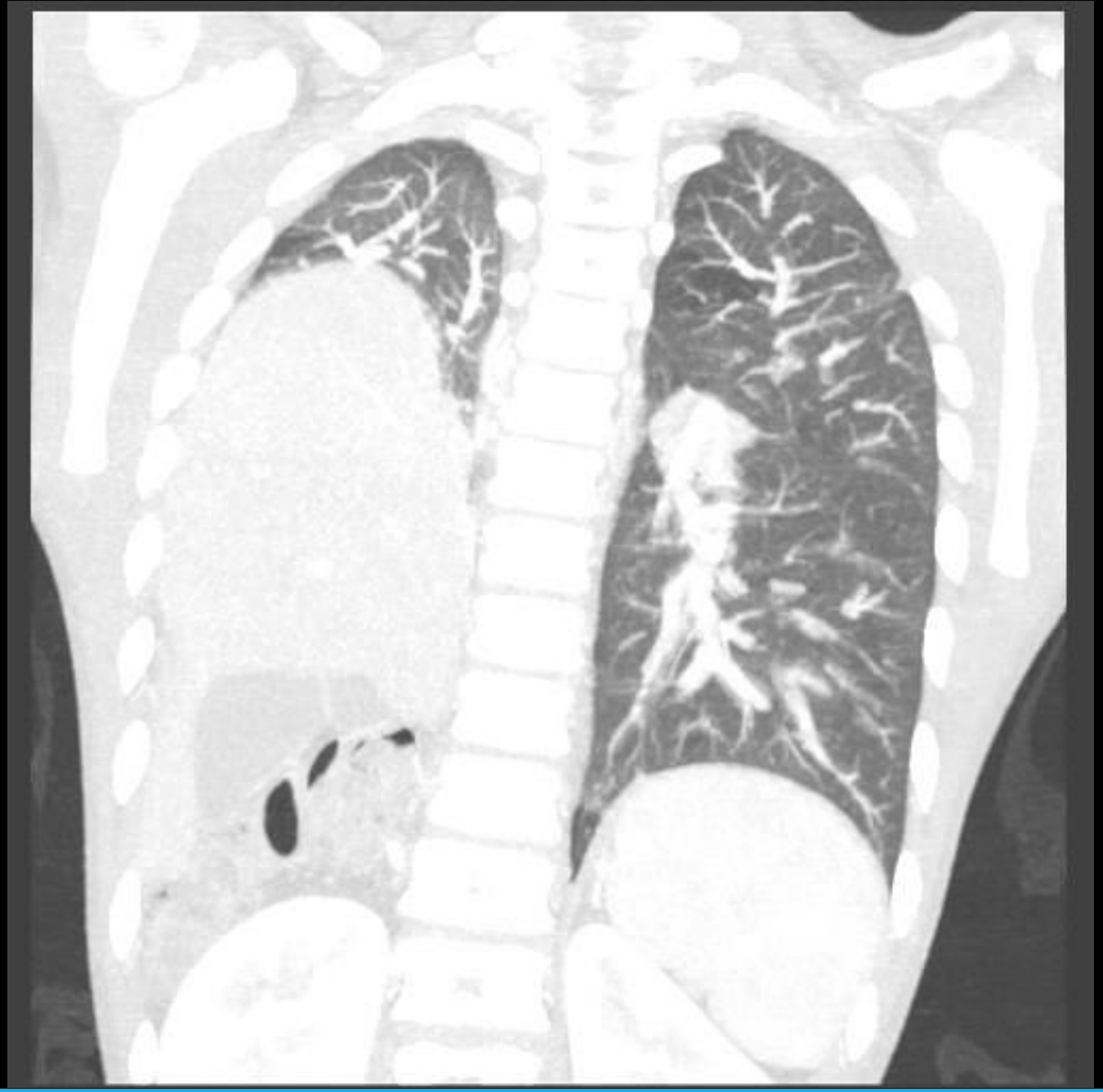
Top

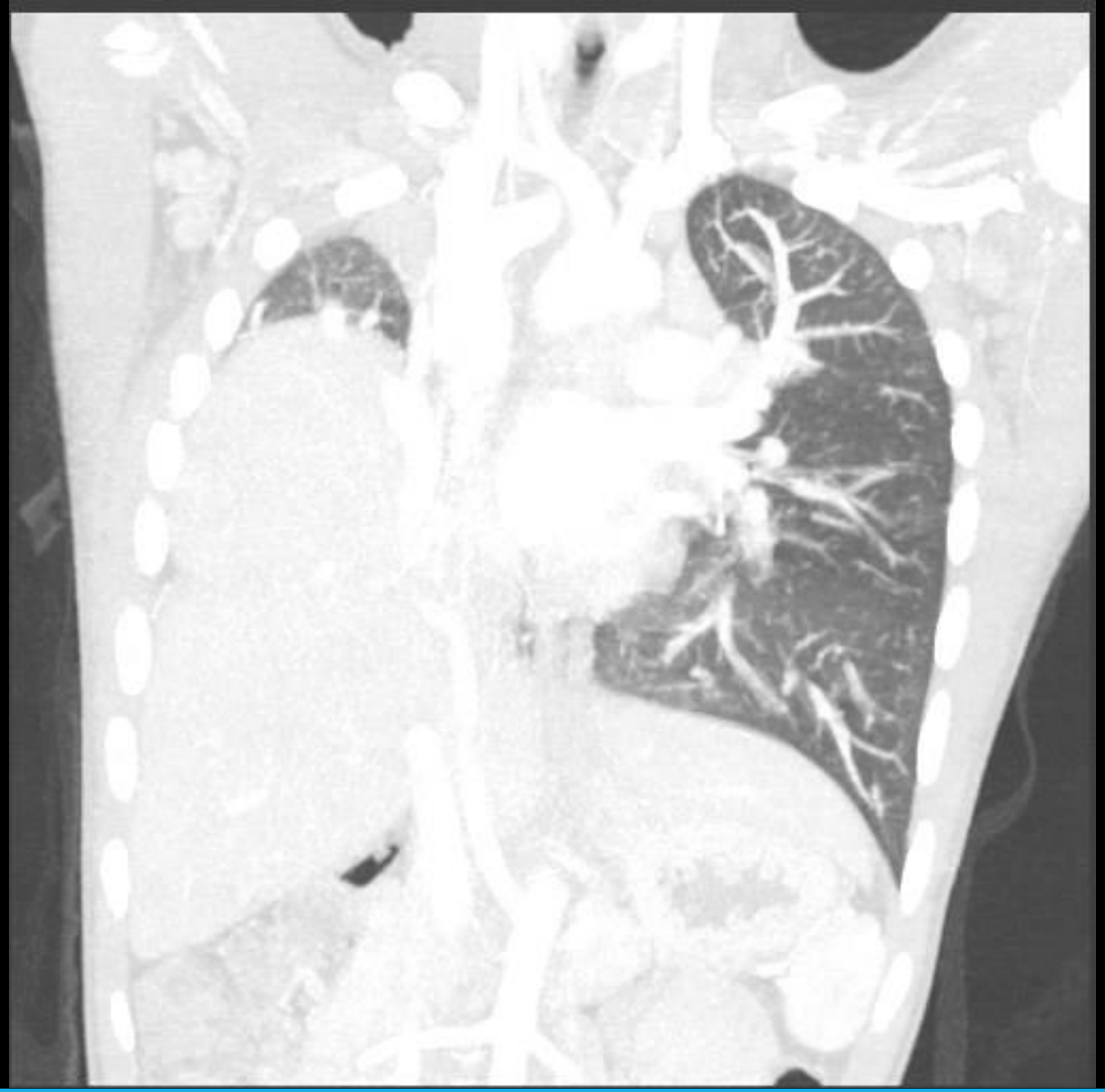












