Rare diseases in pulmonology

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Rare diseases:

- < 1/2000
- genetic, infectious, autoimmune, rare neoplasms, of unknown origin
- in most cases severe, chronic and progressing
- present after birth, during childhood and even adulthood
- problematic – diagnosis, profesional information and care

interstitial lung disease (ILD)

diffuse infiltrative lung disorders characterized by the presence of:

- inflammation
- altered lung interstitium

specific forms of ILD can be differentiated from one another by combining:

- clinical data
  radiologic imaging
- and pathologic findings (if lung biopsy is needed)

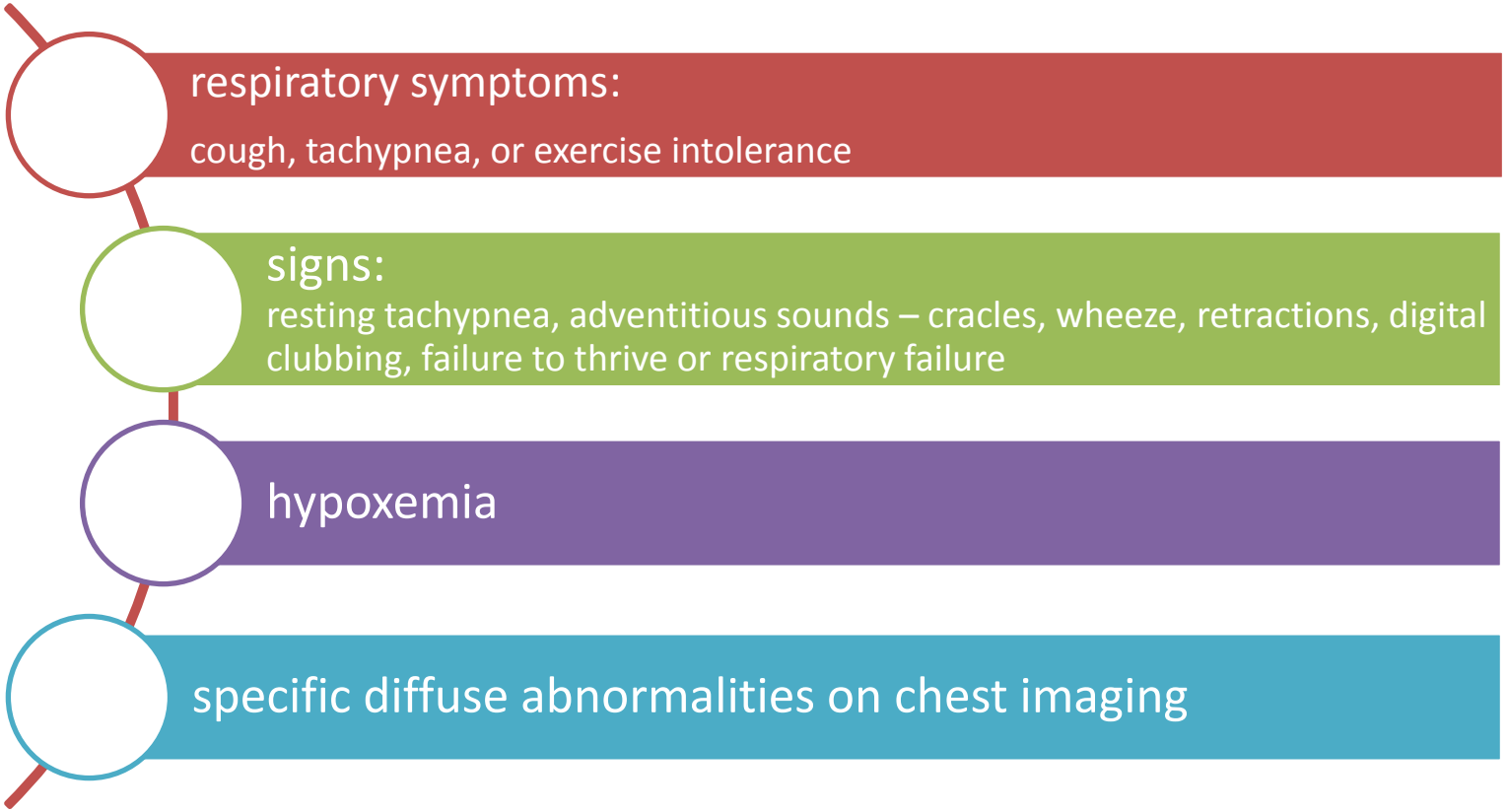
- well > 100 different forms

interstitial lung disease (ILD)

- restrictive
- impaired gas exchange
- abnormal imaging findings - diffuse changes

childhood interstitial lung disease (chILD)

at least three of the following four criteria:



interstitial lung disease (ILD)

Diagnosis:

- O2 saturation: while asleep, on exercise, at rest
- functional tests: spirometry, pletyzmography, DLCO
- thoracic imaging: X-ray, HRCT
- ECG, ECHO
- searching for the trigger: pH-metry, serological tests for connective tissue disease, infections, immunedeficiencies
- LUNG BIOPSY! the gold standard

interstitial lung disease (ILD)

CXR

- Hilar lymphadenopathy
- Septal thickening
- Honeycomb change
- Small nodules
- Cavitating nodules
- Migratory or fluctuating opacities
- Pneumothorax
- Pleural involvement
- Kerley B line prominence

