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CASE REPORT

Kartagener's Syndrome with Short Stature – A Rare Association

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Abstract

Kartagener's syndrome [KS] is a rare, autosomal recessive disease comprising a triad of situs inversus, bronchiectasis and sinusitis. Usually KS manifest in early childhood and the manifestations include chronic, thick, mucoid rhinorrhea, recurrent otitis media, wheeze, chronic wet cough with unexplained respiratory distress Most of the times clinicians would have seen many times before the diagnosis is made. Here with we are reporting a 16 year old girl who presented with recurrent infections since childhood days, diagnosed as KS at the age of 16 years with short stature due to Growth hormone deficiency.

Key Words

Kartagener's Syndrome, Recurrent Infections , Short Stature

Introduction

Kartagener's syndrome [KS] is a rare, autosomal recessive disease comprising a triad of situs inversus, bronchiectasis and sinusitis.[1] KS is classified under the group of primary cilliary dyskinesias [PCD].[1] Approximately 50% of primary ciliary dyskinesia patients have situs inversus and are classified as having Kartagener syndrome. KS is rare with frequency of 1 case per 10,000-20,000 live births years.[2]

Genome analysis has found gene mutation of *DNAH5* and *DNA11* on bands 5p15.1 and 9p13,3 respectively, are known to cause primary ciliary dyskinesia.[2] In KS, the gene mutation leads to impaired ciliary motility, which predisposes to recurrent sinopulmonary infections and infertility.[1] We are reporting a 16 year old girl who presented with recurrent infections since childhood days now diagnosed as KS at the age of 16 years with short stature due to Growth hormone deficiency.

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Case Report

A 16 year old girl presented with complaints of nasal block and mucopurulent nasal discharge since 3 months. She has been visiting many practitioners and consultants since child hood days for respiratory symptoms, nasal symptoms and not gaining adequate weight and height. She started consulting doctor at the age of 2 years for cold and running nose. She has been visiting doctor 6-7 times a year since then. At 13 years of age she was suspected to have KS, confirmation was not done in another hospital. There is no ear discharge/ expectoration /breathlessness. She attained menarche at the age of 14 years. Since 3 months she has been taking inhalers for persistent wheeze as prescribed by clinician. She is born out of consanguineous marriage, with history of 3 sibs deaths. On examination her vitals:-Temperature 99^0F, RR 18/minute, PR 84/minute, BP of 112/80 mmHg and saturation of 98% at room air. There was no cyanosis/ lymphadenopathy/clubbing. Anthropometry:- Weght 27

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kgs [less than 3rd centile], height 130 cms [less than 3rd centile] according to CDC growth chart. She was classified as short stature with under nutrition. ENT examination revealed atrophied right nasal cavity and mucopurulent discharge from left nasal cavity. Respiratory system examination revealed equal breath sounds on both sides. CVS examination revealed heart sounds heard well on right side.In view of dextrocardia, recurrent chest infections, sinusitis and height less than 3rd centile she was diagnosed as short stature with Kartagener's syndrome and investigated. Her complete haemogram, KFT, LFT were within normal limits. Chest X ray showed dextrocardia with gastric bubble on right side and liver on left side [Fig 1]. ECG showed predominantly negative P wave, QRS complex, and T wave in lead I.CT Chest and abdomen showed total situs inversus with dextrocardia. Lung parenchyma showed bronchiectatic changes in posterior basal segments of left lower lobes [Fig 1] while PNS X-ray showed, mucosal thickening in both maxillary sinuses suggestive of bilateral maxillary sinusitis [Fig 1]. Echocardiography showed dextrocardia, situs and normal chambers with normal function. Ultra sound abdomen revealed normal ovaries with few follicles seen in both ovaries and situs inversus. Thyroid profile [T3, T4, FREE T4 ,TSH] and her FSH, LH, Oestradiol were normal. Growth hormone assay following clonidine stimulation test was < 7 ng/mL suggestive of Growth harmone deficiency. MRI of brain and pituitary gland were normal. Karyotyping normal 46, XX [Done to rule out Turners syndrome in a short stature girl]

Discussion

KS manifest in early infancy and childhood, which include chronic, thick, mucoid rhinorrhea, recurrent otitis media, wheeze, chronic wet cough with unexplained respiratory distress[2,3] Most of the times clinicians would have seen more than 50 times before the diagnosis is made at an average age of 10-14 years.[2] Our child also had visited many practitioners multiple number of times for respiratory/nasal symptoms/inadequate weight and height gain and KS diagnosis was made, investigated at the age of 16 years. In KS, nasal examination reveals pale and swollen nasal mucosa, mucopurulent secretions, and an impaired sense of smell.[2] The current girl visited us with mucopurulent secretions since 3 months continuously with nasal block. In PCD also same problems occur, therefore diagnosis of PCD is frequently delayed.[3] In the present child also it may be true.

