ISSN NO: 2770-2936
Date of Publication:06-01-2022

Nasal obstruction and productive cough in a 12 years old girl in Basrah, Iraq; the diagnosis of Kartagener syndrome is established.

Ziyad Tariq Malghooth [(1)], Haider Kadhem Saeed [(2)]
1: Respiratory physician, Basra Teaching Hospital, Basrah, Iraq
2: Otolaryngologist, Sadar Teaching Hospital, Teacher at Alzahra Medical College, University of Basrah, Basrah, Iraq
Corresponding Author:
Dr. Haider Kadhem Saeed
Alzahra Medical College, university of Basrah, Basrah, Iraq
E-mail: haiderkadhem2012@gmail.com

Abstract:

Background:

Kartagener syndrome (KS) is an autosomal recessive genetic disorder characterized by ciliary dyskinesia that causes defects in the action of ciliary movement affecting the nose, paranasal sinuses, lung and others. The syndrome comprises of triad situs inversus, chronic sinusitis with some times nasal polyposis and bronchiectasis.

Kartagener syndrome regarded as a subset of the primary ciliary dyskinesia which manifested early in life with frequent respiratory infections, frequent sinus infections, frequent ear infections, chronic nasal congestion and Sino nasal polyposis and male infertility.

Case report:

Our reported case is a 12 years old girl presented with repeated lower respiratory tract infections (Pneumonia) and nasal obstruction without family history among her siblings (one brother and three sisters). On detailed review of the patient history, she had repeated attacks of pneumonia which treated as an outpatient with antibiotics and mucolytic. She also reported chronic bilateral nasal obstruction, profuse mucoid nasal discharge, headache with diminished sense of smell. Repeated ear infections along with hearing impairment was also mentioned in the patient history. Naso-endoscopic Examination confirmed the presence of chronic rhinosinusitis with bilateral Sino nasal polyposis, bilateral tympanic membrane perforation and conductive hearing loss, bilateral lower lobe pneumonia and right sided heart location. Chest and abdominal X- with both right and left sided ECG leads and ultrasonography of the abdomen confirmed the dextrocardia and the situs inversus. Treatment in form of antibiotics, saline nasal wash and received.

Conclusion:

Kartagener syndrome is an inherited rare condition characterized by the presence of a triad of bronchiectasis, sinusitis and situs inversus. The discovery of such a case usually earlier, although some cases worldwide were discovered later than this. There is no cure for this syndrome and the treatment is mainly directed towards the symptoms of repeated chest and sinus infections and other infections in the ear.

Key words: Kartagener syndrome, immotile cilia syndrome, Basrah, bronchiectasis, dextrocardia, situs inversus

Introduction:

Manes Kartagener (1897 – 1975) an Austrian-Swiss physician was the first to report the triad of situs inversus, chronic sinusitis and bronchiectasis in 1933, which is now took his name. [1] Other names are: immotile cilia syndrome and Siewert\Kartagener syndrome.

Kartagener's syndrome (KS) is inherited via an autosomal recessive pattern. Its incidence is about 1 in 30,000 live births. [2] This genetic disorder causes a defect in the action of the cilia lining the respiratory tract

ISSN NO: 2770-2936
Date of Publication:06-01-2022

including the paranasal sinuses, middle ear and Eustachian tube and fallopian tube. Patients usually present with chronic recurrent rhinosinusitis, otitis media, pneumonia, and bronchiectasis caused by pseudomonas infection. Situs inversus can be seen in about 50% of cases. [3] Male individual who had this syndrome are usually infertile because of immotile spermatozoa caused by the numerous ultrastructural defects in respiratory cilia and sperm tail. [4]

The main pathophysiology for KS is a genetic defect in the dynein arms of the motile cilia of the respiratory tract as shown in figure [1], ear canal and others caused by mutations involve the heavy (dynein axonemal heavy chain 5) or intermediate (dynein axonemal intermediate chain 1) chain dynein genes in ciliary outer dynein arms. [5]

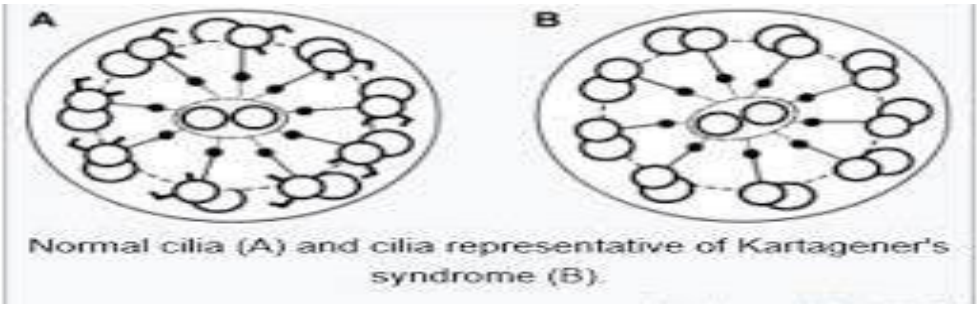

Figure [1]: a diagram representing the difference between normal cilia and cilia in case of Kartagener syndrome from K. Takeuchi et al. [6]