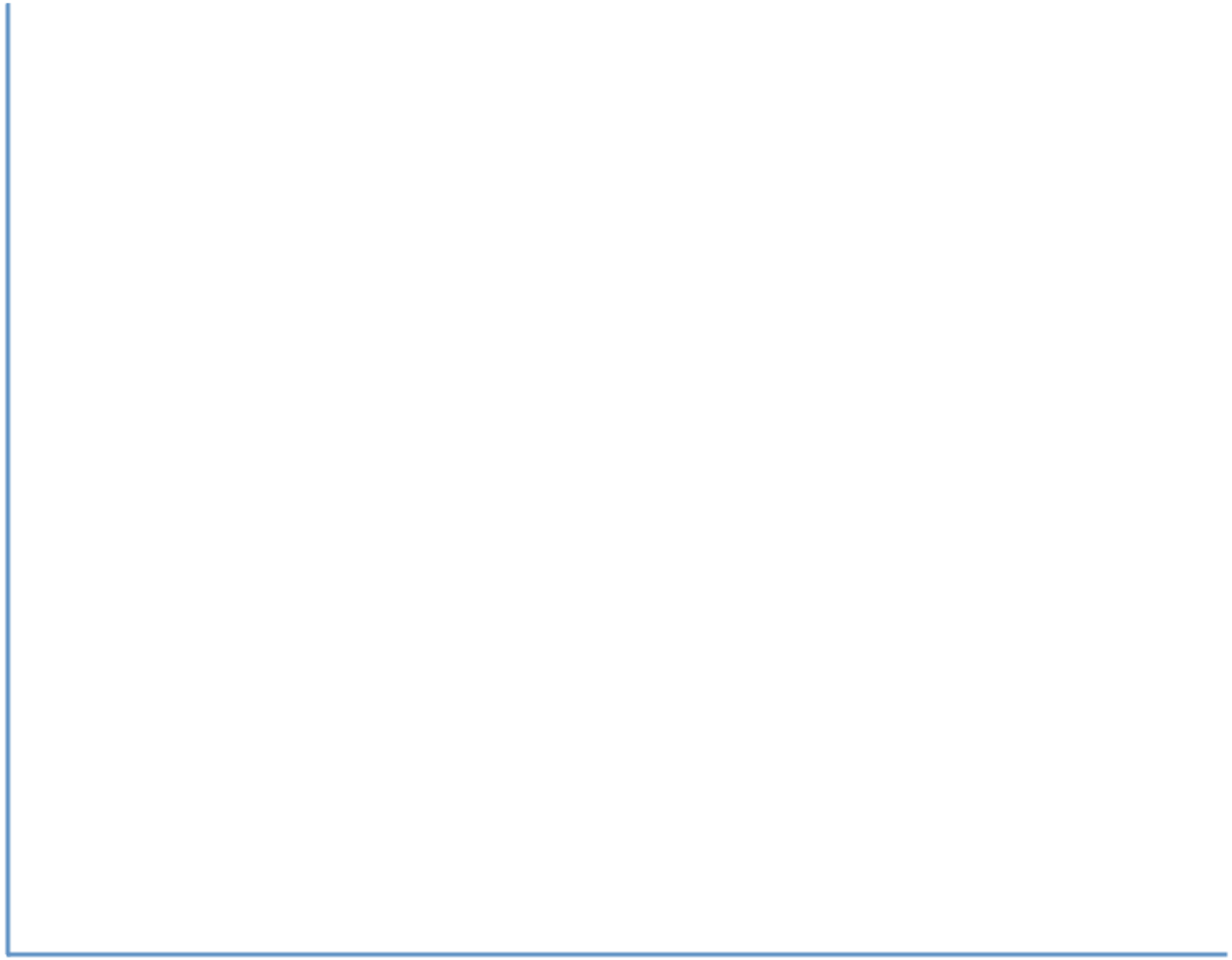
Which of the following is true:

