

Kartagener's syndrome

Synonyms: Afzelius' syndrome, KS, Kartagener's triad, Siewert's syndrome, dextrocardia-bronchiectasis-sinusitis syndrome, primary ciliary dyskinesia

What is Kartagener's syndrome?

Kartagener's syndrome is described as autosomal recessive inherited syndrome. Primary (genetic) defects in the structure and function of sensory and motile cilia result in multiple ciliopathies.

A reduction in the number of arms which propel mucus (dynein arms) is a common abnormality but many other structural defects of the cilia have been found. Patients with normal cilia morphology but abnormal mucus propulsion have been detected.

In diagnosing the syndrome, therefore, both ciliary structure and motility need to be assessed.^[1]

Kartagener's syndrome consists of a triad of features:^[2]

- Situs inversus (transposition of the viscera).
- Abnormal frontal sinuses (producing sinusitis and bronchiectasis).
- Primary ciliary dyskinesia (PCD).^[3]

The defective cilia lining the respiratory tract are unable to clear the airways of secretions and pathogenic bacteria, resulting in mucus retention and chronic or recurrent respiratory tract infection - leading to damage to airway walls.

Approximately half of patients with PCD have the full triad of Kartagener's syndrome (KS).^[4] There may also be a link with [retinitis pigmentosa](#) and hearing loss.^[5]

It is suspected that visceral rotation in the embryo is dependent upon normal ciliary action – hence, the association between primary ciliary dyskinesia and situs inversus abnormality.

How common is Kartagener's syndrome? (Epidemiology)

The incidence of the genetic disorder is 1 in 32,000 births.^[6] However, higher incidences have been found in communities in which consanguineous marriages are common.^[7]

Kartagener's syndrome symptoms^[1] ^[8]

History

- Neonatal respiratory distress may occur.^[9]
- Upper respiratory symptoms may include: chronic rhinorrhoea from early childhood, reduced sense of smell and chronic rhinitis.
- Recurrent otitis media may occur.
- [Chronic obstructive pulmonary disease \(COPD\)](#), [bronchiectasis](#) and recurrent [pneumonia](#) may all be components of the syndrome.
- [Male infertility](#) due to immobile spermatozoa and decreased fertility in females may also occur.

Examination

Findings may include dextrocardia and situs inversus, asplenia, nasal polyps, rhinitis, corneal abnormalities and conductive deafness. Extremities may exhibit distal clubbing.

Differential diagnosis^[9] ^[10]

Conditions which may need to be considered include:

- [Allergic rhinitis](#).
- Adenoid hyperplasia.
- Allergic bronchopulmonary [aspergillosis](#).
- Bronchial obstruction.

- Conditions linked to [bronchiectasis](#):
 - Acquired obstruction- foreign body, tumour, lymphadenopathy, COPD, mucoid impaction and connective tissue diseases.
 - Congenital obstruction - bronchomalacia, tracheobronchomegaly, ectopic bronchus, pulmonary sequestration, pulmonary artery aneurysm and [yellow nail syndrome](#).
 - [Immunodeficiency states](#) with recurrent infections - IgG, IgA deficiencies, abnormalities of leukocyte function, conditions affecting primary antibody production.
 - Abnormal secretion clearance - immotile cilia syndrome, [cystic fibrosis](#), Young's syndrome.
 - Miscellaneous disorders - [alpha-1 antitrypsin deficiency](#), recurrent [aspiration pneumonia](#), inhalation of poisonous dust and fumes and chronic rejection following organ transplantation.
- Chronic aspiration.
- Congenital cartilage deficiency.
- Idiopathic [nasal polyposis](#).
- Interstitial lung diseases - [idiopathic pulmonary fibrosis](#), idiopathic interstitial pneumonias.
- Malignancies - bronchoalveolar carcinoma.
- Samter's triad (asthma, aspirin sensitivity and nasal/ethmoidal polyposis).
- Severe atopy.

Investigations^[8] ^[10] ^[11] ^[12]

- CXR may show dextrocardia, lung over-inflation, bronchial wall thickening and peribronchial infiltrates. Right-sided heart disease with chronic respiratory tract symptoms is highly indicative and occurs in half of patients.

