Case Presentation

By

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Profile

Name: Imran

Age: 14 yrs

Sex: Male

Residence: Sadiqabad, Rawalpindi.

Date of Admission: 1\textsuperscript{st} Dec. 2008
Presenting Complaints

- Productive cough
- Progressive breathlessness

1 yr
History of Present Illness

- Cough – early morning & after activity
- Copious and tenacious sputum
- Sputum volume around 2-3 cups/day
- Purulent & foul smelling, no hemoptysis
- Breathlessness
- Orthopnea
- Occasional cyanosis
- Wheeze
- High grade fever
- No H/O stridor & chest pain
Systemic Inquiry

- Bulky and greasy stools
- Lack of appetite and weight loss
- No H/O
  - Nasal obstruction or nasal discharge
  - Earache & discharge,
  - Polyuria, polydypsia,
  - Jaundice,
  - Hematemesis, petechiae & bruises
  - Night blindness, Ataxia
  - Rectal prolapse
Past History

- Recurrent pneumonia from 1 month of age
- Frequent hospitalizations
- Adverse effects on growth & development
- Usual treatment with antibiotics
- Received ATT twice at the age of 5 & 9yrs
Birth History

- Antenatal history was unremarkable
- Spontaneous vertex delivery at home
- No perinatal complications
Developmental History

Delayed motor milestones

Neck holding: 7 months
Sitting: 1 yr
Walking: 2 ½ yrs

Immunization History

Vaccinated according to EPI schedule

BCG scar is present
Family History

14yrs  12yrs  10yrs  6yrs  4yrs  2yrs
Social History

- Discontinuation of schooling
- Limitation of physical activity
- Psychological effects of the illness
- Financial considerations
Examination: General Physical Examination

- An ill looking young boy in respiratory distress

Vitals

- R/R: 45/min
- Pulse: 102/min
- Temp: 99 F
- Blood Pressure: 100/60 mmHg
General Physical Signs

- Clubbing
- Pallor
- Cyanosis
- Edema feet
- Absent secondary sexual characters
- No lymphadenopathy and stigmata of chronic liver disease
Anthropometric Measurements

- Height: 132 cm < 5\textsuperscript{th} centile
- Weight: 24 kg < 5\textsuperscript{th} centile
- OFC : 51 cm < 5\textsuperscript{th} centile
Systemic Examination

Respiratory System:

Inspection:

- Barrel shaped chest
- Pectus Carinatum
- Bilaterally equal chest movements
Palpation:

- Central trachea
- Reduced Chest expansion
- Normal & equal vocal fremitus on both sides
Percussion

- Resonant on both sides

Auscultation

- Vesicular breathing
- B/L coarse crepts and rhonchi
- Vocal resonance normal and equal on both sides
Examination of CVS

**Pulse**
- 102/min, Regular, Normal volume & character
- No radiofemoral delay

**JVP**
- Raised
Precordium

**Inspection:**
- Pulsations in epigastrium
- No bulge, prominent veins & scar marks

**Palpation**
- Apex beat – 4th intercostal space medial to midclavicular line with normal character
- Parasternal heave
- No palpable heart sounds or thrill
Auscultation

- $S_1$ $S_2$ audible, normal intensity & character
- No murmur
Abdominal Examination

**Inspection**

- Normal shaped abdomen with Central umbilicus
- No prominent veins or scar marks
- Intact hernial orifices

**Palpation**

- Tender Hepatomegaly
Percussion

- No evidence of free fluid in peritonium

Auscultation

- Normal bowel sounds
- No bruit
Examination of the CNS

Higher Mental Functions

- Fully conscious, well oriented
- Mood swings
- Good memory & general intelligence

Cranial Nerves

- Intact
Motor System

- Wasting of muscles
- Tone & power is normal in all muscle groups
- Deep & superficial reflexes intact
- No cerebellar signs

Sensory System

- Intact primary & cortical sensations
Examination of ENT

**Ear**
- Normal tympanic membrane
- No conductive or sensorineural hearing loss

**Nose**
- Normal nasal mucosa
- No polyps

**Throat**
- Normal Oropharynx, Nasopharynx and laryngopharynx
Summary

A 14 yrs old boy with recurrent respiratory infections since infancy. Now presented with acute exacerbation of his respiratory symptoms. He is having clubbing, cyanosis, barrel shaped chest and pectus carinatum with signs of right heart failure.
Differential Diagnosis

Cystic Fibrosis
Primary Ciliary Dyskinesia
Immunodeficiency Syndrome
Resistant Pulmonary Tuberculosis
Investigations

**BLOOD CP**

- Hb: 9.7 gm/dl
- TLC: 11x $10^9$/L
- PLATELET COUNT: 214 x $10^9$/L
- ESR: 40 mm/1st h
DNA testing for CFTR Mutations

ΔF508 Mutation +ve

Sweat Chloride Test

Serum Amylase 
Normal

Stool for fat globules
Positive
Obstructive pattern with reduced FVC & FEV1 / FVC 76%
Sputum for Microbiologic Studies

Pseudomonas
E.coli

Sputum for AFB
Negative
Serum Immunoglobulins – Normal

IgA  302mg/dl  (70-400 mg/dl)
IgG  1809mg/dl (700-1600mg/dl)

Complement levels – Normal

C3   93mg/dl  (70-196mg/dl)
C4   10.3mg/dl  (13-38mg/dl)
LFTs

Serum bilirubin: 0.8 gm/dl
ALT: 35
Alkaline Phosphatase: 310u/l

RFTs

Serum urea: 17 mg/dl
Creatinine: 0.4 mg/dl
Uric Acid: 3.9 mg/dl
Serum Electrolytes

Sodium: 135 mmol/L
Potassium: 4.3 mmol/L
Calculus: 8.6 mg/dl

Random Blood Sugar

82 mg/dl
ECG

Right ventricular hypertrophy
Echocardiography Report

Patient Name: IMRAN  Age/Sex: 12 yrs/m.c
Ward / Bed / OPD/ No.  WD/5  Ref No.  556  Date: 01/01/2009

Diagnosis:

REPORT

Dilated RAD RV with mild TR.
Good LV systolic function. No focal defect seen. No clot or vegetation.
No PE seen.

Manzoor 01/11/2009
Echocardiographer
Final diagnosis
Cystic Fibrosis
Multidisciplinary approach

- Pulmonologist
- Cardiologist
- Physiotherapist
- Psychiatrist
Treatment

**IV Antibiotics**

- **Inj: Cefotaxime** 1.5 gm TDS
- **Inj: Ampicloox** 1.25 gm QID
  
  **For 2 wks**
  
- **Inj: Vancomycin** 650mg TDS
- **Inj: Fortum** 1.5 gm TDS
  
  **For 2 wks**
  
- **Inj: Cefoperazone** 1.5 gm TDS
- **Inj: Amikacin** 500mg BD

Continued
Inhalational Therapy

Nebulization with:
- Ventolin
- Clenil
- Atrovent

Oral Corticosteroids

Oxygen inhalation

Chest Physiotherapy
Antifailure drugs

Digoxin: 0.75 mg P/O OD
Spiromide: 25 mg P/O BD
Captopril: 15 mg P/O BD

Dietary Therapy

High protein diet
vitamin supplementation
Treatment Response:

- Improved symptomatology
- Cough & shortness of breath
- O₂ requirement
- Activities of daily life
Thank You