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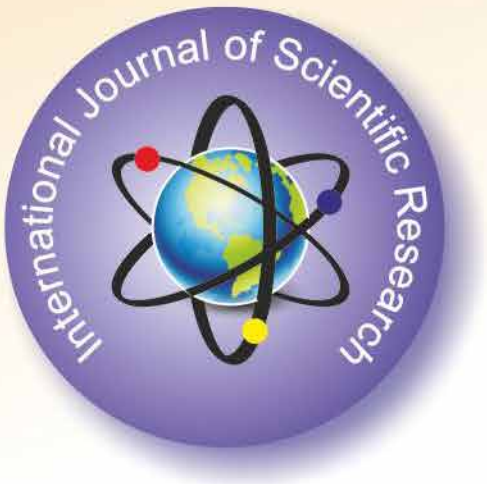

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INDEX

Sr. No.	Title	Author	Subject	Page No.
1	Ionic composition of a freshwater lake and its implications on aquaculture	Dr. Shankar P. Hosmani	Biotechnology	1-2
2	Growth and Performance of Mutual Fund Industry in India	Dr. M. K. Maru	Commerce	3-4
3	Waste Management: A New Paradigm of Contemporary Business	Dr. Vipul Chalotra	Commerce	5-6
4	Rural Financial Services in J&K (A study in the field of financial services sector development)	Tarsem lal	Commerce	7-8
5	Banyan, the National Tree of India	Dr. J.K. Sehgal	Commerce	9-10
6	Impact of Online Marketing on Customers with Special Reference to Coimbatore City	Dr. R. Ganapathi	Commerce	11-15
7	Customers' Attitude towards Housing Loan With Reference to Commercial and Rural Banks	Dr. R. Ganapathi, Mrs. B. VIDYA	Commerce	16-23
8	Consumer Behaviour towards Broiler Chicken Retail Stores With Reference to Madurai City	P. Easwaran, J. Gnanadevan, Dr. R. Ganapathi	Commerce	24-30
9	Data Security and Protection in Cloud Computing	Shameena Begum, V. Ratna Vasuki, K.V.V.Srinivas	Computer Science	31-34
10	Foreign Direct Investment in India – An Explanatory Study	Dr. K.Madhu Babu	Economics	35-38
11	Growth - Saving Causality in India: A Cointegration Analysis	Dr. Shradha H. Budhedeo	Economics	39-42
12	Constitutional perspectives on Labour Wages in India	Dr. Shankar Ambhore, Dr. Dilip Arjune, Manish Parshuram Pawar, Dr. Ashok Shankarrao Pawar	Economics	43-45
13	A Critical Study of Special Economic Zones in India	Dr. Shankar Ambhore, Dr. Dilip Arjune, Manish Parshuram Pawar, Dr. Ashok Shankarrao Pawar	Economics	46-48
14	Industrial Relations - Settlement of Disputes in India	Dr. Shankar Ambhore, Dr. Dilip Arjune, Manish Parshuram Pawar, Dr. Ashok Shankarrao Pawar	Economics	49-50
15	AMLA - ITS MEDICINAL USES	Manisha Gaur	Economics	51-52
16	The Role Of Total Quality Management In Higher Education	Ramesh B. Sakhiya	Education	53-55
17	Perceived Competencies Of Graduate Teacher Trainees In The Intensive Teaching Practice [I T P] Session	Dr M. Parimala Fathima, N.Sasikumar, M. Panimalar Roja	Education	56-58
18	Uchch Siksha Ki Rah Men Dushvariyan	Dr. Anup Chaturvedi	Education	59-60
19	Fault Diagnoses of Rotating Machinery with Advance Signal Processing Methods	Prof. Divyang H. Pandya, Prof. Ankit A. Darji	Engineering	61-63
20	A Hybrid Neural Network Approach for Wind Speed Prediction	S.N Deepa, K.gnana Sheela	Engineering	64-67
21	A Study on Phishing: Preventions and Anti-Phishing Solutions	V.Karamchand Gandhi, Prof R.Senthil Kumar	Engineering	68-69
22	The Killari 1993 Intracratonic Earthquake- a Comparative Study	S.S. Patil, K.L. Karkare, I.B. Ghorade	Environment	70-72
23	Cosmic Plants as Alternative Medicine	Dr. Sneh Harshendra Sharma	Environment	73-77