A. Environmental factors do not play a role in the development of CDH

B. Most cases of CDH are detected prenatally

C. CDH infants should immediately be ventilated using a bag mask

D. Right-sided hernia is easily detectable on ultrasound



Congenital diaphragmatic hernia

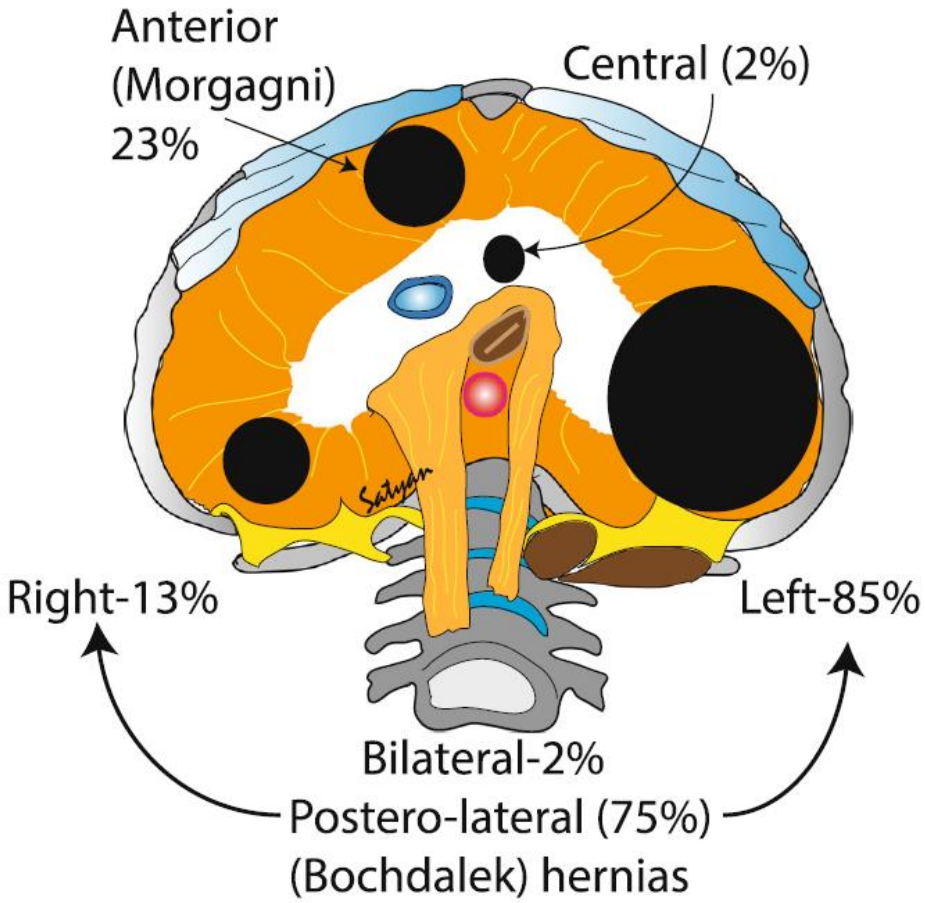
1 in 3300 live births

Genetic and environmental influences

- Vitamin A pathway
- Aneuploidy

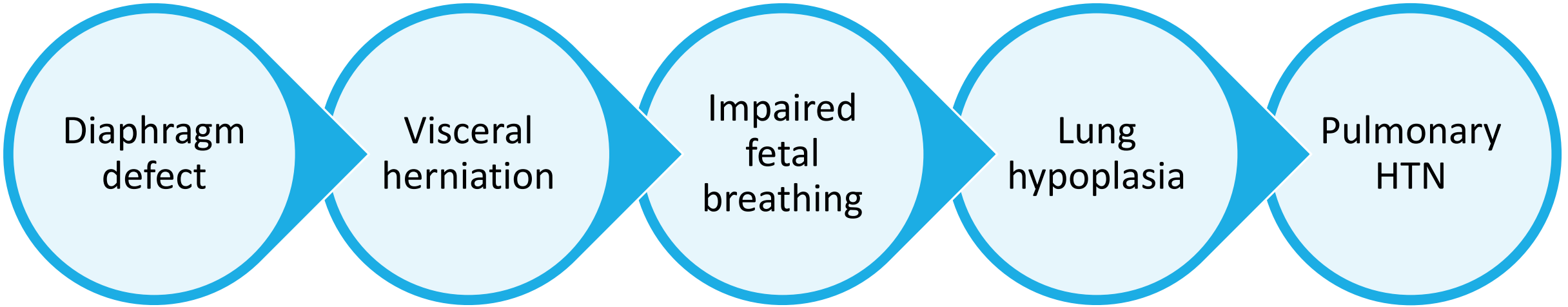
Associated defects

- Cardiac, GI, GU



