Cystic fibrosis and PCD in children

Cystic fibrosis - introduction

- The most common life-shortening autosomal recessive disease among Caucasian populations
- **Frequency** – 1 in 2000-3000 live births
- Median predicted survival for CF patients is around **35** years…

Cystic fibrosis - gene

- Mutation in CFTR protein
  - a complex chloride channel
  - and
  - regulatory protein
  - found in all exocrine tissues

- The gene that causes CF is located on chromosome 7.
- There are over 1500 mutations, and the most common mutation is delta F508 del.

Cystic fibrosis – clinical features

- RESPIRATORY TRACT INVOLVEMENT
- **repeated infections,**
- **persistent, productive cough**
- sputum production, morning sputum
- decreased ability to exercise
- *Staphylococcus aureus, Haemophilus influenzae, and Pseudomonas aeruginosa are* the three most common organisms causing lung infections in CF patients
- inflammation eventually leads to bronchiectasis

Cystic Fibrosis - clinical features

HRCT is the „gold standard" for detecting bronchiectasis.

Classically, cylindrical bronchiectasis may appear as "signet rings"

Bronchiectasis

Chronic cough with massive sputum production

Persistens abnormalites on physical examination of the chest.

Persistent abnormalities on chest x - ray

Bronchiectasis

- Types of Bronchiectasis
  - **cylindrical** (the most common)
  - varicose
  - saccular

Cystic Fibrosis – clinical features

- SINUS DISEASE

- chronic rhinosinusitis,
- nasal polyposis,
- chronic nasal congestion, headeaches,
- post nasal drip and cough,

- 100% CF patients have panopacification of the paranasal sinuses on radiographs

Cystic Fibrosis – clinical features

- PANCREATIC DISEASE
- Pancreatic insufficiency
- Fat malabsorption
- Steatorrhea (frequent, bulky, foul-smelling stools that may be oily),
- Poor weight gain
- CF-related diabetes