24	Green Initiatives for Reducing Carbon Footprint	Dr Mahalaxmi Krishnan	Environment Science	78-79
25	Prediction of Urban Sprawl in Hyderabad City using Spatial Model, Remote Sensing and GIS Techniques	S. Indhira Gandhi, Dr. V. Madha Suresh	Geography	80-81
26	Tectono-Provenance and Reservoir Rock Characteristics of the Tipam Sandstones in Parts of Upper Assam Basin	Dr. Pradip Borgohain	Geology	82-84
27	(Jansanchar Aur Bharatiya Samaj)	Dr Subodh Kumar	Journalism	85-86
28	An Overview of Industrial Disputes Settlement Authorities in India	Manish Parshuram Pawar, Dr. Ashok Shankarrao Pawar	Law	87-88
29	Innovative Method of Role Play for Developing English Language Teaching and Learning	K Rajkumar	Literature	89-91
30	Impact of Workers Participation in Management on Industrial Relations	Anuradha Averineni	Management	92-93
31	Consumers Preferences , Behaviour and Satisfaction with respect to banking services quality in Ghaziabad(NCR Region)	Prof(Dr.)H. P. Pandey, Mr. Ashish kumar Singh	Management	94-96
32	Factors Influencing Employee Branding in Higher Educational Institutions: A Special Reference to Management Institutions in Virudhunagar District in Tamilnadu	Jegadeeswari. Mani, Dr. S. Franklin John S.	Management	97-98
33	Evaluation of Service Quality in Internet Banking: An Empirical Study in Coimbatore	Ms. R. Gokilavani, Dr. R. Ganapathi	Management	99-101
34	To Study the Relationship Between Gender & Banking Preferences of Management Graduates at Ibmrd, Ahmednagar	Rajendrasingh Pardeshi, Gadekar Vithal Laxman	Management	102-103
35	FCB model of Advertising Strategy	Prof. Arvind Rathod	Management	104-107
36	Assessing Beneficiary Satisfaction with Service Delivery of Non Governmental Organizations (NGOs)	Dr Papori Baruah, Bhaskar Jyoti Barthakur	Management	108-111
37	Current Trends in Human Resource Management	Dr. Kalyani Kenneth, Mrs.R.Aruna jayamani	Management	112-113
38	“Indian Banking – A Future Ahead”	Haresh B. Barot	Management	114-116
39	Financial Inclusion-Banking Services to the Common Man	Dr. M. Venkata Subba Reddy, Mr. M.s.udaya Banu	Management	117-118
40	A study of Service Marketing Mix w.r.to b-schools in Mumbai	Dr. Balaji S. Mudholkar	Management	119-120
41	A Study on the Customers Opinion on the Benefits of the Credit Cards Around Combatore District	Mrs. G. Murali Manokari	Management	121-123
42	A Study on the Job Satisfaction of the Employees at Sri Kannan Departmental Stores, Coimbatore	Mrs. G. Murali Manokari, Mrs.r.kanaka Rathinam, Mr. G. Lenin Kumar	Management	124-126
43	Foreign Direct Investment In Indian Retail Sector: A Critical Evaluation	Dr. Raghavendra Dwivedi, Ram Kumar	Management	127-128
44	Emerging Challenges to Cyber Security-Internet Monitoring with Specific reference to National Security	Triveni Singh	Management	129-131
45	An Empirical Study of Consumer Impulse Buying Behavior in domestic Markets (special reference to Ahmednagar, (M.S) India.)	Gadekar Vithal Laxman	Marketing	132-135
46	Insomnia and the performance of general population: Results from the Insomnia Survey	Miss Ketaki Sathe, Dr G S Shekhawat	Medical Science	136-137
47	Transition in Human Resource for Health: Challenges Ahead	Dr. Pawan Kumar, Dr. Abdul Majeed Khan	Medical Science	138-139

48	Kartageners Syndrome- A Case Report	Dr. Ramakrishna Ghubde, Dr. Archana Shekokar	Medical Science	140-141
49	Perceptual challenges in auditory neural processing in neurodegenerative conditions like Fredereich Ataxia	Mr. Ayas Muhammed, Ms. Archana, Dr. Rajashekhar	Medical Science	142-143
50	Transient Auditory Dysynchrony Due to Non-Maturational Causes Evidenced by ABR – A Case Report	HariPrakash. P, Sangeetha. G, Bhargavi P.G	Medical Science	144-146
51	Study on Sphenoid Sinsuses Variants in Magnetic Resonance Imaging of South Indian Population	Suresh Sukumar, Sushil Yadav	Medical Science	147-148
52	A Study to Find out the Prevalence and Effectiveness of Occupational Therapy Intervention for Pain and Activity Performance in Mobile Users with Risk of Repetitive Strain Injury	KR.Banumathe, V.Guruprasad, Leena Ann Lukose	Medical Science	149-151
53	Modified Falls Behavioral Scale for Indian Community Dwelling Older Adults	V.Guruprasad, Sebestina A D'Souza, KR.Banumathe	Medical Science	152-154
54	The Essence of Employees Training and its Impact on the Work Force in an Industry	Dr. Mohan Singhe	Organization Behavior	155-156
55	Scientific Behaviourism of Watson and Hull : A Philosophical Perspective	Dr. Jatinder Kumar Sharma	Philosophy	157-158
56	The growth of manganese oxide thin films by spray pyrolysis technique	M.Sudha, P.Duraisamy	Physics	159-161
57	Terrorism and Competitive Terrorism in India	S. Sreejith, P. Sakthivel	Political Science	162-164
58	Kuposhan Se Karahta Bachpan	Dr. Anup Chaturvedi	Social Science	165