Chandraskharan et al. Matern Heal Neonatol Perinatol 2017

Pathophysiology



Late congenital diaphragmatic hernia

5-25% of CDH

- 80% left;
- 20% right; 2/3 dx < 1 year

2 male : 1 female

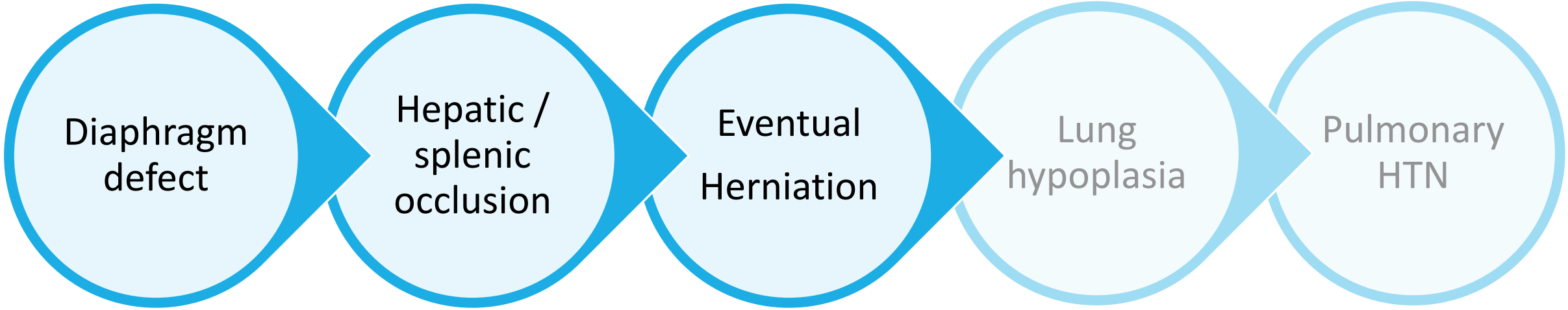
Few associated anomalies

- Malrotation in 29%