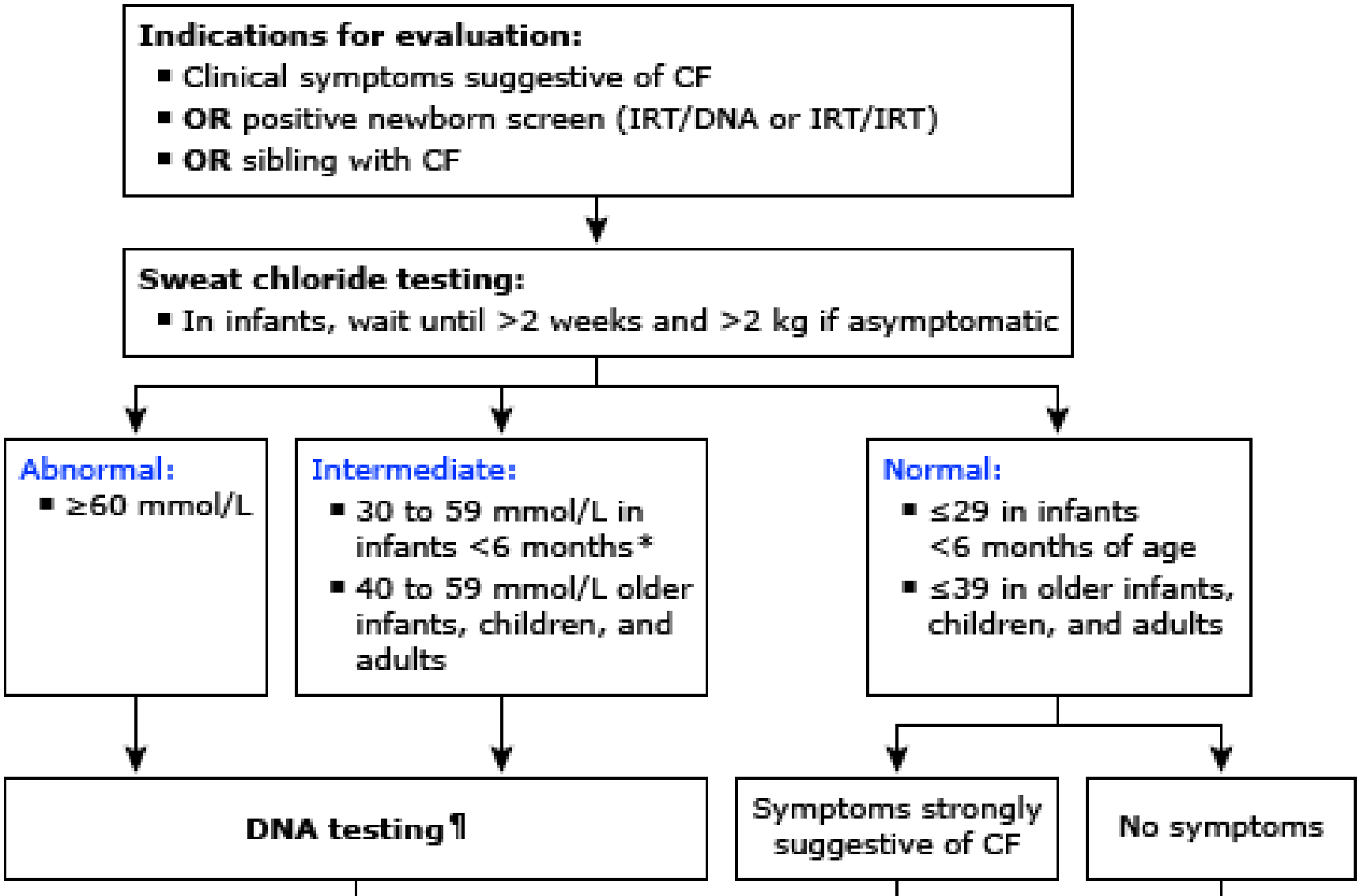
Cystic Fibrosis – clinical features

- **Rectal prolapse** (now rare, ineffetive pancreatic enzyme therapy)
- **Meconium ileus** (obstruction of the bowel by meconium in a newborn infant)
- **DIOS** – distal intestinal obstructive syndrome (episodes of small bowel obstruction)
- **Hepatobiliary disease**