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Research Paper

Kartageners Syndrome- A Case Report

Medical Science

KEYWORDS : Primary ciliary dyskinesia (PCD), Kartagener's syndrome

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ABSTRACT

Kartageners syndrome is a rare disorder characterized by the triad of chronic sinusitis, bronchiectasis male infertility & situs inversus. It is classified under the group of disorders called primary ciliary dyskinesias. It is a genetic disorder with an autosomal recessive mode of inheritance . Special cells that are abnormal cells in respiratory tract lead to problems. It is caused by a defect in the structure of cilia, due to which ciliary movement, and consequently, its function, are impaired.

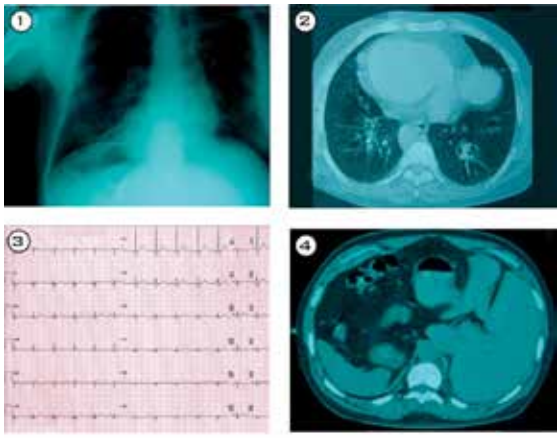
Introduction

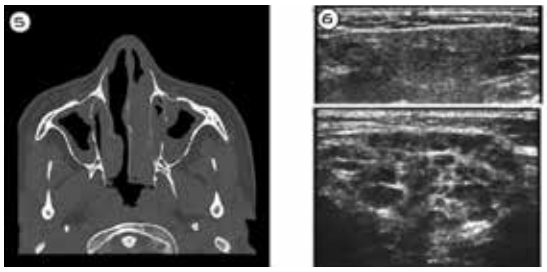
The human respiractory tract is lined by special cells that have hair like projections on them called cilia .the cells sweep inhaled molecules of dust ,smoke ,bacteria out of the respiratory tract . In primary ciliary dyskinesia ,the cells are defective & don't work properly .This means that mucus & bacteria in the lungs cant be expelled out ,as a result frequent lung infections develop. It is characterized by the classic triad of situs inversus, bronchiectasis and sinusitis. The condition was first described by Siewert in 1904 but details of the condition were given by Manes Kartagener in 1933 and it is known by his name ever since.

The primary defect is abnormal function of cilia, which causes immotile cilia syndrome. The immotile cilia cause recurrent sinopulmonary infection. KS was seen in consanguinity especially first cousin Patients with the Kartagener syndrome may have anosmia The sperm of males with Kartagener syndrome are immotile, which is the same as every cilliary cell in the body. The spermatozoa have fertility capacity and with using assisted reproductive technique, they will fertilize the egg. Women with Kartagener syndrome usually are fertile, although eggs are moved down in fallopian tube by cilia. So using **in vitro** fertilization in the management of such patients is a possibility 1 .

Case report

A 29 years old man presented with cough & expectoration 7days .since birth he had recurrent episodes of respiratory tract infections .Symptoms Chronic sinus infection,Frequent lung infections such as pneumonia & bronchitis .Bronchiectesis ,ear infectionsn examination apex beat was in the sixth right intercostals space with heart sounds heard on the right side of the chest .ECG (fig3) showed evidence of dextrocardia (fig1) Cardiac apex ,along with aortic arch & gastric bubble on chest radiograph were seen on the right side & bilateral bronchiectasis.(fig2)High resolution CTof chest & upper abdomen (fig2,4) showed bronchiectasis with situs inversus totalis. Radiograph & CT of sinuses(fig5) showed mucosal thickening in maxillary sinuses & hypoplastic frontal sinuses . fibreoptic bronchoscopy showed transposition of right & left bronchus .Semen analysis-done thrice showed decreased sperm viability with no motility.





