

Suspected Primary Ciliary Dyskinesia in Chronic Rhinosinusitis with Nasal Polyposis and Situs Inversus Totalis: Diagnostic challenge in a resource-limited setting.

Dr. Sanyam Mahajan¹, Dr Rubina Galib², Dr Mehtab Alam³

Department of Otorhinolaryngology, Jawaharlal Nehru Medical College and Hospital, Aligarh Muslim University, Aligarh, Uttar Pradesh, India

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ABSTRACT

A 28-year-old man presented with chronic rhinosinusitis and bilateral nasal polyposis, and was found to have situs inversus totalis. This combination raised the suspicion of primary ciliary dyskinesia. Computed tomography showed features of allergic fungal rhinosinusitis with no bronchiectasis on chest imaging. As specialised testing was unavailable, the diagnosis could not be confirmed. This case report highlights the need to consider Primary ciliary dyskinesia (PCD) in patients with chronic sinonasal disease and situs inversus, even without bronchiectasis.

Keywords: Primary ciliary dyskinesia; Chronic rhinosinusitis; Situs inversus totalis; Allergic fungal rhinosinusitis.

Background

Primary ciliary dyskinesia (PCD) is an inherited disorder of motile cilia that impairs mucociliary clearance which can cause chronic disease of both the upper and lower airways including chronic rhinosinusitis, otitis media, recurrent respiratory infections and bronchiectasis [1, 2]. Abnormal left-right organ development occurs in a substantial proportion of individuals with PCD and may manifest as situs inversus totalis (SIT) [1, 3]. Finding situs inversus in a patient with longstanding sinonasal disease should thus raise the suspicion of PCD. However, situs inversus alone does not confirm the diagnosis, laterality defect can occur in individuals without motile-cilia disorder [3, 4].

Kartagener syndrome describes the classical combination of chronic sinus disease, situs inversus, and bronchiectasis. Present patient had no signs and symptoms of bronchiectasis and PCD was not confirmed by specialised testing. Allergic fungal rhinosinusitis (AFRS) is a distinct sinonasal inflammatory disorder characterised by type I hypersensitivity to fungal antigen associated with nasal polyposis, raised immunoglobulin E (IgE) and a characteristic appearance on computed tomography (CT) [5, 6, 7]. Although the patient's clinical and CT findings were strongly suggestive of AFRS, histopathological confirmation was not available. There are several key differences between primary ciliary dyskinesia, bronchiectasis, chronic rhinosinusitis, and allergic fungal rhinosinusitis [Table 1].

Table-1

Condition	Primary etiology	Upper airway feature	Lower airway feature	Key findings
Primary ciliary dyskinesia	Inherited defect in motile ciliary structure or function	Early-onset chronic rhinosinusitis and nasal polyposis	Recurrent infections, progressive bronchiectasis	Laterality defects such as situs inversus totalis; diagnosis requires appropriate specialized testing
Bronchiectasis	Multifactorial (post-infectious,	Sinonasal symptoms are variable or minimal	Chronic productive cough, irreversible bronchial dilatation	Bronchial dilatation on CT; the underlying cause should be determined clinically.

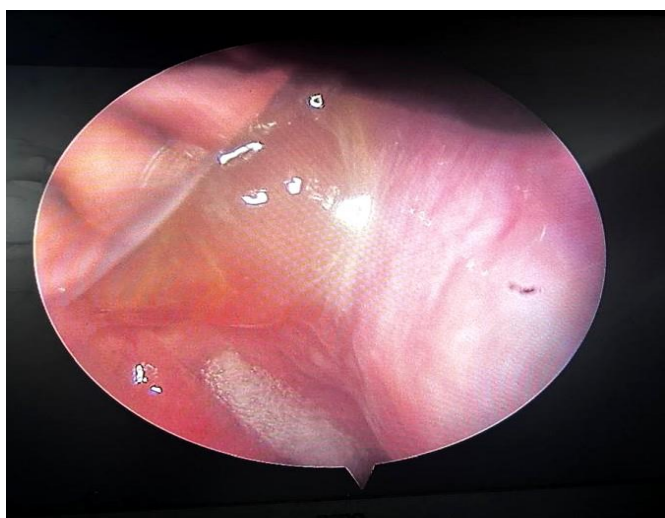
	immunodeficiency, inflammation)			
Chronic rhinosinusitis with nasal polyps	Type 2 eosinophilic mucosal inflammation	Bilateral nasal polyps, nasal obstruction, diffuse opacification	May coexist with asthma and other lower-airway disease	Usually normal organ laterality; ciliary dysfunction is not a defining feature.
Allergic fungal rhinosinusitis	Type 1 hypersensitivity to fungal component	Asymmetric or bilateral nasal polyps, allergic mucin, bone remodelling	Can be associated with Allergic asthma	Elevated total/fungal-specific IgE and characteristic CT findings; allergic mucin and fungal elements may support confirmation