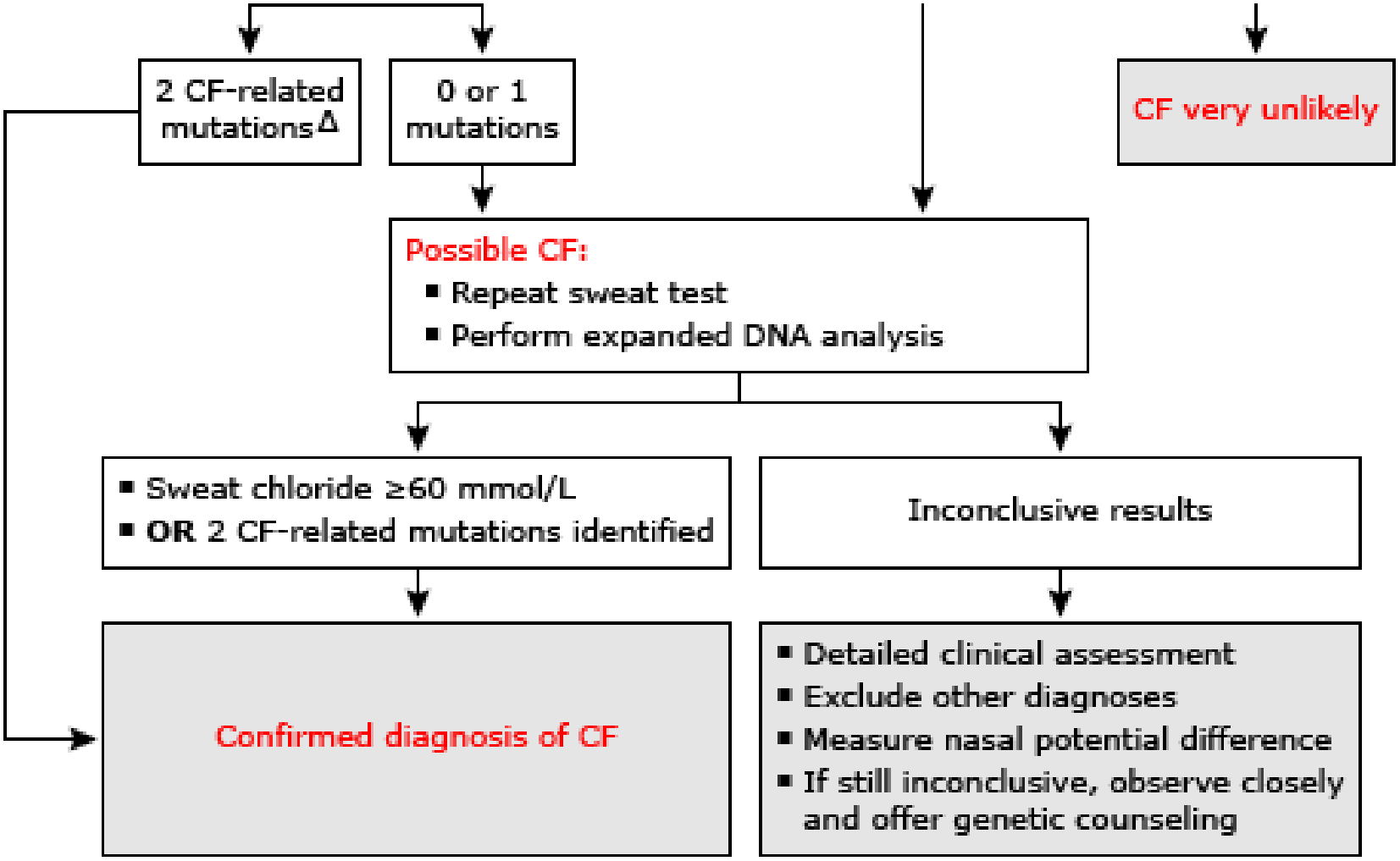
Cystic Fibrosis – clinical features

Infertility (defects in sperm transport, abnormally tenacious cervival mucus)

Cystic Fibrosis – diagnosis



Cystic Fibrosis – diagnosis



Cystic fibrosis - management

- The management of the child with CF requires close a multidisciplinary team approach, which should include:
- paediatric pulmonologist,
- physiotherapist,
- dietician
- psychologist

Cystic fibrosis - management

- PULMONARY CARE – physiotherapy at least twice a day
- chest percussion
- postural drainage
- deep breathing exercise
- use of flutter or acapello device

Cystic fibrosis - management

- PULMONARY CARE
- antimicrobial therapy: nebulized and oral, iv for acute exacerbations,
- mucolitycs: recombinant DNAase, inhaled hypertonic (7-10%) saline,
- Immunizations
- CFTR modulators

Cystic fibrosis - management

- GASTROINTESTINAL CARE
  - For DIOS: Lactulose, Oral ACC solution, Gastrografin
  - NUTRITION
- pancreatic supplements taken with all meals and snacks
- high-calorie diet – 120-150% of normal energy intake
- salt supplements
- fat-soluble vitamins - ADEK


Cystic Fibrosis
Care

Primary ciliary dyskinesia (PCD)

Congenital impairment of mucociliary clearance

Underlying cause=defect of cilia in airways, making them:

- Unable to beat (ciliary immotility)
- Unable to beat normally (ciliary dyskinesia)
- or absent altogether (ciliary aplasia)

PCD

- Freqency : one in 10,000 to 30,000 individuals
- Inherited as autosmal recessive disease
- More than 30 different genetic variants described

PCD - clinical manifestations

- Considerable variation exists in the clinical presentation of PCD.
- The most common features are reccurent infections of the upper and lower respiratory tract.
- Most patients with PCD present in childhood (age of diagnosis is over 5 years).

PCD - clinical manifestations

- Approximately 50% have situs inversus totalis (defective embryonic, nodal cilia)
- When
- - situs inversus,
- - chronic sinusitis,
- - and bronchiectasis occur together,
- an individual is said to have
- KARTAGENER'S SYNDROME

PCD - clinical manifestations

- PULMONARY
- **Newborns** – mild respiratory distress
- **Infants/older children** – increased incidence of respiratory infections with chronic cough and expectoration of mucopurulent sputum (symptoms tend to increase during the day, rather than peak in the morning)

PCD - clinical manifestations

- PULMONARY
- Bronchiectasis…
- It usually affects the middle and lower lobes and lingula

PCD - clinical manifestations

- RHINOSINUSITIS
- Constantly runny nose
- Year-round nasal congestion
- Severe course of common colds

- Cardinal feature of PCD

PCD - clinical manifestations

- OTITIS – chronic secretory otitis media with reccurent episodes of acute otitis media,
- Conductive hearing loss is common
- HYDROCEPHALUS
- INFERTILITY – immotile spermatozoa, impaired ciliary function in the fallopian tubes

PCD - diagnostic evaluation

- No „gold standard" diagnostic test has been established
- Each test has adv. and disadv.

- A combination of tests is necessary for accurate diagnosis of PCD.

PCD - diagnostic evaluation

- Measuring nasal nitric oxide **(nNO)** – it is very low or absent in patients with PCD
- **HSVA** – high speed videomicroscopy analysis; samples obtained from nasal bruishing
- **TEM** – transmission electron microscopy – to confirm and find type of ciliary abnormality

PCD - diagnostic evaluation

- Genetic testing – it is usually reserved for patients with normal or equivocal HSVA nad TEM and a strongly suggestive history

- PICADAR scale

PCD - management

- Treatment is based on experience with treating patient with CF and other forms of bronchiectasis.
- daily chest physiotherapy including postural drainage and coughing,
- antibiotics for pulmonary exacerbations,
- polypectomy has been beneficial in a number of patients for relief of sinusitis.

Thank You