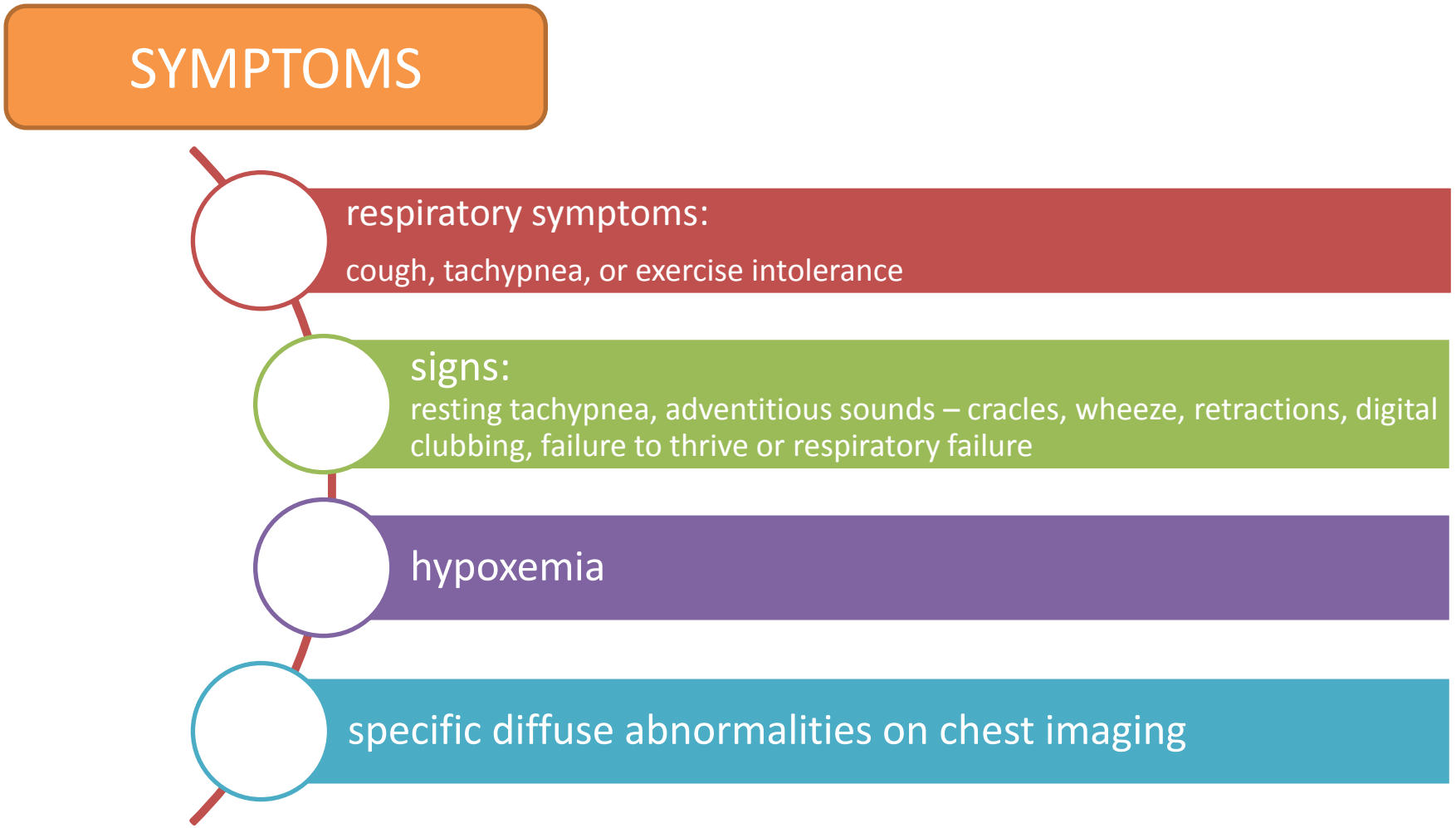
HRCT

- Nodules
- Septal thickening
- Cyst formation
- Reticular lines
- Traction bronchiectasis
- Honeycomb change
- Ground-glass opacity

neuroendocrine cell hyperplasia (NEHI)

- a prevalent sub-type of childhood interstitial lung disease (chILD) in infants and children younger than 2 years of age
- etiology unknown – genetic?
- previously described as transient tachypnoe of infancy

NEHI


SYMPTOMS
respiratory symptoms:
cough, tachypnea, or exercise intolerance
signs:
resting tachypnea, adventitious sounds – cracles, wheeze, retractions, digital clubbing, failure to thrive or respiratory failure
hypoxemia
specific diffuse abnormalities on chest imaging

NEHI

DIAGNOSIS

HRCT

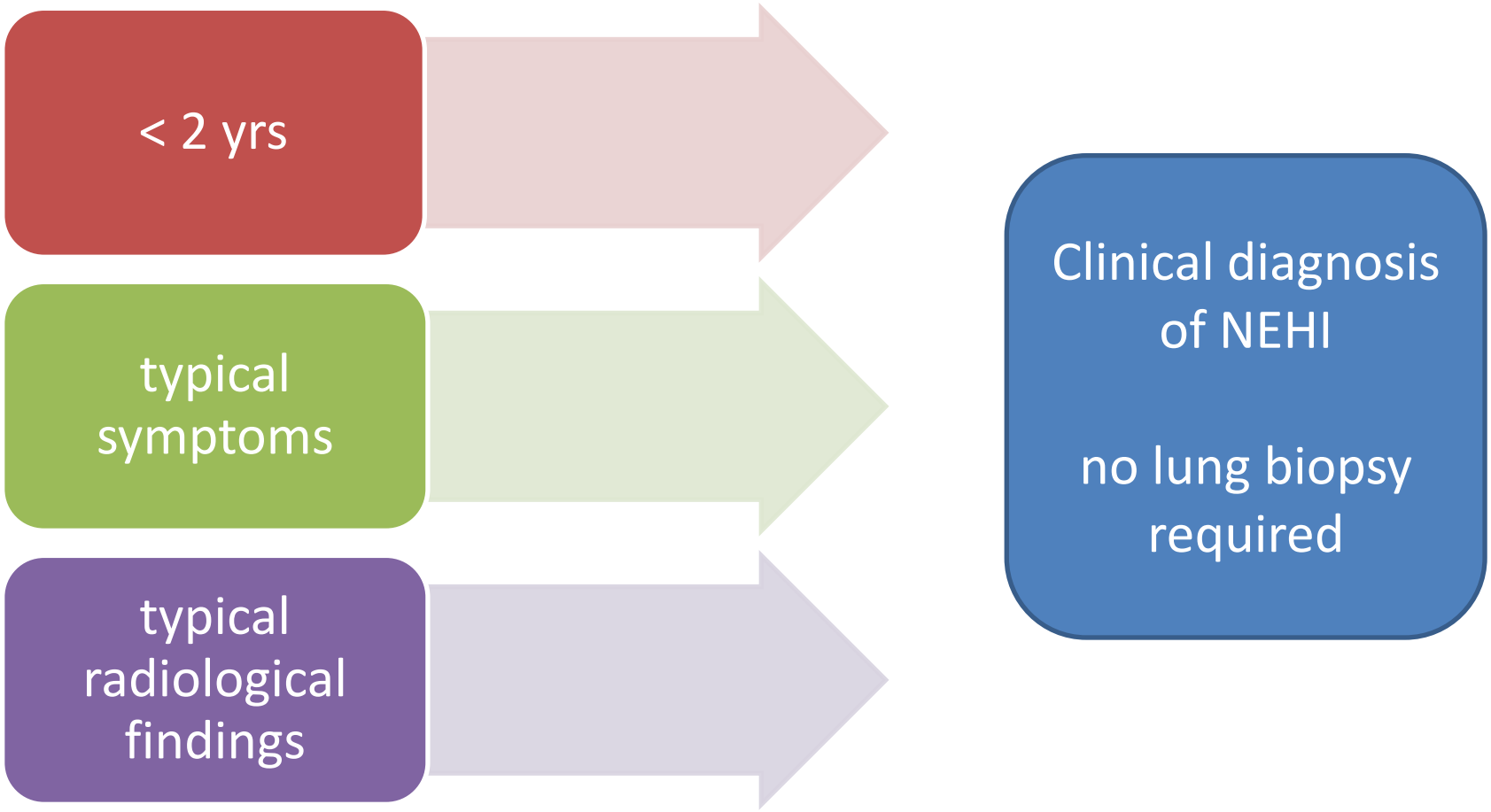
- ground-glass opacification in the right middle lobe and lingula
- air trapping with a mosaic pattern