The Main anomaly is seen in the respiratory tract as recurrent pneumonia caused by mucus statis in the bronchi with productive caught represent most presenting symptom. Nasal obstruction, congestion and polyposis in those patients is an associated finding with features of anosmia, snoring, nasal speech and chronic nasal discharge. [6]

Case Report:

A female patient, seen in figure [2], consult the respiratory clinic with her mother complaining from prolong and repeated history of productive cough and repeated chest infections since infancy. On history, the patient report that these infections occur frequently as low grade fever, productive cough, whizzy chest and some shortness of breath. The rest of demographic data is illustrated in table [1].

Figure [2]: The patient with the broad nasal bridge.

ISSN NO: 2770-2936
Date of Publication:06-01-2022

Table [1]: the general demographic data of the case

	General demographic data	Our patient
1	Gender	Female
2	Age	12 years
3	Occupation	household
4	Residency	Rural
5	Siblings	4 [1 brother younger and 3 sisters older]

The general examination of the patients reveal normal vital signs. Chest examination revealed crackles in lower and middle lung lobes with occasional rhonchi. Pulmonary function test for the patient reveal obstructive curve. The heart sound were picked on the right side of the chest and it was normal. Chest X-ray reveled bilateral bronchial dilatation and consolidation in upper, middle and lower lung lobes with dextrocardia.

The patient's history reveal nasal problems so otolaryngological consultation was asked. The patient reported chronic nasal obstruction, mucoid nasal discharge, anosmia associated with some attacks of epistaxis. She also reported repeated ear discharge and some hearing impairment. Examination revealed broad nasal bridge, mouth breathing, nasal speech and negative cottle test. Naso-endoscopic examination revealed congested nasal mucosa, hypertrophied both inferior turbinate and sino-nasal polyposis. Ear examination showed bilateral tympanic membrane perforation with negative Rinne test and weber lateralized to the left. Pure tone audiometric examination revealed bilateral mild to moderate conductive hearing loss and tympanometric examination revealed type B tympanogram bilaterally. Oral, laryngeal and neck examination were normal. Abdominal examination revealed left sided liver with no tenderness or masses. So, the diagnosis of Kartagener syndrome is taken into consideration.

Radiological investigation in form of Computed Tomography (C-T scan) of the nose, paranasal sinuses, temporal bone and the chest confirmed the diagnosis of bronchiectasis, chronic rhinosinusitis with nasal polyposis and chronic supportive otitis media with mastoiditis as seen in figure [3].

