Case Report

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Kartagener Syndrome with Ectodermal Anomalies in an Adolescent Female: A Case Report

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Abstract

Background: Kartagener's syndrome is a rare autosomal recessive disorder characterized by the triad of bronchiectasis, chronic sinusitis, and situs inversus due to defective motile cilia. Occasionally, ectodermal anomalies such as hair, nail, and dental abnormalities coexist, reflecting involvement of ectodermal structures. the present report is written for the purpose of reminding readers of this rare and severe situation and to emphasize the necessity for further research on how to deal with in best.

Case Presentation: A 17-year-old female presented with recurrent productive cough since childhood, progressive hearing loss, delayed eruption of permanent teeth, patchy alopecia, and dystrophic nails. Imaging revealed dextrocardia and bilateral bronchiectasis. Sweating and secondary sexual characteristics were normal. The constellation of findings suggested Kartagener's syndrome with associated ectodermal anomalies.

Conclusion : This represents a rare overlap of Kartagener syndrome with ectodermal anomalies (Alopecia, dental agenesis, micronychia). Genetic testing is warranted to determine whether this represents a dual diagnosis or a novel syndromic variant.

Key words: Bronchiectasis; Dental agenesis; Dextrocardia; Ectodermal anomalies; Kartagener's syndrome; Primary ciliary dyskinesia.

Introduction

Kartagener's Syndrome (KS) is a rare autosomal recessive disorder and a subset of Primary Ciliary Dyskinesia (PCD) characterized by the classical triad of chronic sinusitis, bronchiectasis, and situs inversus totalis. The condition arises from structural or functional defects in motile cilia, which play a critical role in mucociliary clearance, embryonic organ laterality and fertility.[1,2] Impaired ciliary motility leads to recurrent upper and lower respiratory tract infections, chronic rhinosinusitis, otitis media, bronchiectasis and in approximately 50% of cases, situs inversus.[1.2] Early recognition is essential to prevent progressive lung damage and associated complications.

Ectodermal anomalies including abnormalities of hair, nails, teeth, and sweat glands represent a heterogeneous group of disorders known as ectodermal dysplasias. These arise from defective development of ectodermal derivatives during embryogenesis.[3,4] Although classically considered distinct from PCD, recent evidence suggests a rare phenotypic overlap, possibly due to shared genetic or developmental pathways affecting both ciliary function and ectodermal morphogenesis.

Several studies have reported associations between KS and unusual dental morphology, supporting a potential developmental link between ciliary dysfunction and odontogenesis. Merrett described a patient with Kartagener's syndrome exhibiting congenital absence of an upper lateral incisor, enamel hypoplasia, and abnormal tooth form.[5] Similarly, Pawlaczyk-Kamie ska et al. documented dental agenesis and enamel defects in a ciliopathic disorder with overlapping features of PCD.[6] In a more recent report, Abed et al. discussed dental management challenges in a KS patient presenting with significant structural tooth anomalies.[7] These observations suggest that defective ciliary signaling may extend beyond mucociliary clearance to influence craniofacial and dental development.[5-7]

The coexistence of KS with ectodermal abnormalities thus presents a unique diagnostic and research challenge. Recognizing such overlapping phenotypes is critical, as it refines the understanding of genotype–phenotype correlations, informs genetic counseling and guides multidisciplinary management. The present report describes a 17 year-old female patient exhibiting

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classical features of Kartagener's syndrome along with dental agenesis, dystrophic nails and scarring alopecia, highlighting the clinical and radiological spectrum of this rare association.

Case Presentation

A 17-year-old female, born of a consanguineous marriage, was admitted to Chattogram Medical College Hospital (CMCH) in Endocrinology Department from 24.09.25 to 16.10.25, with a history of recurrent productive cough, delayed eruption of permanent teeth, dystrophic nails and patchy scalp hair loss. According to her mother, eruption of primary dentition was delayed and several permanent teeth failed to appear. Since early childhood, she had developed brittle, thickened toenails and progressive hair thinning that later evolved into scarring alopecia. The patient experienced recurrent episodes of productive cough with purulent sputum, which worsened during seasonal changes and partially responded to intermittent courses of antibiotics and mucolytics. At 15 years of age, she noted gradual-onset hearing loss, more pronounced in the right ear.

Menarche occurred at 13 years of age, followed by regular menstrual cycles and normal secondary sexual characteristics. There was no history of heat intolerance, anhidrosis, recurrentskin infections, developmental delay, or intellectual impairment. No similar illness was reported among siblings or extended family members.

On general assessment, the patient appeared alert and cooperative but exhibited a subdued affect. Vital signs were within normal limits: blood pressure 100/70 mmHg and pulse rate 86 beats per minute. Her sexual development corresponded to Tanner Stage IV. There was no lymphadenopathy, thyromegaly, bony tenderness or mucosal discoloration.