LUNG BIOPSY

- Immunohistochemistry →

a significant number of bombesin-positive cells in the small airways walls

NEHI


< 2 yrs
typical symptoms
typical radiological findings
Clinical diagnosis of NEHI
no lung biopsy required

NEHI

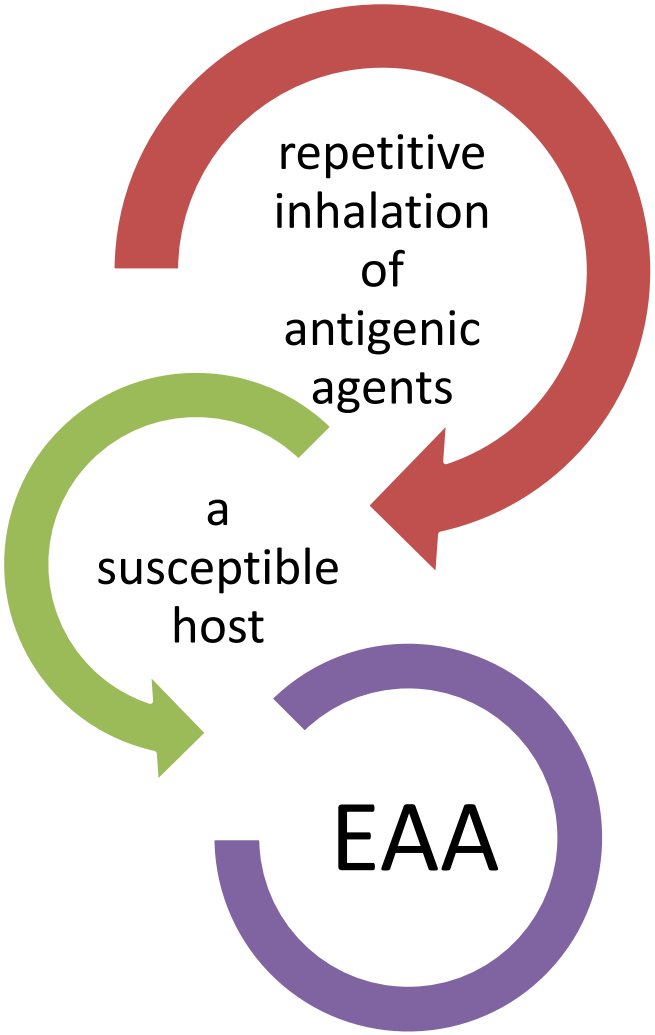
TREATMENT

Symptomatic:

- proper diet
- O2
- inhalant CSs
- bronchodilators in case of bronchoconstriction
- systemic CSs in severe cases

prognosis → self-limitting disease

Hypersensitivity Pneumonitis (Extrinsic Allergic Alveolitis, EAA)