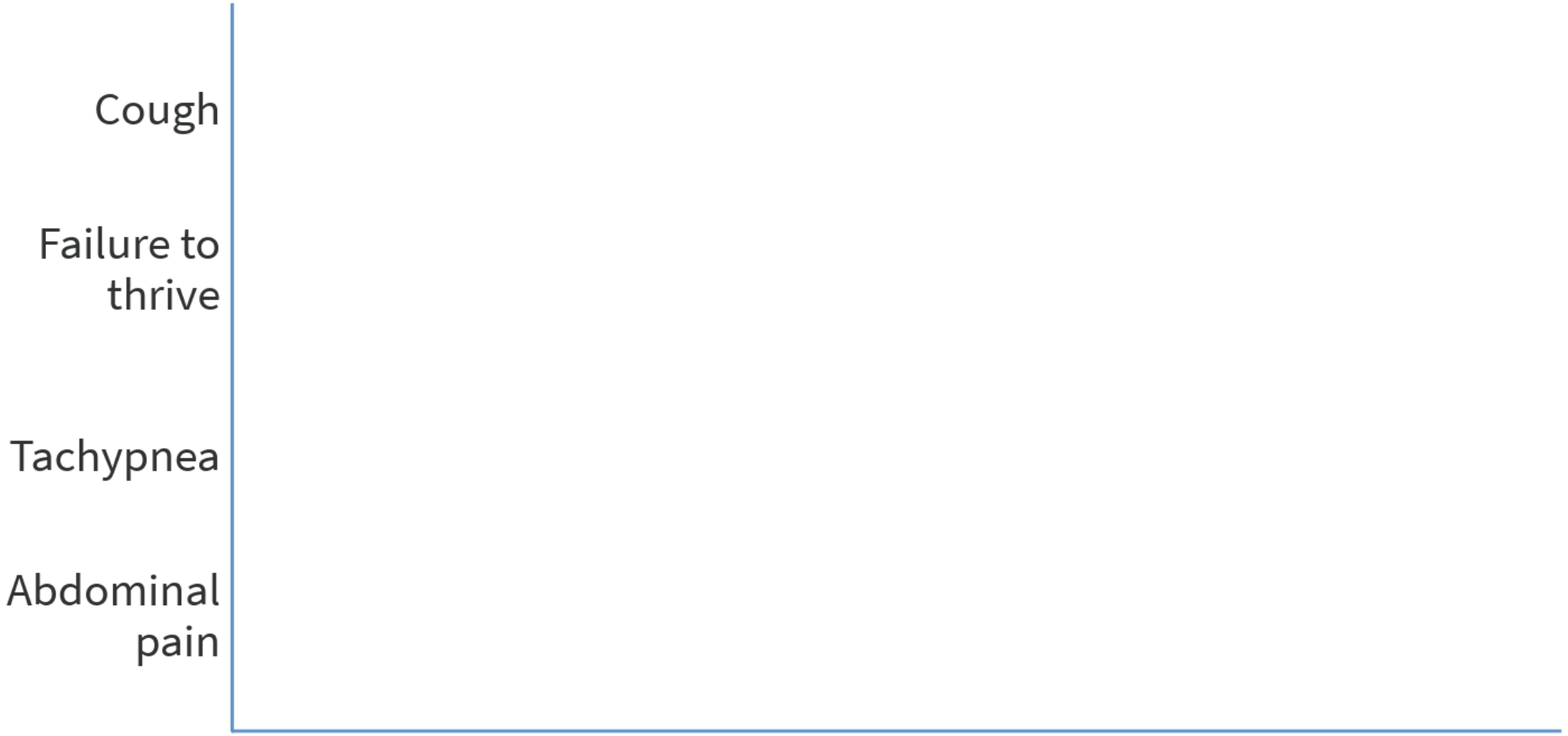
No apparent heritability

Congenital vs acquired debate is ongoing

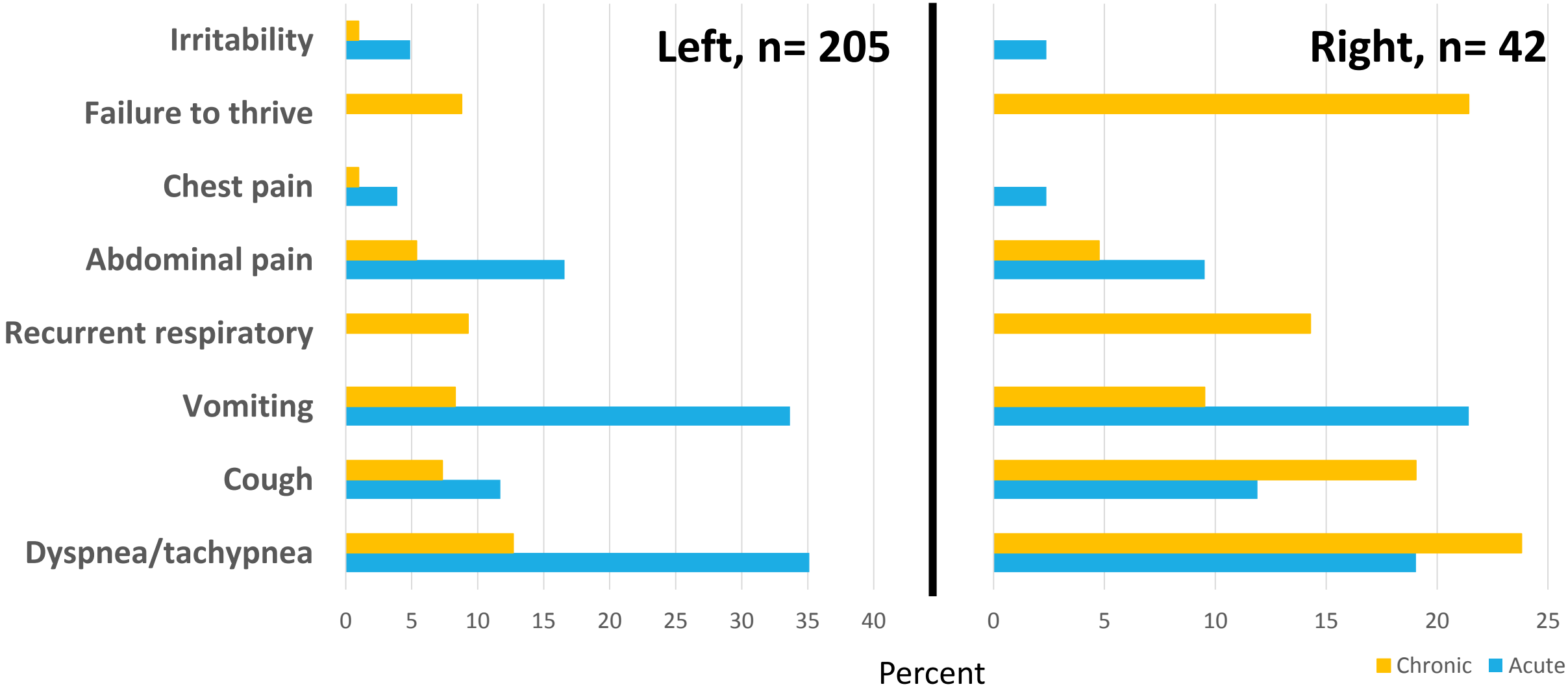
Pathophysiology



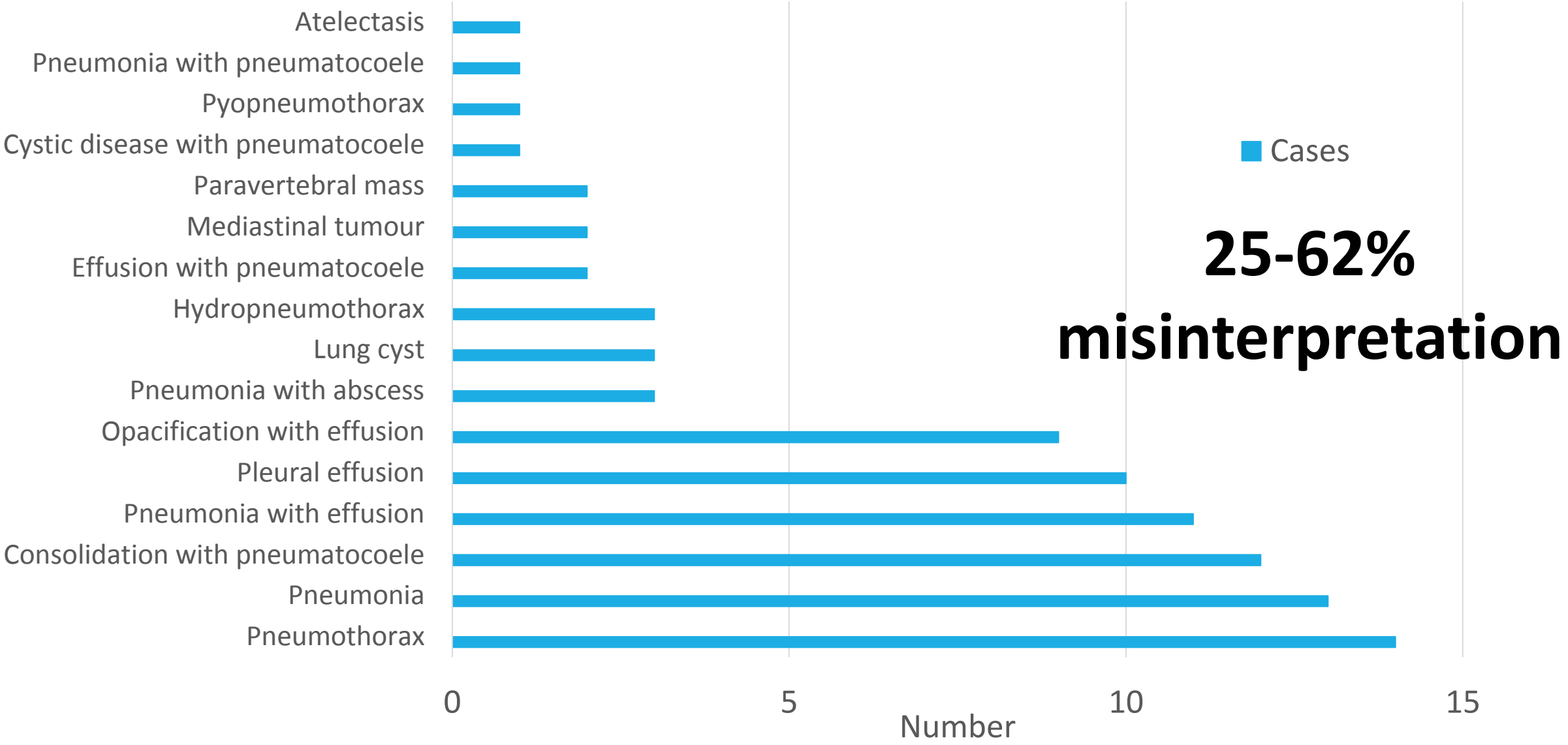
What is the most common presenting symptom in late CDH?