Case Report

A 28-year-old man presented to the otorhinolaryngology outpatient department with nasal obstruction and nasal discharge that had progressively worsened over the preceding four years. He recalled intermittent nasal obstruction since childhood. He denied chronic productive cough, sputum production, recurrent chest infections, or symptoms suggestive of recurrent pneumonia. There was no history of recurrent otitis media, chronic ear discharge, or hearing impairment. He had no history of neonatal respiratory distress, prolonged oxygen requirement, or neonatal intensive care admission. There was no reported history of asthma or aspirin or other nonsteroidal anti-inflammatory drug (NSAID) sensitivity. He had no history of facial pain or pressure, reduced sense of smell or anosmia, visual disturbances, or symptoms suggestive of orbital involvement. He had not undergone previous sinonasal surgery. There was no known family history of situs inversus, chronic respiratory disease, infertility, or consanguinity. He was a non-smoker, with no significant occupational exposure to dust, fumes, or other respiratory irritants. There was no relevant history of tuberculosis or previous antitubercular treatment. He had no history of prolonged or recurrent antibiotic use or systemic corticosteroid therapy for recurrent respiratory infections. He had, however, used a nasal decongestant and intranasal corticosteroid spray intermittently for short periods, with no continuous use exceeding two weeks.

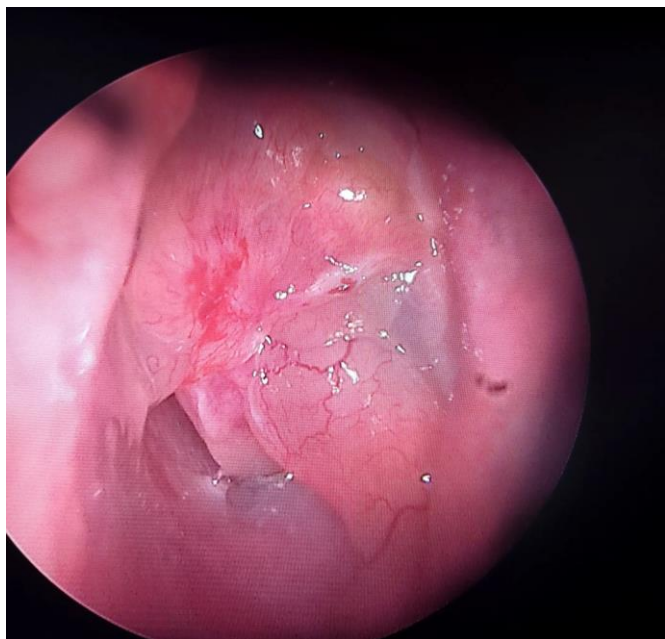
The combination of childhood-onset sinonasal symptoms and situs inversus totalis (SIT) raised suspicion for an underlying mucociliary disorder. However, the absence of chronic wet cough, recurrent lower respiratory tract infections, and bronchiectasis made the clinical phenotype less typical of PCD. As specialised diagnostic testing was unavailable, PCD remained clinically suspected but unconfirmed. Diagnostic nasal endoscopy examination showed bilateral nasal polyps (Fig. 1a, b).

Figure 1a — Nasal endoscopy (view 1)



Diagnostic nasal endoscopy showing polypoid mucosal disease.