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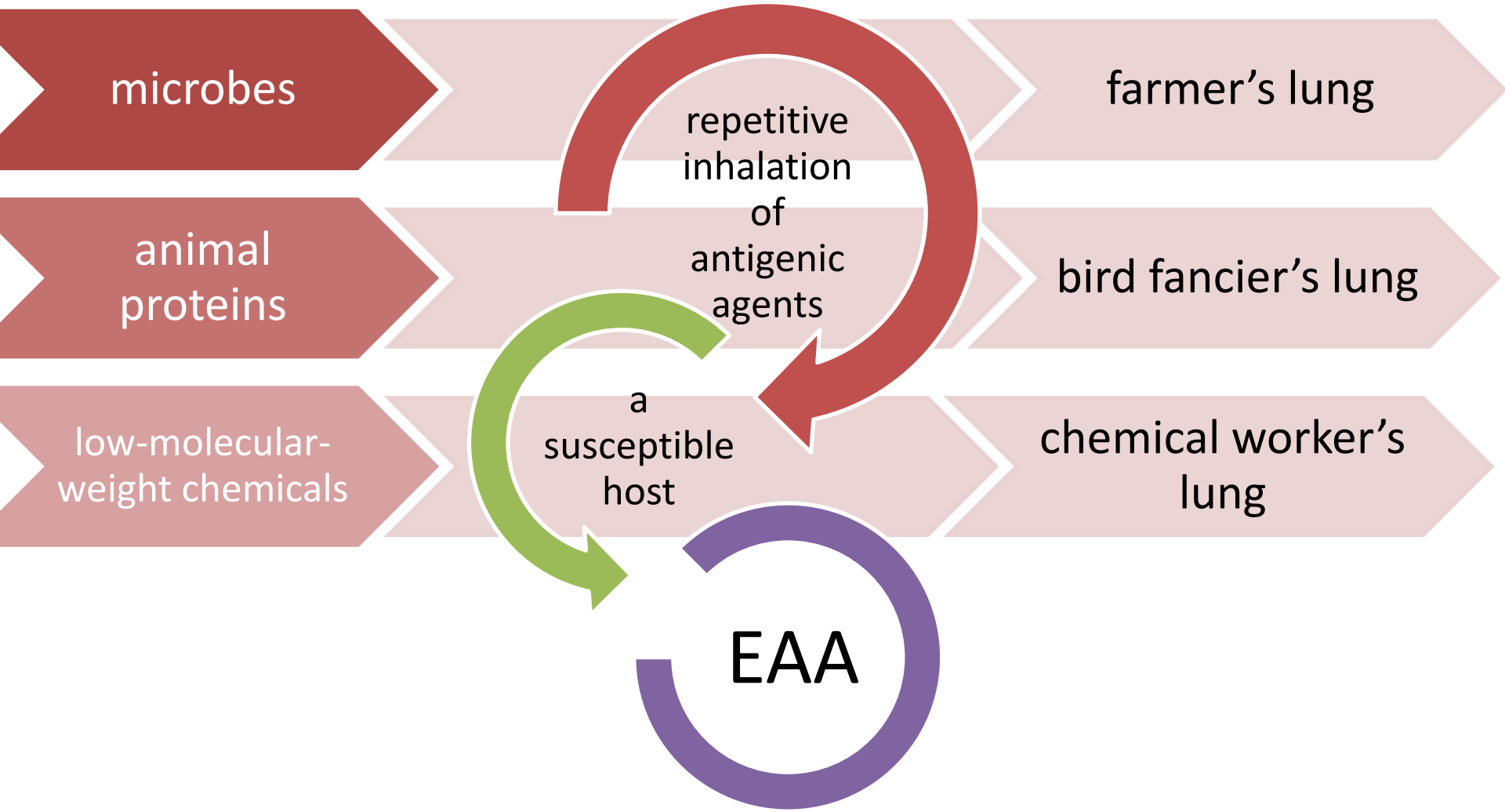
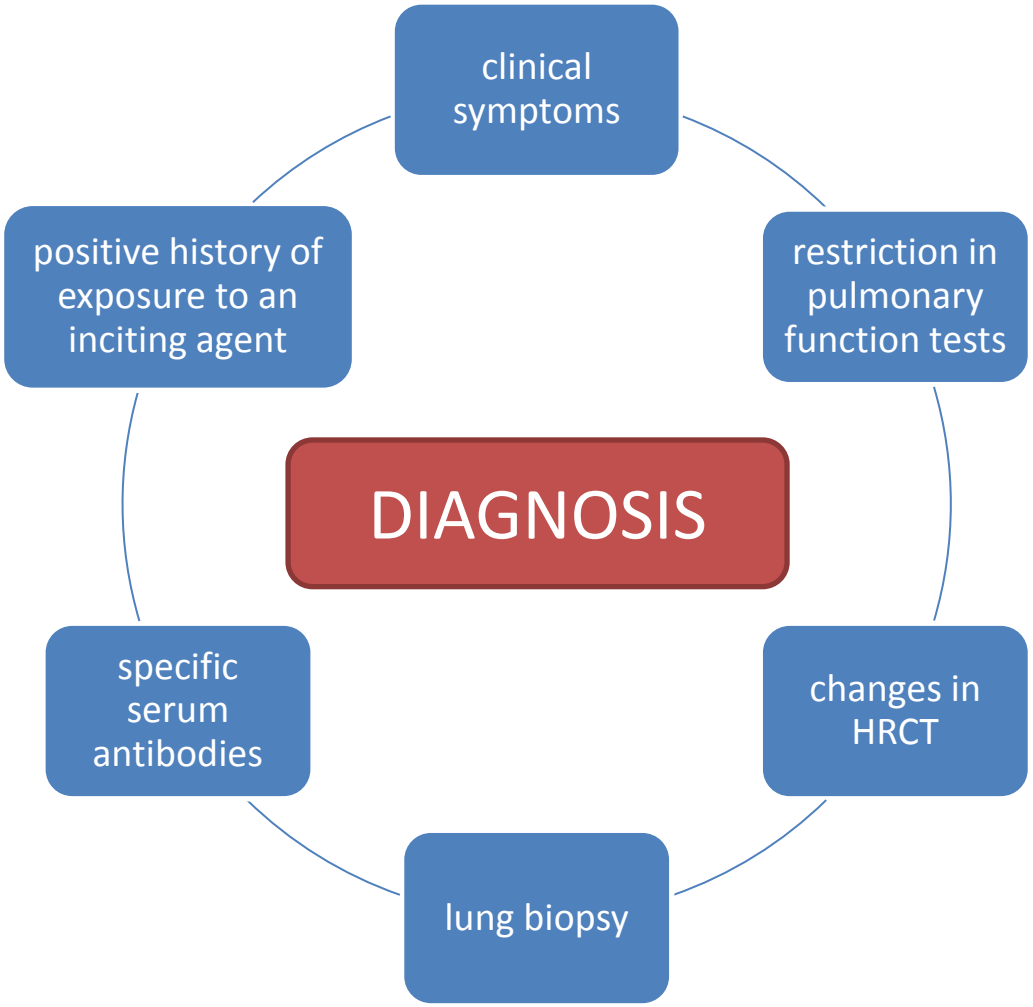


Hypersensitivity Pneumonitis (Extrinsic Allergic Alveolitis, EAA)

acute	• symptoms 2-9 h after massive exposure • cough, dyspnea, fever, chills, malaise, single cracles • symptoms cease within 24-48 h after exposure stops
subacute	• symptoms after several weeks of repetetive exposure • chronic (productive) cough, poor exercise tolerance, weight loss, poor apetite, cracles
chronic	• symptoms as in subacute course, worsening with time up to respiratory failure → finger clubbing • restriction in pulmonary function tests • fibrosis in HRCT - irreversible

EAA


clinical symptoms
restriction in pulmonary function tests
changes in HRCT
lung biopsy
specific serum antibodies
positive history of exposure to an inciting agent
DIAGNOSIS

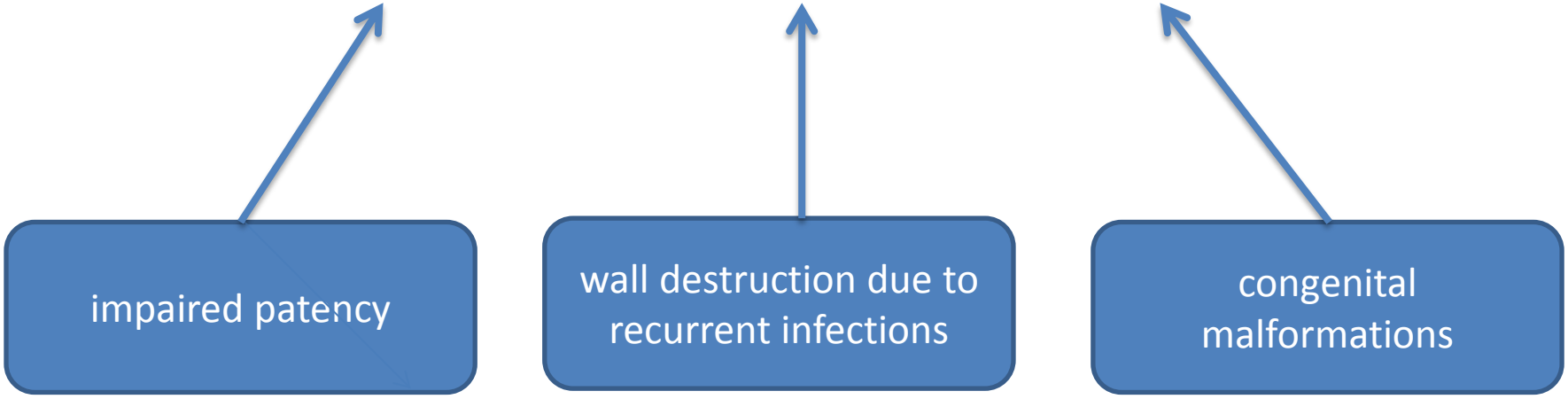
EAA

TREATMENT

- avoidance of exposure to an inciting agent
- CSs
- lung Tx, lung-heart Tx

Bronchiectasis:

abnormal, permanent, and irreversible distortion of one or more of the conducting bronchi or airways