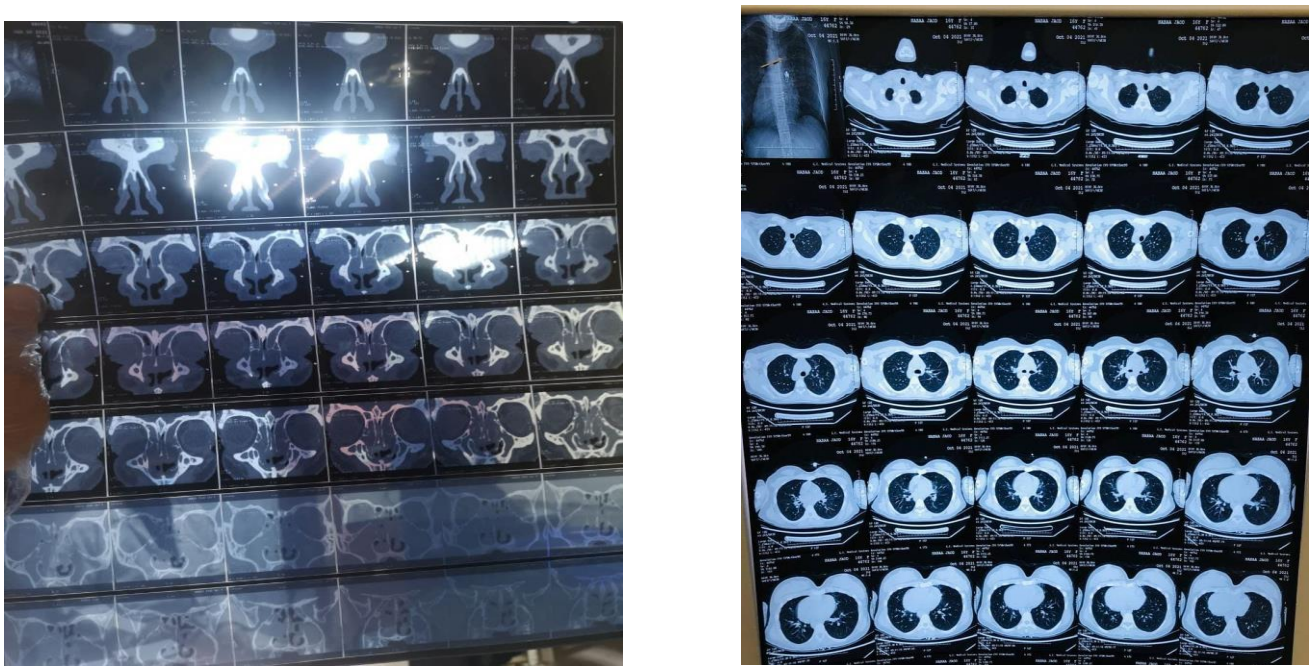

Figure [3]: C-T scan images of the chest, nose and paranasal sinuses.

Ultrasonography of the abdomen confirmed left sided liver and gall bladder, right sided spleen (situs inversus). ECG and Echocardiography confirmed the dextrocardia with normal cardiac function. A summary of all finding is listed in table [2].

No access to genetic study analysis or transmission electron microscope available in our province in order to confirm the diagnosis of ciliary abnormalities.

ISSN NO: 2770-2936
Date of Publication:06-01-2022

The triad of bronchiectasis, situs inversus and sinusitis made the diagnosis of immotile cilia syndrome or Kartagener syndrome. Treatment in form of physiotherapy for the chest, mucolytic, antibiotic, steroid inhaler, steroid nasal spray, saline nasal douche and tonics were described and regular follow up.

Table [2]: the result of investigations for our patient.

	Investigation sent	Result
1	Pulmonary function test (PFT)	Obstructive ventilatory defect with decreases in the ratio of forced expired volume in 1 second to forced vital capacity, reduced forced expired volume in 1 second, and a reduced forced expiratory flow 50%.
2	Chest X-Ray (PA view)	Dextrocardia bilateral bronchial dilatation and consolidation in upper and lower lung lobes
3	Pure tone audiometry	Both ear show conductive hearing loss of mild to moderate in severity
4	Tympanogram	Both ear show type B tympanogram with increased canal volume
5	Echocardiography	Dextrocardia with mirror image of normal heart and normal cardiac function
6	US of Abdomen	Liver and gall bladder are normal in shape and feature but on the left side with spleen on the right side (situs inversus)
7	C-T scan of chest	Bronchiectasis changes with tree in bud sign of endobronchial infection
8	C-T scan of nose, paranasal sinuses and temporal bone	Mucosal thickening in both maxillary, ethmoidal and frontal sinuses with sino-nasal polyposis, bilateral middle ear opacification with mastoidites.

Discussion:

Repeated chest infections in otherwise healthy child should rise the suspicion of an anatomical or physiological abnormalities in the respiratory tract. Detailed history and precise examination should pick up any serious pathology like Asthma, bronchiectasis, cystic fibrosis, immunodeficiency syndromes and congenital abnormalities of the respiratory tract or other where in the body like Kartagener syndrome. [7]

Siewert first described the combination of situs inversus, chronic sinusitis, and bronchiectasis in 1904. However, Manes Kartagener first recognized this clinical triad as a distinct congenital syndrome in 1933 and the syndrome named after him. [8]. since then, a lot of Kartagener syndrome cases had been reported worldwide. Most of these cases were children, as the diagnosis is discovered early in life. But several cases in adult or even elderly has been reported. [3][6][9][10]

Kartagener syndrome is a rare, ciliopathic, genetic disorder caused by defect in the gene responsible for ciliary movement. The major clinical manifestations are: (1) in the respiratory tract, immotility or dyskinetic beating of cilia in epithelial cells impairs the mucociliary clearance, leading to recurrent infections, sinusitis and bronchiectasis; (2) in the urogenital tract, infertility occurs in some patients because of the dysmotility of sperm tails; (3) mirror reversal of body organ positioning (situs inversus) is seen in approximately in 50% of patients. [11]

Our case, a 12 years old female presented with long history of repeated chest infections and nasal obstruction since childhood. Examination and investigation confirmed the presence of bronchiectasis, sinusitis with nasal polyposis and situs inversus. This triad made the diagnosis of Kartagener syndrome in mind. Our finding of this case is similar to the findings of Sanjay Gupta et al [1], Abilo Tadesse et al [2], Nedaa Skeik et al [3], Margaret W Leigh et al [5], Barthwal et al [9], Bi Cui Liu et al [10], DC Arunabha [11] and Hailu SS et al [12]. All of the above mentioned authors reported similar case presentation with different in time of