Respiratory findings included a centrally placed trachea and bilateral rhonchi with coarse crepitations over the mid and lower lung zones, varying with coughing. Cardiovascular assessment revealed the apex beat located in the right fifth intercostal space, 9 cm from the midline along the midclavicular line—consistent with dextrocardia. Heart sounds were normal across all four auscultatory areas, with no murmurs, palpable P or epigastric pulsation.

Neurologically, higher mental functions and cranial nerves were intact, muscle strength was 5/5 in all extremities and both superficial and deep tendon reflexes were preserved.

Intraorally, several permanent teeth were absent, including molars in the left mandibular and right maxillary arches, with retention of incisors, canines and a single premolar in the right mandibular arch; Image 1 and Image 7. Gingival atrophy was evident, while the palate and tonsils appeared normal. Dermatological findings included patchy facial hyperpigmentation, Image 2, scarring alopecia, Image 3 and dystrophic nails mainly of the feet, Image 4.

Other systemic evaluations, including abdominal and musculoskeletal assessments, were unremarkable.

Laboratory Parameters

Investigation	Findings	Interpretation / Remarks
Hemoglobin (Hb)	11.5 g/dL	Within normal limits
Total WBC count	9.98*10^9/L	Normal
Platelet count	434*10^9/L	Normal
ESR	11mm/hr	Mildly elevated
Renal function tests	0.6mg/dl	Within reference ranges
Liver function tests	Normal	Within reference ranges
Serum electrolytes	Normal	Within reference ranges
OGTT	normal	
UrineR/M/E	NAD	
TSH	0.014uIU/ml,	Temporary thyroid
	0.110uIU/m,	dysfunction due
	1.89uIU/ml	to thyroiditis
Free T4 (FT4)	4.39ng/dl	
Free T3 (FT3)	2.19pg/ml	
Thyroid scan	normal	subacute thyroiditis
		(Recovery phase)
Basal cortisol	212µg/dL	Suggests cortisol excess
Plasma ACTH	37pg/ml	normal
Dexamethasone	7.90µg/dL,	Confirms abnormal
suppression test	Non-suppressible	HPA axis regulation
Chest X-ray	Dextrocardia, Image 5	Suggestive of situs inversus
HRCT Chest	Early bronchiectatic	As documented in the report
	changes, Image 6	
Abdominal Ultrasound	Situs inversus totalis, liver	Confirms visceral organ
	and gallbladder on left,	laterality anomaly
	spleen and stomach on right	
Audiometry	Clinical chronic sinusitis,	Supports ciliary
	right-sided conductive	dysfunction,
	hearing loss	
Sputum GeneXpert	Negative	No evidence of
		Mycobacterium tuberculosis
Sputum Culture &	Viridans group	Non-pathogenic flora; no
Sensitivity	streptococci isolated	major pathogenic growth
Genetic testing	Not performed	Resource limitation
Specialized ciliary studies (Electron microscopy / nasal NO)	Not performed	Not available

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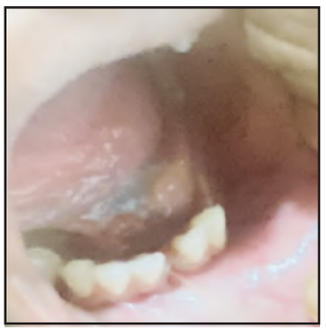

Image 1 Several permanent teeth absent, including molars in the left mandibular and right maxillary arches, with retention of incisors, canines, and a single premolar in the right mandibular arch