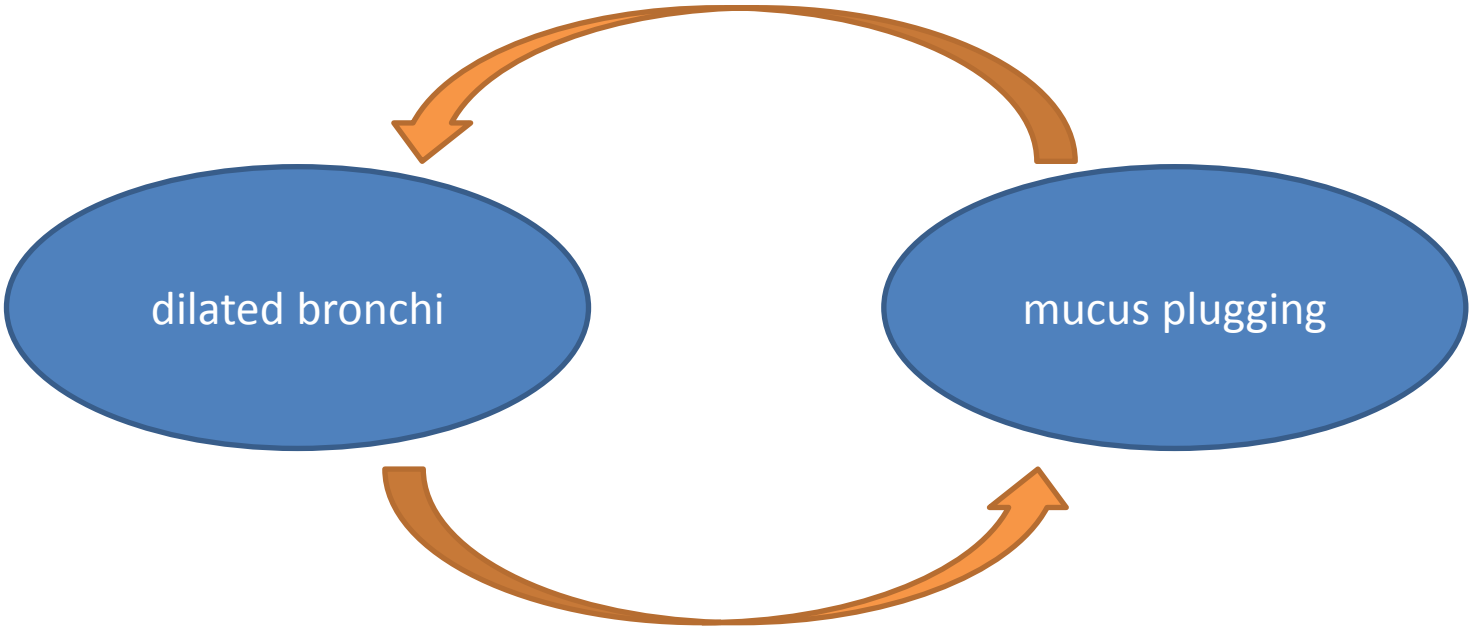


foreign body aspiration

CF
PCD
GERD
immunodeficiency

connective tissue
cartilage

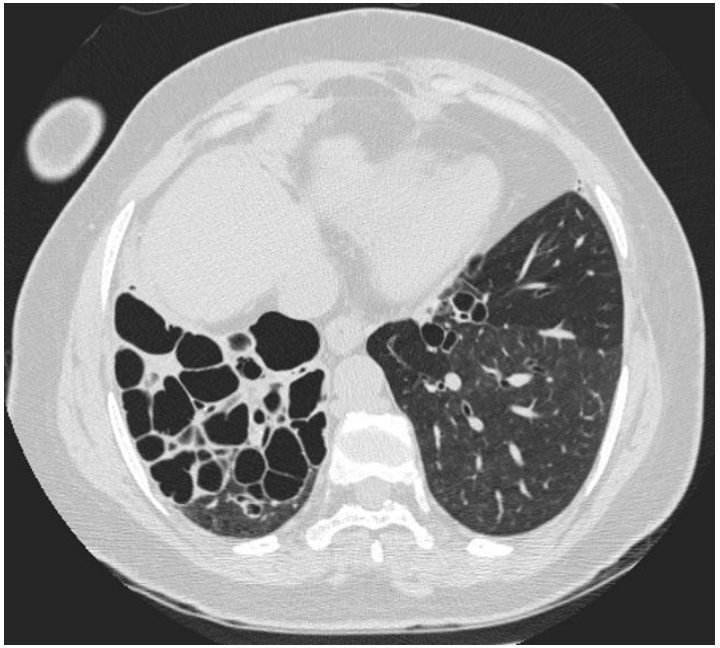
Bronchiectasis:



- productive cough
- dyspnea
- recurrent infections (pneumonias in the same localisation)
- in severe cases - wasting and weight loss

Bronchiectasis:

localised cracles, sometimes ronchi and wheeze



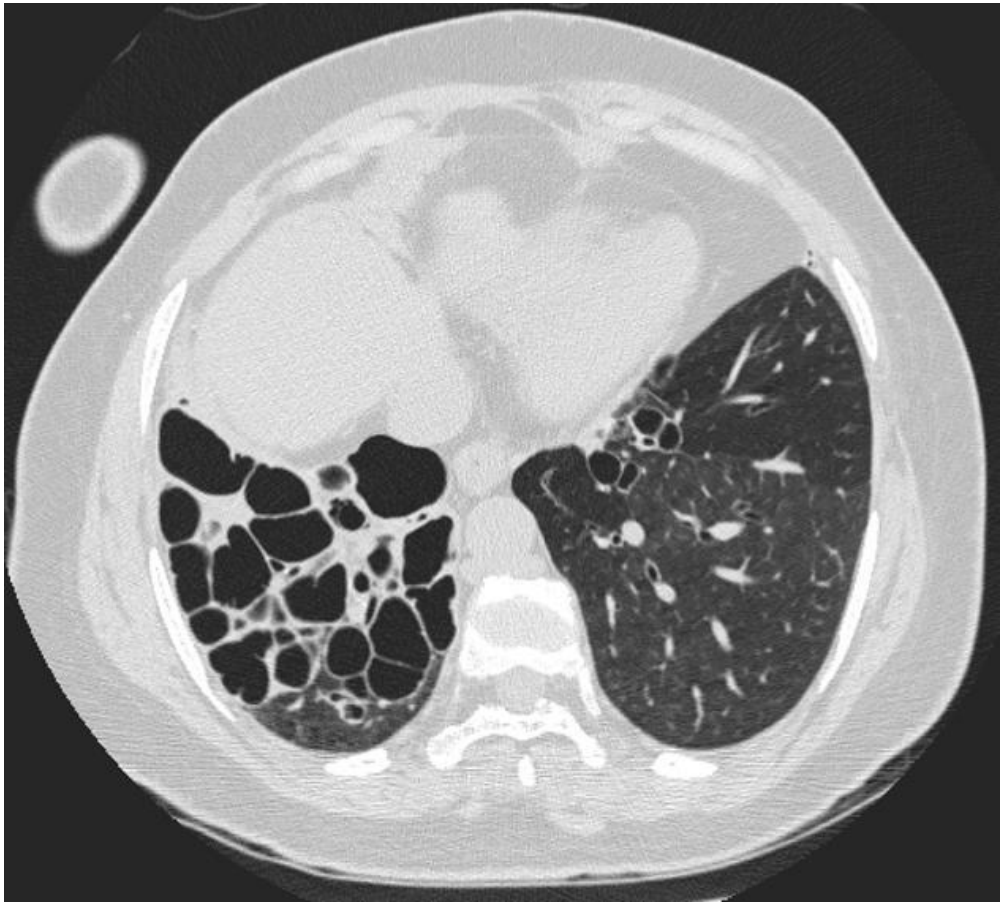
HRCT

bronchoscopy – diagnostic + therapeutic

searching for the trigger

Case courtesy of Dr Hani Salam, Radiopaedia.org, rID: 13104

Bronchiectasis:



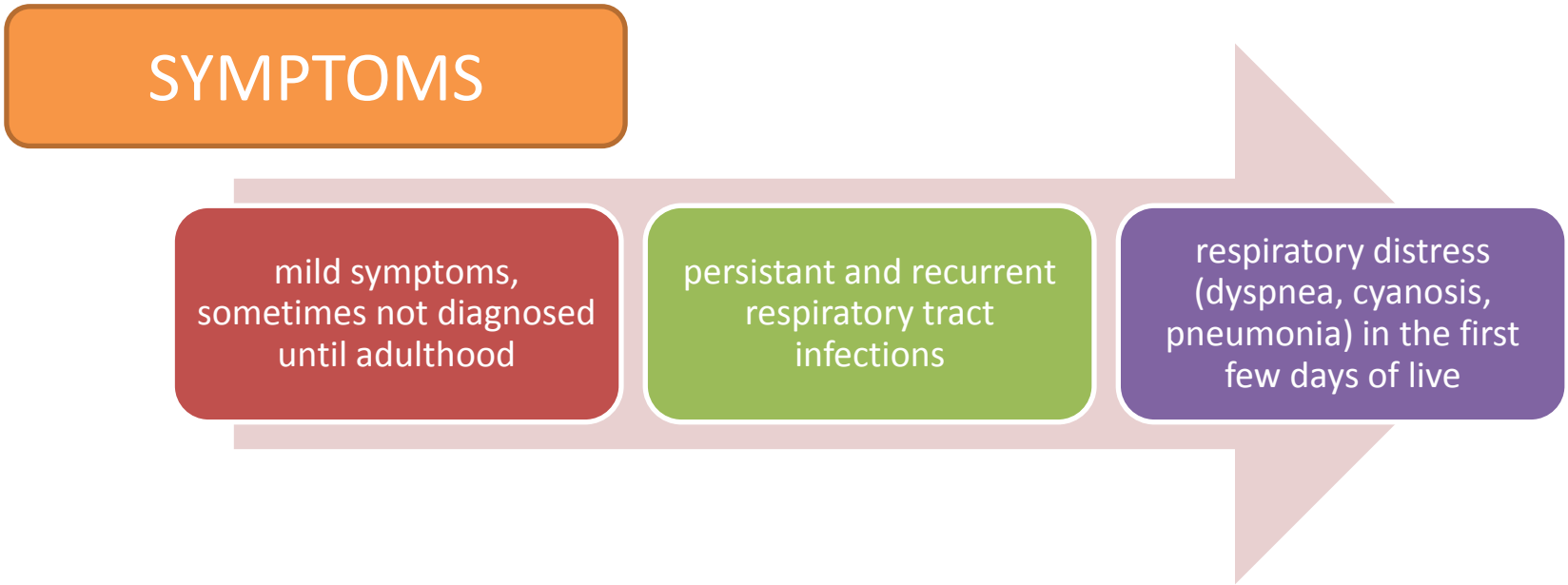
1. profilaxis
   - immunizations
   - antibiotics
2. efficient treatment of infections
3. bronchodilators
4. **evacuation of mucus**
   - mucolitics
   - clearance physiotherapy
5. surgery

Congenital anomalies

- <4% of total congenital defects in children
- Congenittal anomalies are responsible for 1/3 of all infant deaths

1. cardiovascular system
2. respiratory system

(due to ↑ selective termination of pregnancies with diagnosed nervous system malformations)



Congenital anomalies

- airway
- lung parenchyma
- pulmonary vessels
- diaphragm
- chest wall
- neuro-muscular

Tracheo-esophageal fistula (TEF)

- defective septation of the primitive foregut = incomplete separation of respiratory and gastrointestinal tract during embionic stage

- incidence 1:3000-5000 live births

- in 85-90% TEF is accompanied by the esophageal atresia

➢ The lung appears around day 26 as a ventral bud of the esophagus

Tracheo-esophageal fistula (TEF)

Typa A 85% Type B 6-8% H-type 3-5%

Tracheo-esophageal fistula (TEF)

SYMPTOMS

TEF with the esophageal atresia

- maternal polyhydramnios
- Neonates : copious, fine white frothy bubbles of mucus in the mouth and nose, dyspnea, tachypnoe, cyanosis, aspiration → pneumonia. Secretions recur despite suctioning.
- Infants : wheezing and episodes of coughing and choking in association with cyanosis.
- Symptoms worsen rapidly during feeding

TEF with the esophageal atresia

H-type TEF

- 3-5%
- mild symptoms, recurent RTI (pneumonia), persistant cough and cough after feeding

Tracheo-esophageal fistula (TEF)

DIAGNOSIS?

