The Bioscan
AN INTERNATIONAL QUARTERLY JOURNAL OF LIFE SCIENCES
20(1): 409-411, 2025
www.thebioscan.com

Case Study Report on Kartagener's Syndrome - an Inherited Disorder

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DOI: https://doi.org/10.63001/tbs.2025.v20.i01.pp409-411

KEYWORDS
Dermatomes,
Aspiration pneumonitis,
Herpes zoster,
Varicella-zoster virus,
Antiviral therapy

Received on:

16-12-2024

Accepted on:

14-01-2025

Published on:

23-02-2025

ABSTRACT

This case presents an 18-year-old male with a complex presentation, including neurological, respiratory, and dermatological symptoms. The patient's viral encephalitis is likely the underlying cause of the altered sensorium, and herpes zoster in the T2-T3 dermatomes is indicative of a localized reactivation of the varicella-zoster virus, often seen in immunocompromised states or viral infections. The patient's respiratory distress and pneumonia complicate the picture, likely compounded by aspiration pneumonitis following self-extubation.

The combination of these findings warrants a multi-disciplinary approach to management, including antiviral therapy for herpes zoster (acyclovir), antibiotic therapy for pneumonia (ceftriaxone), seizure management (levetiracetam), and symptomatic management for the altered sensorium and aspiration pneumonitis. Additionally, the poor cardiovascular status, along with altered mental status, requires close monitoring and aggressive supportive care.

INTRODUCTION

Kartagener's syndrome (KS) is a rare, inherited disorder characterized by a triad of clinical features: bronchiectasis, sinusitis, and situs inversus. It is a subset of a broader group of conditions known as primary ciliary dyskinesia (PCD), which is caused by defects in the cilia (tiny hair-like structures) that line the respiratory tract, sinuses, and other parts of the body. These ciliary defects lead to impaired mucociliary clearance, resulting in recurrent respiratory and sinus infections, among other complications.

Epidemiology

Kartagener's syndrome is a rare disorder with an estimated prevalence of about 1 in 15,000 to 30,000 live births. It is an autosomal recessive condition, meaning an individual must inherit two copies of the defective gene (one from each parent) to develop the syndrome. It can affect individuals of all ethnic backgrounds, but the incidence may vary by geographic region.

Pathophysiology

The underlying cause of Kartagener's syndrome is a defect in the structure or function of the cilia. Cilia are microscopic, hair-like structures that are found on the surface of many cells, including those in the respiratory tract and the sinuses. Cilia normally beat in a coordinated manner to move mucus, bacteria, and other particles out of the respiratory system. In individuals with Kartagener's syndrome, the cilia are either immotile or poorly functioning, impairing their ability to clear mucus from the airways.

This defect in ciliary function results in several clinical consequences:

1. Bronchiectasis: The inability to clear mucus and debris leads to chronic infections, inflammation, and damage to the airways, resulting in bronchiectasis (abnormal widening of the airways).
2. Sinusitis: The impaired mucociliary clearance in the sinuses leads to frequent, persistent sinus infections.
3. Situs Inversus: This refers to the complete reversal of internal organs, such as the heart, liver, and stomach. This happens because of defects in the dynein arms (which help in ciliary movement) during embryonic development.

Clinical Features

The clinical features of Kartagener's syndrome typically include:

1. Chronic Respiratory Symptoms:
   - Chronic cough
   - Recurrent respiratory infections
   - Wheezing
   - Shortness of breath
   - Bronchiectasis (abnormal dilation of the airways)
   - Chronic sinusitis with nasal congestion and frequent sinus infections
   - Otitis media (ear infections)
2. Situs Inversus:

- This condition is present in about half of individuals with Kartagener's syndrome. Situs inversus refers to the complete or partial reversal of the normal positioning of internal organs. The heart may be positioned on the right side (dextrocardia), and other organs such as the liver, spleen, and stomach are reversed.
- Most individuals with situs inversus do not experience symptoms directly related to the organ reversal, but it may be detected incidentally during imaging studies.