Presenting features of late CDH



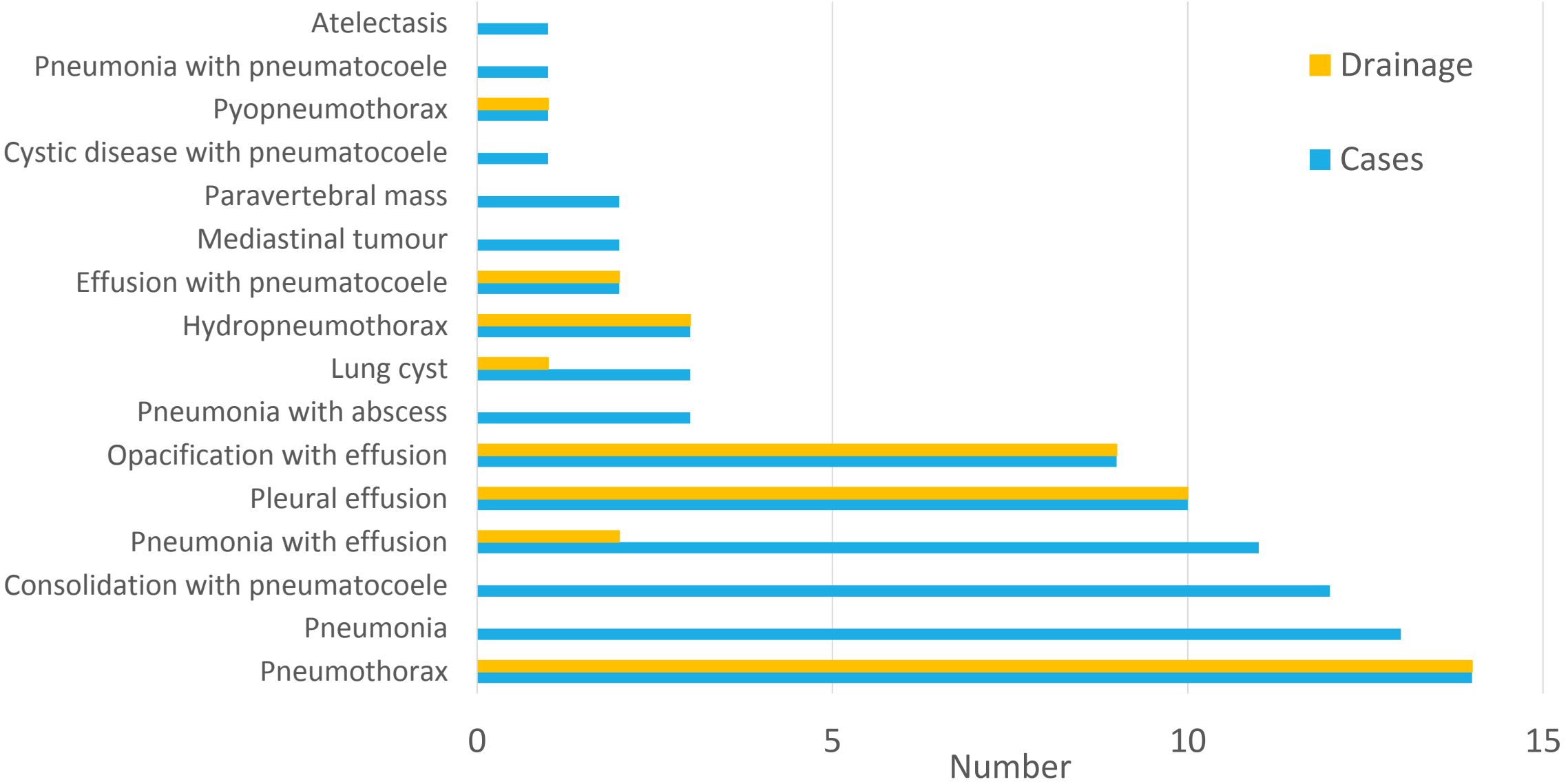
Initial radiographic diagnoses of late CDH