- polyhydramnios

- tracheal compression and deviation on chest Xray
- Absence of a gastric bubble
- Aspiration pneumonia
- Contrast studies

- bronchoscopy

Tracheo-esophageal fistula (TEF)

TREATMENT

surgery → complications → anastomotic stenosis
recurrence of the fistula
gastroesophageal reflux ~ 100%

Bronchogenic cysts

- abnormal fetal development of bronchial tree – a fragment of bronchial tissue separates from developing bronchus to form the bronchial cyst
- app. 5% of mediastinal masses in infants and children
- solitary, unilocular cystic structures containing fluid or mucus, 1-3cm ∅ (up to 10cm)
- thin walled, lined with ciliated respiratory epithelium and mucus glands, surrounded by smooth muscle and fibrous tissue, cartilage presence allows differentiation from esophagal duplication

SYMPTOMS	**compression:** cough, wheezing, dyspnea, athelectasis, emphysema, recurrent infections, dysphagia **infection of the cyst:** exacerbation of compressive symptoms, hemoptysis, lung abscess, pneumothorax when ruptured **no symptoms**
DIAGNOSIS	diminished/absent vesicular sound; chest X-ray, CT, MRI
TREATMENT	surgical excision

Pulmonary sequastration

- an abnormal accesory tracheobronchial bud arising from the foregut
- typically without a connection to the normal tracheobronchial tree and supplied by an anomalous systemic artery
- **intralobar sequestration (ILS)** – more common, visceral pleura shared with the adjacent normal lobe, most commonly in the lower lobes, venous drainage usually through the inferior pulmonary vein; may be connected to the normal tracheobronchial tree → recurrent RTIs, repeated episodes of pneumonia in the same localization
- **extralobular sequestration (ELS)** – separate visceral pleura, posteromedially in the left lower chest, venous drainage through the systemic circulation; early presentation – 60% diagnosed < 6mo, respiratory distress, feeding problems, tachypnoe
- solid, nonaerated, pyramid-shaped/ovoid mass: chest X-ray, CT, MRI
- differential diagnosis: foreign body aspiration, TEF, bronchial anomaly, neuroblastoma!
- Treatment: symptomatic – surgery, asymptomatic – consider surgery

Congenital lobar emphysema (CLE)

- a partial bronchial obstruction creating a ball-valve effect → abnormal hyperinflation of one or two lobes, most often the left upper lobe
- obstruction may be intrinsic (bronchomalacia, cartilaginous rings, absence of cartilage) or less commonly extrinsic (vascular, bronchogenic cyst)
- rarely detected prenatally, typically presents in neonates and infants when air gets into the lung
- progressive tachypnoe with lobar hyperinflation, mediastinal shift, and sometimes contralateral atelectasis; hyperresonance on percussion
- diagnosis: clinical presentation, phisical examination, chest X-ray
- Differential diagnosis: foreign body aspiration, tension pneumothorax
- Treatment: surgery - lobectomy

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- Differential diagnosis: foreign body aspiration, tension pneumothorax

- Treatment: surgery - lobectomy

Congenital Cystic Adenomatoid Malformation (CCAM)

- Glandular hypertrophy of the lung tissue with formation of multiple cysts
- Symptoms: respiratory distress
- Diagnosis: in utero – ultrasound, chest x-ray, CT, hist-pat
- Treatment: surgical resection

resection of even asymptomatic masses is recommended because of the risk for infection, hemorrhage, acute respiratory compromise (which may occur anytime), and neoplastic transformation.

Congenital Diaphragmatic Hernia (CDH)

- developmental abnormality of the diaphragm allowing the abdominal viscera to enter the thoracic cavity; 6-8 Hbd
- > 95% occur through a posterolateral defect (Bochdalek hernia)
  - 90% on the left side
- < 5% located anteriorly (Morgani hernia)

- diagnosis:

  prenatal ultrasound (16 - 24 wk) diagnosed in over 50% of cases

  polyhydramnios, chest mass, mediastinal shift, gastric bubble, fetal hydrops, liver in the thoracic cavity and lung-to-head ratio

  (last 2 are predictors of postnatal morbidity)

  postnatal: X-ray

Congenital Diaphragmatic Hernia (CDH)

Bochdalek hernia (>95%)

- posterolateral defect (90% on the left side)
- immediate postnatal period: respiratory distress, a scaphoid abdomen, apparent dextrocardia (90% CDH on the left) and decreased breath sounds over the involved chest
- varies according to the severity of **pulmonary hypoplasia** and **pulmonary hypertension**
- EMERGENCY !!!!!

High mortality if not diagnosed in the first postnatal 24 hours.

Treatment:

- avoidance of bag-and-mask ventilation
- prompt endotracheal intubation with limited ventilation pressures
- surgical correction

Diagnosis:

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- polyhydramnios,
- chest mass,
- mediastinal shift,
- gastric bubble,
- fetal hydrops,
- liver in the thoracic cavity
- lung-to-head ratio