- CT scan for bronchiectasis and to demonstrate involvement of paranasal sinuses (poorly aerated mastoids ± absence of frontal sinuses).
- Transmission electron microscopy of cilia from airway biopsy (nasal mucosa or tracheal mucosa taken when the patient is not acutely ill). The specimen is examined for ciliary movement, beat frequency, coordination and amplitude. The diagnosis of two newborns, using nasal mucosal biopsy, has been reported.^[13] The most common ultrastructural defect is an absence or decrease in the number of inner or outer dynein arms. This investigation is helpful in differentiating primary from secondary ciliary dysfunction.
- Semen analysis in postpubertal males may reveal abnormal sperm motility and ultrastructure.
- The saccharin test is only used occasionally due to pragmatic difficulties. Saccharin is placed in the nose and the speed of transport to the nasopharynx is measured.
- Measurement of nasal nitric oxide is a useful screening test, which has the advantage of being non-invasive. It involves the patient breathing in nitric oxide and then measuring the level during exhalation through the mouth or nose. The level is low in KS patients due to reduced ciliary clearance in the paranasal sinuses.^[14]
- Pulmonary function tests may be abnormal:
 - Spirometry may reveal an obstructive picture with a reduction in the ratio of forced expiratory volume in 1 second to forced vital capacity, decreased forced expiratory volume in 1 second and a reduction in respiratory flow of 25–75%.
 - Static lung volumes may demonstrate hyperinflation.
 - The response to bronchodilators is variable.

- Surgical procedures:
 - Examination of mucous membrane is the gold standard investigation. This is best obtained when the patient is not acutely ill, as ciliary morphology or function may be affected. Tracheal biopsy provides the best specimen but needs general anaesthesia. Nasal mucus is more readily available. Nasal brushings are less invasive but often yield inadequate results.
 - Children with KS frequently end up having adeno-tonsillectomies which can provide a fruitful source of material for histopathology and electron microscopy.
 - Nasal endoscopy may be required to detect polyps.

Efforts to standardise the clinical criteria for the diagnosis of KS have centred on dextrocardia, a ciliary beat frequency of less than 10 Hz/second and a mean cross-section dynein arm count of less than 2. If the patient does not have dextrocardia, the identification of primary ciliary dyskinesia becomes the mainstay of diagnosis. Genetic testing ultimately may become the principal means of establishing this diagnosis.^[15]

Kartagener's syndrome treatment^[8] ^[9]

Medical care

- Antibiotics - intravenous or oral, intermittent or continuous - are used to treat upper and lower airway infections. *Haemophilus influenzae* and *Staphylococcus aureus* are the most common organisms. Long-term low-dose prophylactic antibiotics may be necessary in children.
- Obstructive lung disease/bronchiectasis should be treated with inhaled bronchodilators, mucolytics and chest physiotherapy. The effectiveness of deoxyribonuclease and other mucolytic agents such as hypertonic saline and acetylcysteine has not been fully assessed but may be worth trying in patients with recurrent infections or persistent respiratory symptoms.^[10]
- The evidence base is largely anecdotal but there may also be a role for inhaled antibiotics and inhaled and oral corticosteroids.^[16]

- [Influenza](#) and [pneumococcal vaccination](#) should be encouraged.^[16]
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Surgical care^[16]

- Tympanostomy tubes will reduce recurrent infections and conductive hearing loss. Repeated insertions may be required and chronic suppurative otitis media can be an annoying complication. Aural hygiene measures, such as acetic acid irrigations, otomicroscopy, or topical or systemic antibiotic therapy may be required.
- Endoscopic sinus surgery and the formation of a nasal antral window underneath the inferior turbinate, may afford a transient improvement in upper and lower respiratory tract symptoms.
- Lobectomy is sometimes required for the associated bronchiectasis.^[17] Lung transplantation and heart-lung transplantation have occasionally been tried in severe cases, with some success.^[18]

Complications^[13]

Complications include bronchiectasis, pneumonia, conductive deafness and communicating hydrocephalus.

Prognosis^[16]

Treatment of Kartagener's syndrome with antibiotics, physiotherapy and appropriate surgical intervention has improved the prognosis and, in many cases, lifespan may be normal. Early diagnosis is important. Once bronchiectasis is established, prognosis worsens significantly.

Further reading

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