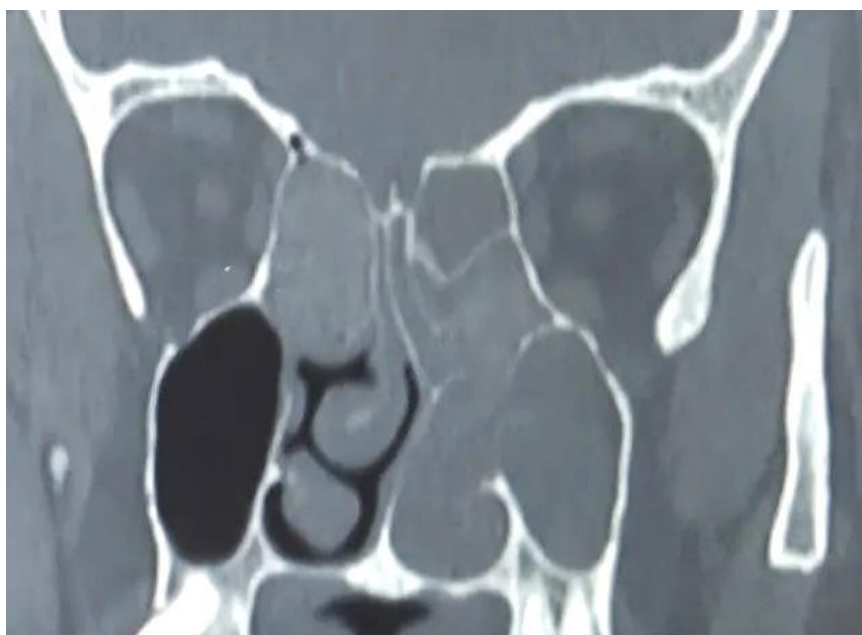
Figure 1b — Nasal endoscopy (view 2)



Diagnostic nasal endoscopy, close-up mucosal view.

The serum total IgE was elevated at 316.7 IU/mL (reference range of 0.1–200 IU/mL). Non-contrast CT of the paranasal sinuses (Fig. 1c, d) demonstrated near-complete opacification of the left maxillary and frontal sinuses and both ethmoid and sphenoid sinuses. The opacified sinuses contained centrally hyperdense material surrounded by hypodense mucosa. The left maxillary ostium was widened, with extension of disease into the left nasal cavity and associated bony remodelling. The nasal septum was deviated to the left, with a mid-septal bony spur contacting the left middle and inferior turbinates and compensatory hypertrophy of the right middle turbinate. Taken together, the appearances were strongly suggestive of allergic fungal rhinosinusitis. However, histopathological confirmation of allergic mucin or fungal elements was not available.

Figure 1c — CT paranasal sinuses (coronal)



Coronal CT: soft-tissue/polypoid opacification with side-to-side asymmetry and double density (AFRS).

Figure 1d — CT paranasal sinuses (coronal)



Coronal CT: maxillary sinus opacification with S-shaped deviated nasal septum.

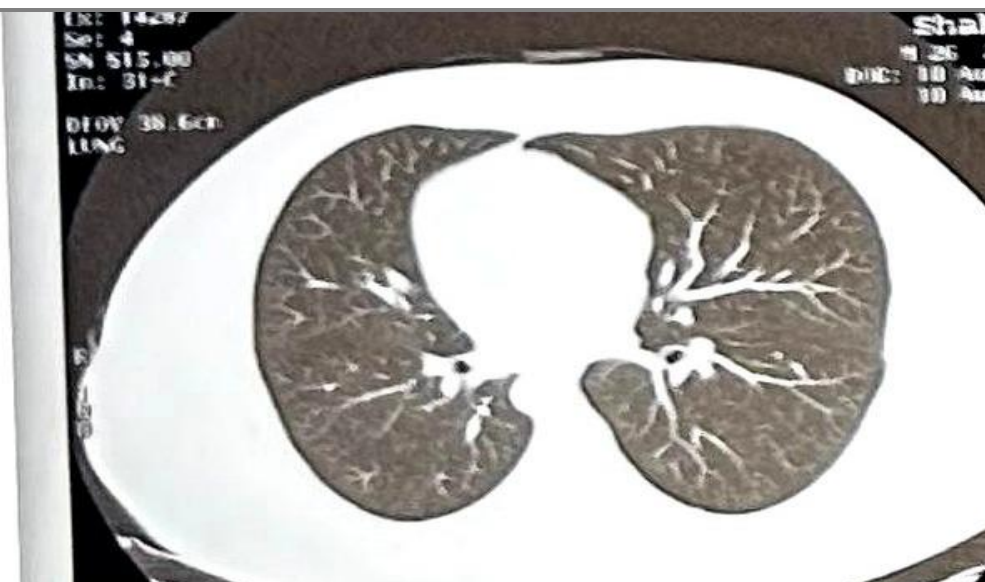
CT of the thorax and abdomen (Fig. 2a, b) demonstrated situs inversus totalis, with dextrocardia, the liver positioned on the left, and the stomach and spleen on the right. Importantly, chest CT showed no bronchiectasis or significant bronchial wall thickening.