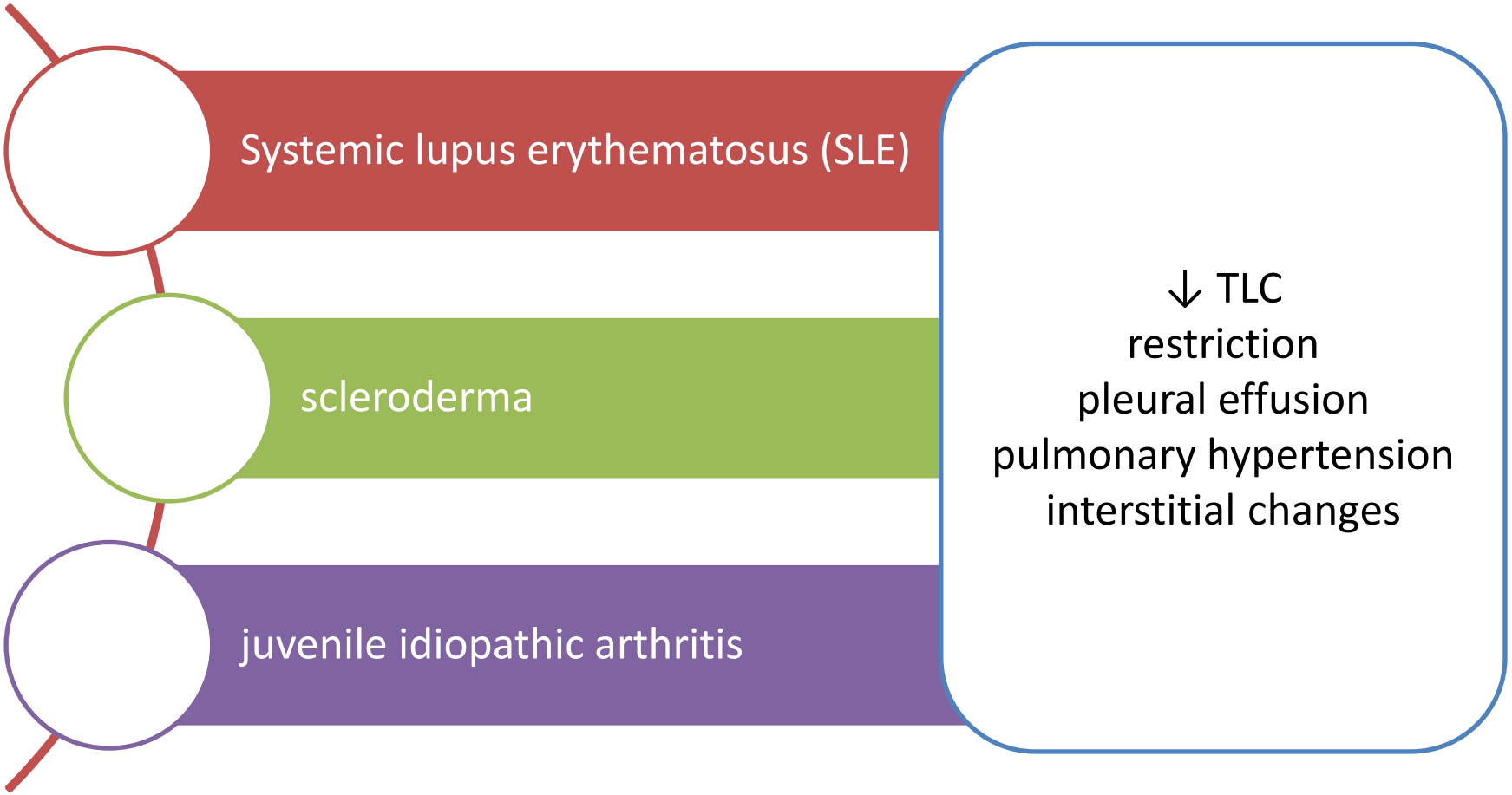
postnatal: X-ray

Congenital Diaphragmatic Hernia (CDH)

Morgani hernia <5%

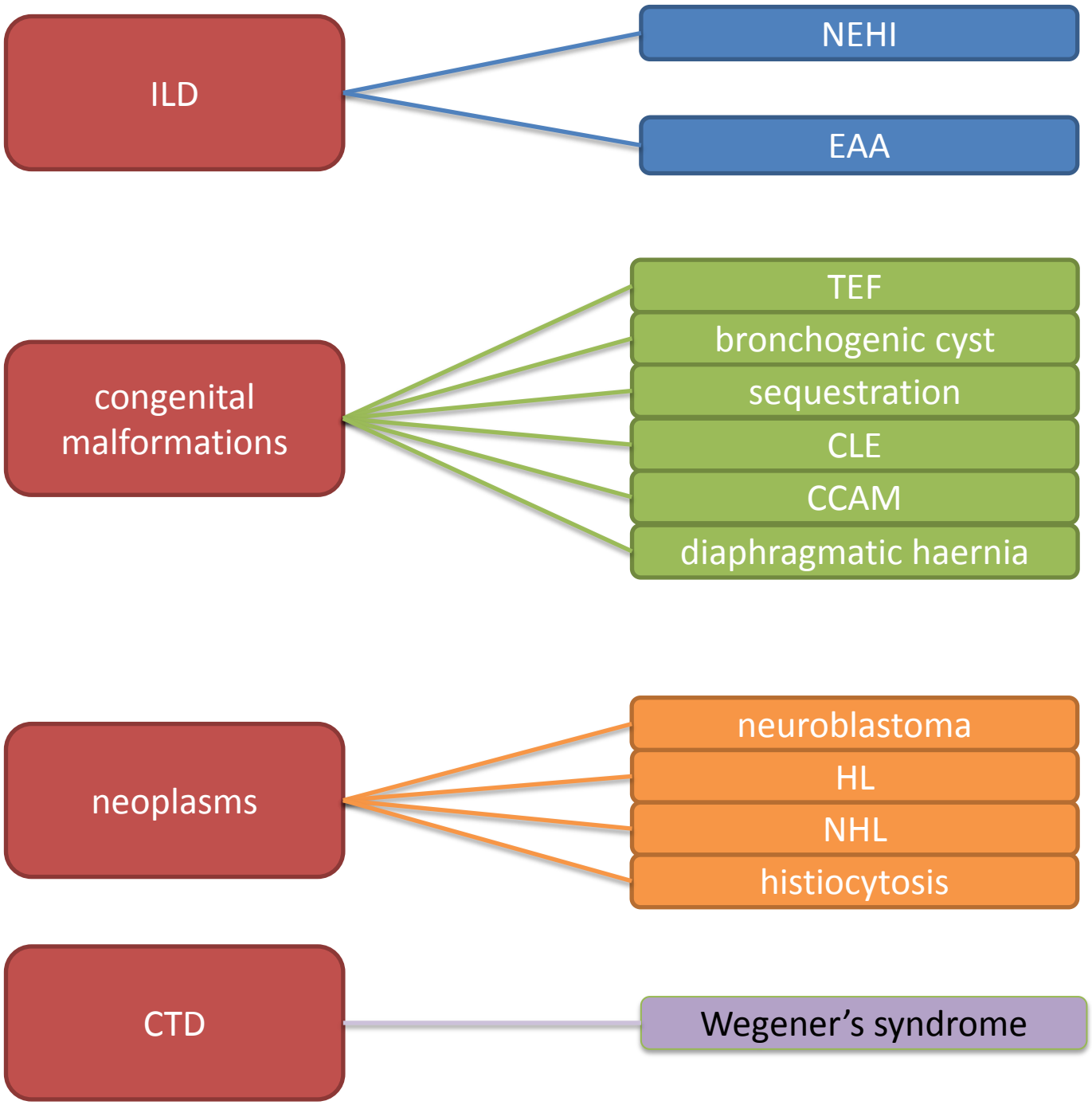
- more common in girls rarely symptomatic at birth
- found incidentally on chest roentgenograms
- usually rightsided – more than ½ have a sac containing omentum or transverse colon
- rarely leftsided– obliterated by pericarduim
- late symptoms suggesting gallbladder problems or peptic ulcer disesase, retroxiphoid pain, dyspnea and cough
- 10% of complications – possibility of incarceration or strangulation

Connective tissue diseases



Vasculitis

- granulomatosis with polyangiitis (Wegener Granulomatosis)
- microscopic polyangiitis


ILD
NEHI
EAA
congenital malformations
TEF
bronchogenic cyst
sequestration
CLE
CCAM
diaphragmatic haernia
neoplasms
neuroblastoma
HL
NHL
histiocytosis
CTD
Wegener's syndrome

Thank you!