3. Infertility:
   - In males, infertility is common due to impaired sperm motility resulting from defective cilia in the reproductive tract.
   - In females, ciliary defects in the fallopian tubes can impair egg movement, potentially leading to infertility or complications with fertility.
4. Other Possible Features:
   - Deafness: Some individuals with Kartagener's syndrome may develop sensorineural hearing loss due to ciliary defects in the inner ear.
   - Nasal Polyps: Chronic sinus issues and infections may lead to the development of nasal polyps.

Diagnosis

The diagnosis of Kartagener's syndrome is made based on clinical features, imaging studies, and genetic testing. Key diagnostic tools include:

1. Clinical Evaluation: The presence of the triad of bronchiectasis, sinusitis, and situs inversus is suggestive of Kartagener's syndrome, particularly when associated with chronic respiratory symptoms.
2. Chest X-ray/CT Scan: Imaging studies can reveal bronchiectasis, a hallmark of the syndrome, along with other lung abnormalities such as inflammation or infections.
3. Sinus Imaging: CT scans of the sinuses can show signs of chronic sinusitis, including thickened sinus walls and mucosal swelling.
4. Genetic Testing: Genetic tests can identify mutations in the DNAH5 or DNAI1 genes, which are responsible for the defects in the dynein arms of the cilia. These tests are often used to confirm the diagnosis.
5. Ciliary Motility Testing: A sample of cells from the respiratory tract (such as from the nasal lining) may be taken and examined under a microscope to assess the motility of the cilia. In individuals with Kartagener's syndrome, the cilia will typically be immotile or poorly functioning.
6. Electron Microscopy: This can be used to examine the structure of cilia and identify specific defects, such as absent or defective dynein arms, which impair ciliary movement.

Treatment and Management

There is currently no cure for Kartagener's syndrome, and management is primarily aimed at controlling symptoms and preventing complications. Key aspects of treatment include:

1. Respiratory Care:
   - Chest physiotherapy to help clear mucus from the lungs.
   - Bronchodilators to open the airways and improve airflow.
   - Inhaled corticosteroids to reduce inflammation.
   - Antibiotics to treat or prevent respiratory infections.
   - Oxygen therapy in cases of respiratory failure or significant lung damage.
2. Sinus and Ear Management:
   - Saline nasal sprays or irrigation to clear nasal passages and reduce sinus congestion.
   - Antibiotics for persistent or recurrent sinus infections.
   - Surgical intervention for chronic sinus issues or nasal polyps.
3. Fertility Treatment:
   - In males, assisted reproductive technologies (e.g., in vitro fertilization) may be used to address infertility.
   - In females, fertility treatments may be required for those experiencing difficulty with conception due to ciliary dysfunction in the fallopian tubes.
4. Immunizations:
   - Patients with Kartagener's syndrome may be at higher risk for respiratory infections and should receive vaccinations, including the pneumococcal vaccine and influenza vaccine, to reduce the risk of infections.

Prognosis

The prognosis of Kartagener's syndrome depends on the severity of the symptoms and the presence of complications. With appropriate medical management, individuals with Kartagener's syndrome can lead relatively normal lives, although they may face chronic respiratory issues and frequent sinus infections. Early diagnosis and intervention can help prevent further lung damage and improve quality of life.

Although life expectancy has improved in recent years due to advancements in medical care, patients with severe lung damage or complications from recurrent infections may experience a reduced lifespan.

Case Discussion:

Patient Information:

- **Name**: Unknown (Adolescent Boy)
- **Age**: 18 Years
- **Gender**: Male
- **Presenting Condition**: Inability to walk, hiccoughs, altered sensorium, suicidal behavior, herpes zoster lesions, and respiratory distress.