Figure 2a — CT upper abdomen (situs inversus)



Axial CT: liver on the left, stomach on the right — (situs inversus).

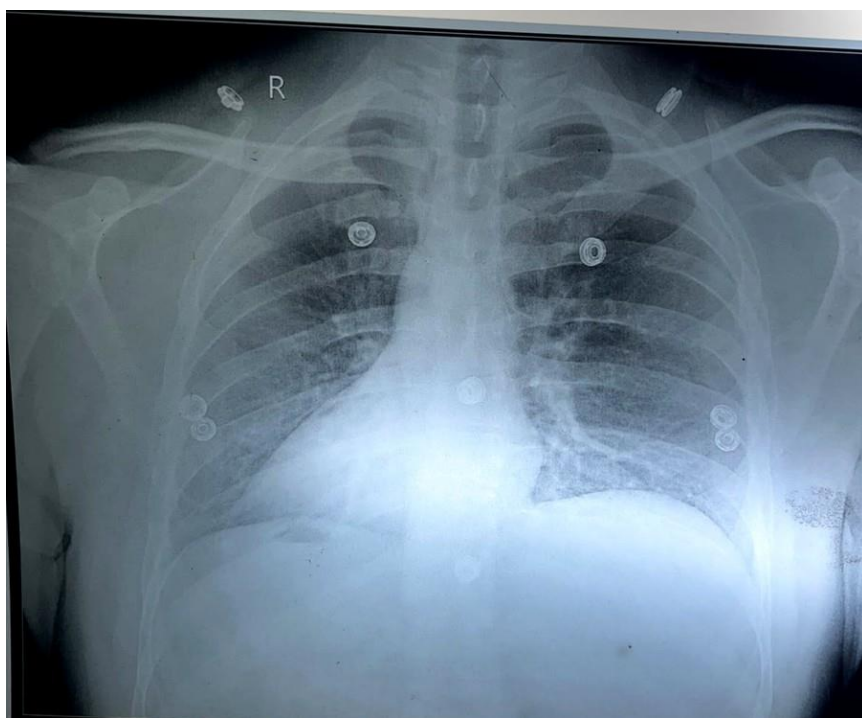
Figure 2b — CT chest (lung window)



Axial lung-window CT: clear lung fields, no bronchiectasis.

Chest X-ray also shows dextrocardia and situs inversus (Fig. 3).

Figure 3 — Chest radiograph (dextrocardia)



Chest radiograph: cardiac apex to the right (dextrocardia), consistent with CT-confirmed situs inverss.

The combination of chronic rhinosinusitis with nasal polyposis and SIT raised clinical suspicion for PCD. However, the patient lacked chronic wet cough, recurrent lower respiratory infections, and bronchiectasis. These findings did not exclude PCD but made the phenotype less typical.

Treatment and Follow-up

Treatment was initiated at the initial outpatient visit with antibiotics, intranasal corticosteroid spray, and saline nasal irrigation for chronic rhinosinusitis with nasal polyposis. Endoscopic sinus surgery was not performed, and fungal investigations, including histopathological examination and fungal studies, were not undertaken,

representing limitations of the case. The patient was referred for specialized PCD assessment, including nasal nitric oxide measurement, high-speed video microscopy, transmission electron microscopy, and genetic testing; as these investigations were not available locally. He was advised to continue ENT follow-up and long-term respiratory surveillance.