25-62% misinterpretation

Baglaj & Dorobisz Pediatr Radiol 2005

Initial radiographic diagnoses of late CDH



Baglaj & Dorobisz Pediatr Radiol 2005

Air-filled hollow viscera

Air fluid level

Soft tissue mass

Interruption of the diaphragm

Mediastinal shift

Management

Surgical correction generally completed

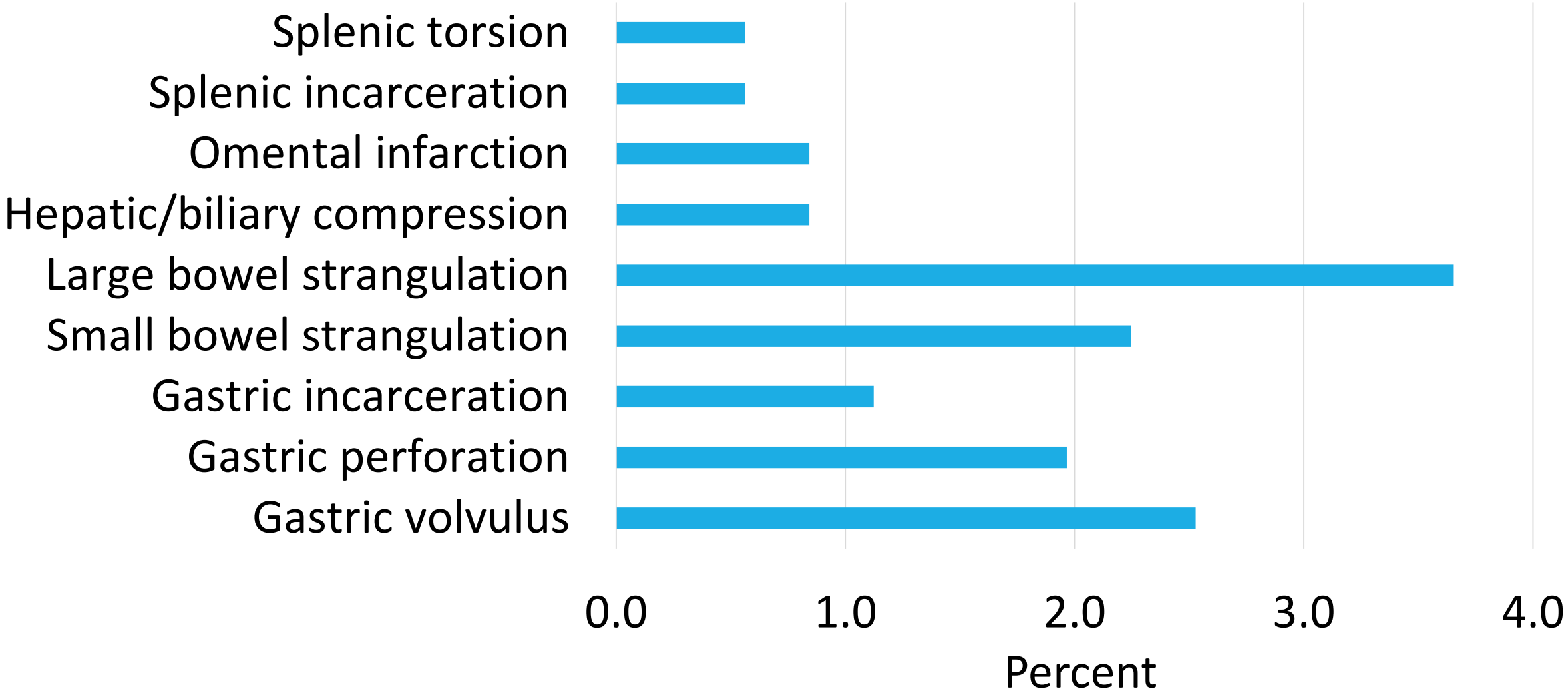
Disposition

No surgical repair
- Risks > benefits

Initiate QVAR

Follow-up
- CDH clinic
- Cardiology

Complications of Late CDH



Baglaj. Pediatr Surg Int 2004

GUIDELINE CPD

Diagnosis and management of congenital diaphragmatic hernia: a clinical practice guideline

The Canadian Congenital Diaphragmatic Hernia Collaborative*

American Academy of Pediatrics
DEDICATED TO THE HEALTH OF ALL CHILDREN™

CLINICAL REPORT

Postdischarge Follow-up of Infants With Congenital Diaphragmatic Hernia

Section on Surgery and the Committee on Fetus and Newborn

Organizational Principles to Guide and Define the Child Health Care System and/or Improve the Health of All Children

Be suspicious!

Key Points

Late CDH ≠ neonatal CDH

Misdiagnosis is common

- Heterogeneous radiographic and clinical presentation

Surveillance guidelines