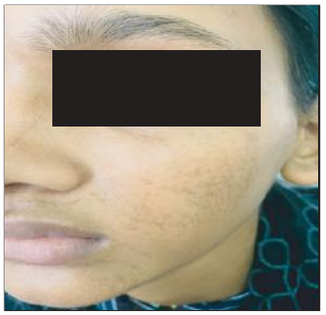

Image 2 Patchy facial hyperpigmentation

Image 3 Scarring alopecia

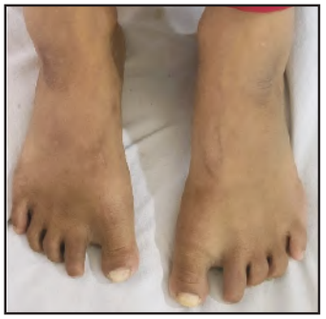

Image 4 Dystrophic nails of the feet

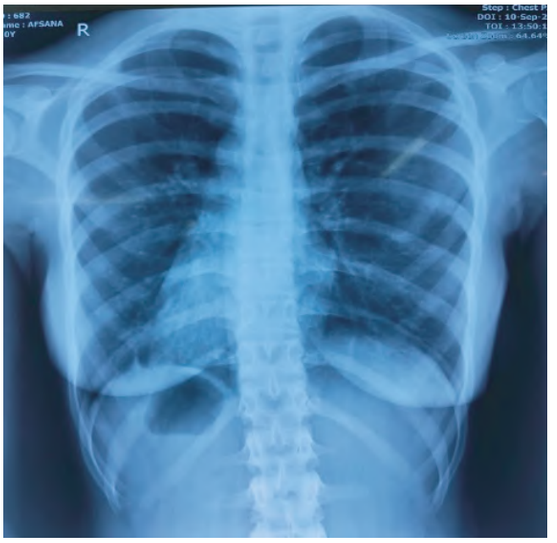

Image 5 CXR reveals
Situs Inversus Early Bronchiectatic Change

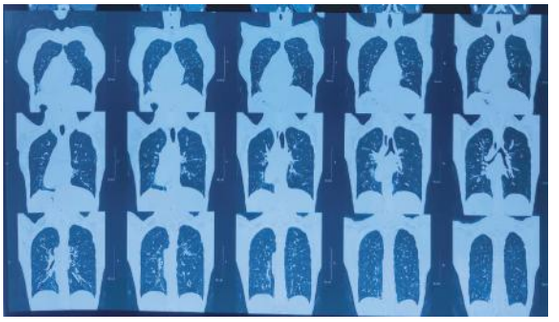

Image 6 HRCT chest \reveals

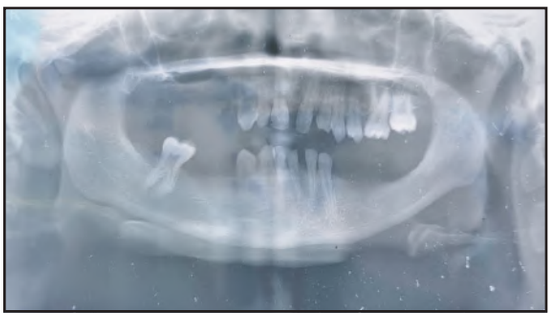

Image 7 Panoramic X-ray (OPG) reveals
Agenesis of left lower jaw and right upper jaw

Management and Follow-up

A multidisciplinary management approach was adopted:

- Respiratory care: Chest physiotherapy, bronchodilator and mucolytic therapy, intermittent antibiotics for acute exacerbations.

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- Sinus care: Symptomatic management of chronic sinusitis.
- Dermatology: Topical Benzoyl peroxide (Azelaic preparation) and emollients for facial pigmentation and nail dystrophy.
- Dental care: Prosthetic rehabilitation and restorative planning for agenetic teeth.
- Endocrinology: Monitoring thyroid function; no antithyroid therapy was required. A repeat dexamethasone suppression test has been advised after psychological stabilization to assess normalization of HPA function.
- Psychological support: Counseling and behavioral therapy aimed at reducing stress and potential HPA-axis over activation.
- Immunization: The patient received pneumococcal and influenza vaccines and was further counselled regarding further vaccination schedule, to reduce the risk of severe respiratory infections associated with impaired mucociliary clearance.
  Necessary permission was obtained from the Department before start the study.

Discussion

Kartagener's Syndrome (KS) a subset of primary ciliary dyskinesia, classically presents with the triad of bronchiectasis, chronic sinusitis, and situs inversus totalis resulting from ciliary motility defects leading to impaired mucociliary clearance and abnormal left–right asymmetry.[1,2] The current case demonstrated characteristic clinical and radiological features consistent with KS, including recurrent productive cough, bronchiectatic changes on high-resolution computed tomography, and ultrasound-confirmed situs inversus totalis.

What distinguishes this case is the coexistence of ectodermal anomalies, specifically dental agenesis, scarring alopecia and dystrophic nails, which are seldom documented in association with KS. While KS primarily involves motile cilia dysfunction, a limited number of reports have highlighted overlapping phenotypes of primary ciliary dyskinesia with ectodermal dysplasia, suggesting that certain ciliopathy-related genes may have pleiotropic effects influencing both ciliary and ectodermal development.[13-5]

In comparison with previous cases, our patient exhibited a broader ectodermal phenotype. Merrett reported congenital absence of a lateral incisor, Pawlaczyk-Kamienska et al. described dental agenesis with enamel hypoplasia and Abed et al. documented structural tooth anomalies.[5,6,7]

Our patient additionally demonstrated scarring alopecia and nail dystrophy, expanding the phenotypic spectrum of KS–ectodermal overlap syndromes.

Transient thyroid dysfunction was observed, characterized by suppressed TSH with elevated FT3 and FT4 that spontaneously normalized, consistent with subacute thyroiditis.[6,7] This mirrors prior reports where thyroid indices spontaneously resolved without pharmacologic intervention, highlighting the self-limiting nature of the condition.