DISCUSSION

Situs inversus arises when the embryo fails to establish the usual left–right positioning of the viscera, a step that depends on motile cilia at the embryonic node. Since these are the same cilia that later clear mucus from the airway, it is unsurprising that laterality defects and chronic sinonasal disease so often travel together in PCD [1, 3]. The presence of situs inversus totalis during evaluation of chronic rhinosinusitis should therefore raise clinical suspicion for PCD and prompt appropriate evaluation, although situs inversus alone is not diagnostic [3,4]. A firm diagnosis of PCD depends on objective tests: nasal nitric oxide, which is typically very low; analysis of ciliary beat by high-speed video microscopy; transmission electron microscopy of ciliary ultrastructure; and genetic testing [1, 2]. None of these investigations were available at our institution. Although the clinical findings raised suspicion for PCD, the diagnosis could not be confirmed. It is also a common predicament, since such testing sits in a handful of reference laboratories; where it is out of reach, the diagnosis has to lean on the clinical and radiological picture and stays a matter of strong suspicion rather than proof- a difficulty already reported for PCD and Kartagener syndrome in resource-limited settings [8, 9, 10]. Chronic rhinosinusitis depends upon predisposing factors, inflammatory mechanisms, allergic rhinitis, genetics, cystic fibrosis, aspirin exacerbated respiratory disease, immunodeficiencies, allergic fungal rhinosinusitis and the relationship between upper and lower airways [11]. The diagnosis would be placed beyond doubt by demonstrating eosinophilic allergic mucin, a positive fungal stain or culture and fungus-specific IgE, which together satisfy the accepted criteria [5, 12].

This case highlights the critical role of clinical acumen in diagnosing rare genetic disorders in low-resource settings and emphasizes the need for broader access to specialized diagnostics [13]. It is worth remembering, too, that situs inversus on its own proves nothing, as most people with mirror-image anatomy have perfectly normal cilia [3, 4]. Our patient's chest was clear, with no signs of bronchiectasis. The absence of bronchiectasis makes the clinical phenotype less typical but does not exclude PCD, particularly when pulmonary disease may not yet be established. Thus, a clear chest CT neither confirms nor excludes PCD, and continued clinical follow-up with ciliary testing when available is appropriate. The raised IgE, bilateral nasal polyps, and characteristic CT findings of heterogeneous, centrally hyperdense sinus opacification with expansion and bony remodelling were suggestive of allergic fungal rhinosinusitis. However, as surgery was not performed and fungal investigations and histopathological examination were not undertaken, AFRS could not be confirmed. AFRS is a distinct sinonasal inflammatory disorder characterized by type I hypersensitivity response to fungal antigen and associated with characteristic clinical and radiological findings [5, 6, 7, 11]. The patient fits the familiar mould: Indian series of AFRS describe patients in their late twenties presenting, as this man did, with nasal obstruction and discharge, bilateral polyps, a deviated septum and a raised IgE [9].

A few lessons stand out. Situs inversus turning up during a rhinosinusitis work-up should prompt a careful look for PCD, whose implications- airway surveillance, fertility counselling, screening of relatives- reach well beyond the nose. Where the confirmatory tests are unavailable, the diagnosis has to rest on the clinical and radiological picture, and the patient deserves long-term respiratory follow-up, because features such as bronchiectasis may only declare themselves later. And finding one sinonasal disease should not be allowed to end the search: as here, a second and potentially more serious disorder of the cilia or of organ laterality may be sitting alongside it.

Situs inversus totalis identified during sinonasal evaluation should raise consideration and evaluation for primary ciliary dyskinesia, even in the absence of bronchiectasis. In this case, PCD remained suspected but unconfirmed because specialized ciliary testing was unavailable. The sinonasal findings were suggestive of allergic fungal rhinosinusitis, although histopathological and fungal investigations were not performed. In resource-limited settings, careful clinical assessment, appropriate referral, and long-term respiratory surveillance are important.

CONCLUSION

Chronic rhinosinusitis with nasal polyposis in a young adult who also has situs inversus totalis is a presentation that should bring primary ciliary dyskinesia to mind and prompt a structured evaluation. In this man the chest was clear, with no bronchiectasis, and the specialised ciliary tests were not available to us, so the diagnosis rests for now on clinical suspicion. Lower-airway disease can appear later, which is why long-term respiratory follow-up and confirmatory ciliary testing wherever it can be arranged matters.

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