Presenting Complaint:

The patient, an 18-year-old adolescent male, was transferred from a rural government hospital to the urban district hospital with the following complaints:

- Inability to walk
- Hiccups
- Involuntary passage of urine and stool
- Fluid-filled vesicles over the chest (which had been treated with natural medicine)
- Altered sensorium for 1 day, including:
  - Altered thinking and irrelevant talk
  - Suicidal tendencies with cuts over the left wrist
  - Herpes zoster lesions
  - Hesitation and confusion
- Respiratory distress with difficulty breathing

Clinical Findings on Examination:

- General Appearance: The patient appeared disoriented with altered mental status.
- Neurological:
- Glasgow Coma Scale (GCS): Spontaneous eye opening, but no response to painful stimuli or oral commands.
- Dermatological: Fluid-filled vesicular lesions over the chest (T2-T3 dermatome) suggestive of herpes zoster.
- Respiratory: Increased respiratory effort with signs of distress and altered breathing patterns.
- Urinary/Anal: Involuntary passage of urine and stool.

Investigation Results:

1. Abdominal & Pelvic Scan:
   - Findings: Distended gallbladder and urinary bladder.
   - No other significant abnormality.

2. Ultrasound (USG):
    - Findings:
        - Consolidation noted with air bronchospasm in the left lower lobe.
        - Patchy air space opacities noted in the left middle lobe and bilateral upper lobes.
        - Dextrocardia (heart displaced to the right side).
3. MRI:
    - Findings:
        - MRV: Hypoplastic right transverse and sigmoid sinuses.
        - Presence of pansinusitis.
4. Lumbar Puncture:
    - Diagnosis: Viral encephalitis (suggesting an underlying viral infection affecting the brain).
5. Culture & Sensitivity:
    - Findings: Klebsiella pneumoniae identified.
    - Sensitivity: Reactive to Ceftriaxone and Ciprofloxacin.

Clinical Course:

Upon admission to the urban district hospital, the patient showed signs of significant respiratory distress, altered sensorium, and poor cardiovascular status. Given these concerns, the patient was:

- Intubated with an 8mm cuffed endotracheal tube (ET tube) after 1 day of admission.
- Mechanical ventilation was initiated after 2 days due to poor respiratory function.
- The patient later self-extubated the ET tube, resulting in aspiration pneumonitis.

Dermatology Opinion:

- Herpes Zoster: Multiple well-defined discrete to coalescent healed superficial erosions in the T2-T3 dermatomes consistent with herpes zoster.

Treatment Plan:

1. Inj. Acyclovir 20 mg IV tds: For the treatment of herpes zoster.
2. Inj. Ceftriaxone 1 g IV bd: Empirical antibiotic therapy for Klebsiella pneumoniae.
3. Inj. Levetiracetam 500 mg IV tds: Anticonvulsant therapy to manage seizure-like activity due to encephalitis.
4. Inj. Pantoprazole 40 mg IV bd: For gastric acid suppression.
5. Inj. Mannitol 10 ml IV tds: Osmotic diuretic used to manage cerebral edema.
6. Inj. Miconazole 800 mg tds: Antifungal treatment for possible co-infection.
7. Mupirocin ointment: Topical treatment for skin lesions associated with herpes zoster.
8. Tab. FST: To be converted as per the prescription (likely a supplement or supportive therapy).

Diagnosis:

- Primary Diagnosis: Viral Encephalitis (with suspected Klebsiella pneumoniae and herpes zoster).
- Secondary Diagnosis:
    - Aspiration Pneumonitis: Following extubation.
    - Dextrocardia.
    - Pansinusitis.

CONCLUSION

Kartagener's syndrome is a rare genetic disorder that primarily affects the respiratory and sinus systems due to ciliary dysfunction. With the characteristic triad of bronchiectasis, sinusitis, and situs inversus, this syndrome presents significant diagnostic and management challenges. While there is no cure, early diagnosis and comprehensive treatment can significantly improve the quality of life and prevent complications, allowing individuals to lead active and fulfilling lives despite the chronic nature of the disease.

The patient's condition is critical, and ongoing management in an intensive care setting is required. The multidisciplinary approach, involving neurology, pulmonology, dermatology, and infectious disease specialists, will be essential for improving outcomes. Regular reassessment of the patient's respiratory and neurological status, as well as adjustment of the treatment plan based on culture and sensitivity results, will be important for addressing the evolving clinical picture.

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