Further, the patient exhibited elevated basal cortisol with normal ACTH level and absence of Cushingoid features, consistent with pseudo-Cushing's state. This pattern is described in patients with major depressive disorder, in whom HPA-axis hyperactivation leads to impaired dexamethasone feedback suppression.[8-10] A repeat dexamethasone suppression test is planned after psychological stabilization to assess normalization of HPA function.

Follow-up is scheduled every three months, including respiratory monitoring, thyroid function reassessment, and repeat dexamethasone suppression testing after improvement in psychosocial stressors.

Prognosis

With adherence to multidisciplinary care, prognosis remains favorable. Early and continued respiratory management may slow bronchiectasis progression. Spontaneous normalization of thyroid function is reassuring. However, psychological well-being and self-image remain key determinants of long-term HPA-axis regulation and overall health, highlighting the importance of continued mental health support.

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Limitation

The primary limitation of this case lies in the absence of confirmatory genetic testing to establish the definitive molecular basis linking Kartagener's syndrome and ectodermal anomalies. While the clinical, radiological and hormonal features strongly support this association, the lack of genotypic confirmation restricts our ability to identify specific gene mutations or their pathogenic correlation.

Conclusion

This case underscores the complex interplay between ciliary dysfunction and ectodermal dysplasia. The coexistence of Kartagener's syndrome with ectodermal anomalies-manifesting as alopecia, nail dystrophy and dental agenesis-widens the clinical spectrum of primary ciliary dyskinesia.

Such overlapping features highlight the need for clinicians to adopt a multisystem diagnostic approach, especially in patients presenting with recurrent respiratory infections and cutaneous or dental abnormalities. Early identification not only aids in targeted management but also prevents progressive pulmonary damage.

This case adds to the limited literature on combined ciliary and ectodermal dysfunction, suggesting that both may arise from shared developmental or genetic pathways influencing epithelial differentiation and ciliary motility.

Recommendations

- Genetic Confirmation:

Future similar cases should undergo comprehensive molecular testing, including whole-exome sequencing or targeted ciliary–ectodermal panels, to elucidate shared or overlapping mutations.

- Interdisciplinary Collaboration:

Effective management requires coordination between pulmonology, dermatology, endocrinology, and genetics to ensure holistic patient care.

- Endocrine Surveillance:

Periodic hormonal evaluation-especially of thyroid and adrenal axes-should be integrated into the long-term follow-up of patients with Kartagener's syndrome exhibiting atypical systemic features

- Longitudinal Monitoring:

Regular radiological and clinical reassessment of pulmonary status, combined with evaluation of hair, nail and dental growth, can help detect progression and tailor therapy accordingly.

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Contribution of authors

NIC-Conception citing references, drafting, critical revision & final approval.

FA-Conception, design, critical revision & final approval.

SA-Citing references, drafting, & final approval.

IAT-Citing references, critical revision & final approval.

HTS-citing references, drafting & final approval.

MF-Citing references, drafting, critical revision & final approval.

Disclosure

All the authors declared no conflict of interest.

References

1. Afzelius BA. A human syndrome caused by immotile cilia. Science. 1976;193(4250):317-319.

2. Lucas JS, Davis SD, Omran H, Shoemark A. Primary ciliary dyskinesia in the genomics age. Lancet Respir Med. 2020;8(2):202-216.

3. Cluzeau C, Hadj-Rabia S, Jambou M, et al. Spectrum of ectodermal dysplasias. Clin Genet. 2011;79(6):511-525.

4. Itin PH, Fistarol SK. Ectodermal dysplasias. Am J Med Genet C Semin Med Genet. 2004;131C(1):45-51.

5. Merrett SJ. Kartagener's syndrome: unusual dental morphology. Br Dent J. 2005;198(3):151-153. doi:10.1038/sj.bdj.4812008.

6. Pawlaczyk-Kamie ska T, Winiarska H, Kulczyk T. Dental anomalies in rare, genetic ciliopathic disorder : A case report and review of literature. Int J Environ Res Public Health. 2020;17(12):4337. doi:10.3390/ijerph17124337.

7. Abed H, Gogandi H, Almutawwif M et al. Dental management of Kartagener syndrome: A case report. Spec Care Dent. 2024;44(3):327-332. doi:10.1111/scd.12917.

8. Belvederi Murri M, Pariante CM, Mondelli V et al. HPA-axis dysregulation in major depression: A meta-analysis. Psychoneuroendocrinology. 2014;41:214-228.

9. Holsboer F. The corticosteroid receptor hypothesis of depression. Neuropsychopharmacology. 2000;23(5):477-501.

10. Carroll BJ, Curtis GC, Mendels J. Neuroendocrine regulation in depression. Arch Gen Psychiatry. 1976;33(4):425-433.