Clinical findings

1. Chest radiograph showing dextrocardia with gastric bubble under right diaphragm. 2. High-resolution CT of chest showing bronchiectasis. 3. Electrocardiogram showing dextrocardia with tall R waves in lead V1 and absent R in V6. 4. High-resolution CT of the abdomen showing situs inversus with spleen on right side and liver on the left 5 chronic sinusitis 6 usg abdomen

DISCUSSION

PCD is a rare genetic disorder with an autosomal recessive mode of inheritance It is caused by a defect in the dynein arm structure of cilia,due to which ciliary movement, and consequently its function, are impaired. Ciliary movements are responsible for the rotation and orientation of internal organs in the 10th to 15th days of gestation. In PCD the underlying ciliary dysfunction causes incomplete rotation or malrotation of one or many internal organs, most commonly the heart 2,3. More commonly, a right-sided heart (dextrocardia) exists along with malrotation of the other internal organs, namely: lungs, liver, spleen, kidneys and intestines (situs inversus)About 50% of PCD patients develop situs inversus and KS, which has been classically described as a triad of dextrocardia, sinusitis and bronchiectasis, and male infertility; the incidence of KS is estimated to be around 1:15000 with variable penetrance, and phenotypic differences have been observed because the underlying genetic mutation has a pleiotropic effect Researchers have found possible explanations for the inflammatory changes in PCD, from Ig A deficiencies4 .exact mechanism remains poorly understood5,6In present study, a 29-years-old man was described with recurrent chronic sinusitis and bronchitis, infertility. Right-sided pancreas and left-sided liver were visualized in sonography. Chest X-Ray confirmed dextrocardia with bronchectasis. Also, the signs of chronic sinusitis were seen in the sinuses graphy. Therefore, Kartagener syndrome was diagnosed for this patient. CT thorax may further delineate malrotation, and bronchiectasis, Proper diagnostic tools are required for the definitive diagnosis of PCD. Screening tests like saccharin clearance test are simple and cheap7, but may be cumbersome in children, time consuming, and most importantly, do not differentiate between primary and secondary ciliary dysfunction.Nasal nitric oxide measurement is a rapid and reliable method for diagnosing PCD8 but is not widely available in all centres and all countries. Recent advances in genetic research have confirmed mutations of DNA H 5 and DNA I 1 9. At present, gene testing and EM are out of reach formany patients of PCD .Medical management for these patients consists of effective control of infections with antibiotics. Immunization

is also advised, Physical exercise and breathing techniques are recommended. Positive expiratory pressure devices are helpful and regular follow-up is strongly advised10,11. Bronchiectasis, pneumonia and other pulmonary complications like collapse and fibrosis are preventable to a large extent;

Conclusion

Kartagener's syndrome is a rare condition that we come across. Association of infertility, dextrocardia and bronchiectasis According to heterogeneity of this disease, genetic evaluation of it is very expensive and usually useless. Genetic counselor recommended this couples to have a baby because the pattern was suggested according to their pedigree is autosomal recessive. The sonography could show the position of internal organ and situs inversus in fetus at 3 months of age, which could help to diagnose Kartagener syndrome in offspring.

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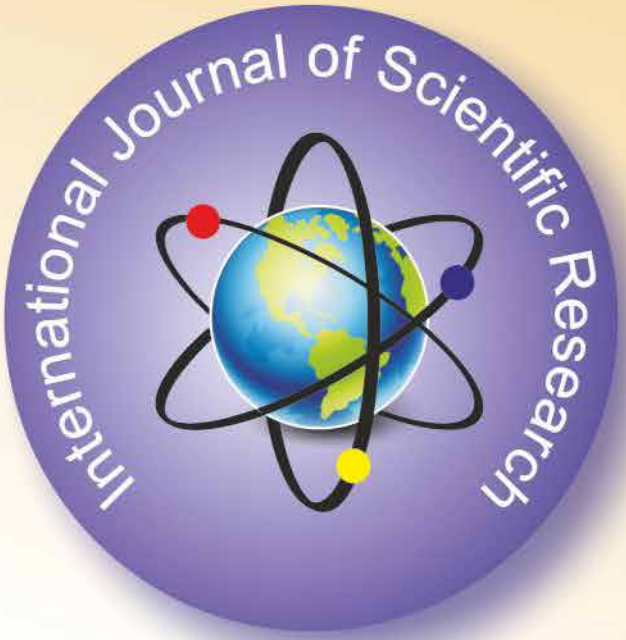

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