ISSN NO: 2770-2936
Date of Publication:06-01-2022

presentation or gender and prove their diagnosis of Kartagener syndrome by genetic analysis or by bronchial wash biopsy or sinus mucosa biopsy examined by transmission electron microscope. Ali MS et al [13] performed laparoscopic cholecystectomy in a Kartagener syndrome patient in Baghdad, Iraq. The case was diagnosed based on clinical triad of situs inversus, bronchiectasis and sinusitis without biopsy or genetic analysis. We have no genetic study in our governorate to confirm the exact gene mutation.

Treatment in form of hospital admission, intravenous antibiotics, chest physiotherapy, mucolytic, steroid inhaler and saline nasal douche was prescribed. The patient is doing well at time of writing this paper on steroid inhaler and saline nasal douche with steroid nasal spray. The nasal congestion is better, cough occur less frequently and she is able to tolerate daily activity. Regular follow up for the patient planned to prevent any deterioration.

Conclusion:

The presentation of child with repeated chest infections should rise the suspension of serious condition affecting the airways. Chest infection, sinusitis and situs inversus rise the suspension of diagnosing Kartagener syndrome. Although, we do not have the necessary genetic analysis to prove the diagnosis of this syndrome in this child, all the clinical, audiometric and radiological assessment had reached us to the diagnosis of Kartagener syndrome in this case.

Acknowledgement:

The authors would like to thanks Dr. Jwad Attia and Dr. Hisham Jebran for their cooperation in following of this case.

Funding:

No funding received

Conflict of interest:

None

Ethical approval:

Verbal consent from the family had been taken to share and discuss this case.

References:

1. Gupta S, Handa KK, Kasliwal RR, Bajpai P. A case of Kartagener's syndrome: Importance of early diagnosis and treatment. *Indian J Hum Genet*. 2012; 18(2):263-267.
2. Tadesse, A., Alemu, H., Silamsaw, M. *et al*. Kartagener's syndrome: a case report. *J Med Case Reports* **12,** 5 (2018).
3. Skeik N, Jabr FI. Kartagener syndrome. *Int J Gen Med*. 2011; 4:41-43. Published 2011 Jan 12.
4. Sammuel I, Kartagener syndrome with normal spermatozoa, JAMA, 1987; 258: 1329-30.
5. Leigh M., Pittman, J., Carson, J. *et al.* Clinical and genetic aspects of primary ciliary dyskinesia/Kartagener syndrome. *Genet Med* **11,** 473–487 (2009).
6. Mohammed Raoufi, Hicham Sator, Jawad Lahma, Ali El Ayoubi, Sophia Nitassi, Abdelilah Oujilal, Mohammed Anas Benbouzid, Leila Essakalli, Hanane Elouazzani, Ismail Abderrahmane Rhorfi, Ahmed Abid, case of Kartagener syndrome with rhinolalia clausa. Pan African Medical Journal. 2016;23:159
7. Michael Barkham, Gillian E Hardy, Counselling and interpersonal therapies for depression: towards securing an evidence-base, *British Medical Bulletin*, Volume 57, Issue 1, March 2001, Pages 115–132.
8. Kartagener M. Zur Pathogenese der Bronchiektasien. Bronchiektasien bei Situs inversus viscerum. *Beitr Klin Tuberk.* 1933;83:489–501
9. Barthwal M S, Kartagener's Syndrome In A Fertile Male – An Uncommon Variant, Lung India: Jul–Sep 2006 - Volume 23 - Issue 3 - p 123-125
10. Liu BC, Huang TX, Liu CT. Kartagener Syndrome. Am J Med Sci. 2020 Jun; 359(6):390-391.

ISSN NO: 2770-2936
Date of Publication:06-01-2022

11. Arunabha DC, Sumit RT, Sourin B, Sabyasachi C, Subhasis M. Kartagener's syndrome: a classical case. *Ethiop J Health Sci*. 2014; 24(4):363-368.
12. Hailu SS, Amerga ED, Gorfu Y, Zewedneh D. KARTAGENER'S SYNDROME: A CASE REPORT. Ethiopian Medical Journal. 2016 Apr; 54(2):91-94.
13. Ali MS, Attash SM. Laparoscopic cholecystectomy in a patient with situs inversus totalis: case report with review of literature. *BMJ Case Rep*. 2013; 2013:bcr2013201231. Published 2013 Oct 7. doi:10.1136/bcr-2013-201231