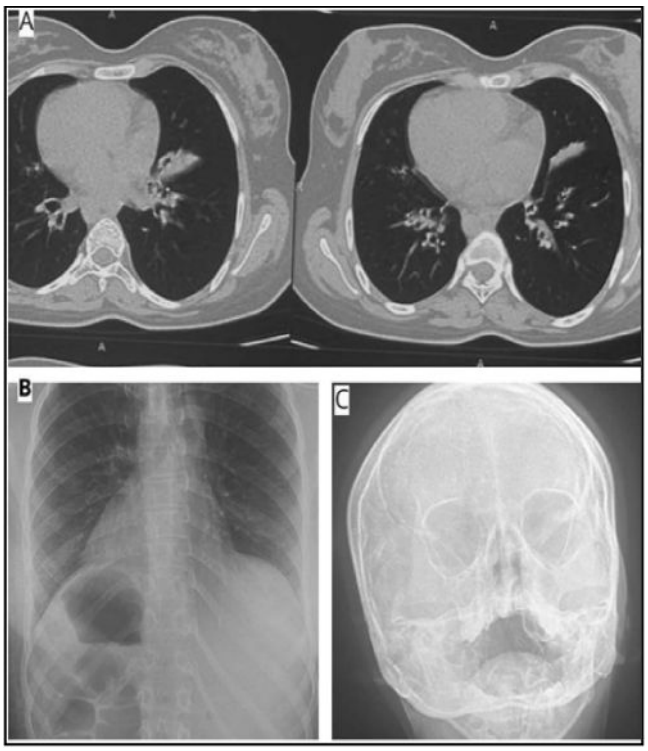

Fig. 1 (A) CT Chest: Lung parenchyma showed bronchiectatic changes in posterior basal segments of left lower lobes. (B) Chest X ray showed dextrocardia with gastric bubble on right side and liver on left side. (C) PNS X-ray showed mucosal thickening in both maxillary sinuses suggestive of bilateral maxillary sinusitis.

Sinus radiographs demonstrate mucosal thickening, opacified sinus cavities, and aplastic or hypoplastic frontal and/or sphenoid sinuses.[2] In KS, chest x-ray reveal bronchial wall thickening (earliest manifestation), hyperinflation, atelectasis, bronchiectasis, and situs inversus [dextrocardia and right-sided stomach air].[2] In KS, bronchiectasis usually involves the lower lobes, while in cystic fibrosis predominantly occurs in the upper lobes.[2] CT findings of bronchiectasis may be variable in severity, changes are much milder than in cystic fibrosis.[4] Our child had mucosal thickening, opacification of bilateral maxillary sinus. CT chest revealed bronchiectatic changes mainly in posterior basal segments of left lower lobes.

Differential diagnosis of KS includes conditions associated with bronchiectasis like foreign body, Immunodeficiency states, cystic fibrosis and Young's syndrome.[5]

In a case series by Özkara *et al*, out of 61 bronchiectasis cases, 6 had Kartagener syndrome. All

those 6 KS cases were misdiagnosed as asthma since their childhood and mistreated with bronchodilator and steroids.[6] High index of suspicion of KS is necessary for the early diagnosis.[7] Svobodová *et al* showed that PCD leads to significant growth deterioration during childhood. They concluded that the general well-being of PCD children can be compromised by the activation of mediators of inflammation during chronic suppurative airway diseases, and by chronic hypoxia.[8] In the present KS girl with short stature, decrease in height may be due to both GH deficiency and chronic infections of the sinus. Nagaraj *et al* reported Kartagener's syndrome with Retinitis Pigmentosa in a 17 years short stature girl and opined that it could be a new phenotypic presentation in PCD.[9] Similar to this report, KS with short staure may be a new phenotypic presentation.

Conclusion

Diagnosis of KS should be considered in a child with dextrocardia having features of recurrent upper or lower respiratory tract infections. Early diagnosis is crucial to avoid complications like bronchiectasis, respiratory failure, cor pulmonale or any lung parenchymal disorders, allergic bronchopulmonary aspergillosis and to improve the quality of life.

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