Case Report

Pediatric Education and Research
Volume 11 Number 2, May – August 2023
DOI: https://dx.doi.org/10.21088/per.2321.1644.11223.4

Learning Disability in a Case of Primary Ciliary Dyskinesia: A Case Report

Jagdish Taur[1], Praveen Unki[2], Santosh Kondekar[3], Suraksha Sharma[4], Qudsiya Ansari[5], Surbhi Rathi[6]

How to cite this article:

Jagdish Taur, Praveen Unki, Santosh Kondekar *et al.*/Learning Disability in a Case of Primary Ciliary Dyskinesia: A Case Report/Pediatr Edu Res. 2023;11(2): 77-80.

Abstract

Primary ciliary dyskinesia (PCD) is a genetic disorder of cilia structure, function and biogenesis leading to chronic infections of the respiratory tract, due to congenital impairment of mucociliary clearance. Mutation in genes coding components of cilia can result in primary ciliary dyskinesia.

Learning disabilities are the disorders that affect the acquisition, retention, comprehension or the application of verbal and or nonverbal information.

Our patient presented with recurrent respiratory tract infection requiring nebulization with antibiotics treatment on Out patient department basis. Child's Radiological investigation and Genetic study were planned and done which later confirmed the diagnosis. Child also had poor Scholastic performance along with behavioral issues for which detailed psychiatric evaluation was done which was suggestive of border line intellectual deficit with learning disability and Attention Deficit Hyperactive Disorder (ADHD) for which child was started on treatment along with behavioral therapy.

Keywords: Primary ciliary Dyskinesia; Learning Disability; Genetic Study.

INTRODUCTION

Primary ciliary dyskinesia (PCD) is usually an heterogeneous genetic condition characterized by structure, function and biogenesis leading to chronic infections of the respiratory tract.[1] The defect can be in the any proteins needed for making functional cilia, different clinical manifestation can be seen as per defect. Repeated infection can cause damage to the lungs and airways leading to development of bronchiectasis.

In some cases, as primary cilia are important in embryonic development of the central nervous system such as neuronal progenitor cell proliferation and generation of neurons in various parts of brain as well as migration.[2-4] Also, some people affected with PCD can have situs inversus in which the position of internal organs in the body are reversed from normal.[5]

Author's Affiliation: [1,4]Junior Resident, 3Associate Professor, [2,5]Assistant Professor, [6]Professor & HOD, Department of Pediatrics, Topiwala National Medical College & BYL Nair Charitable Hospital, Mumbai 400008, Maharastra, India.

Coressponding Author: Praveen Unki, Assistant Professor, Department of Pediatrics, Topiwala National Medical College & BYL Nair Charitable hospital, Mumbai 400008, Maharastra, India.

E-mail: praveenu3@gmail.com

Received on: 03.04.2023

Accepted on: 17.04.2023

Learning disabilities refer to group of disorders that affect the acquisition, retention, organization, comprehension or the application of verbal and or nonverbal information.[6]

Learning disability is condition which affect the information sent, received & processed by brain. They are of various type and are due to problem in brain development and cause difficulty in reading, writing, understanding one or more concepts and or subjects.[7]

CASE

A 12-year-old male child with a history of recurrent respiratory tract infection with borderline intellectual deficit with difficulty in school performance. Child had significant birth history with pneumonia on day of life 3 requiring mechanical ventilation and prolonged NICU stay. Later on, child had 4 admissions requiring oxygen therapy and intravenous medications for respiratory tract infections with recurrent episodes of cough, cold requiring treatment on out patient department (OPD) basis. There is a history of delay in achieving milestones for the child along with hyper activity and learning disability. Currently child has poor academic performance suggestive of intellectual deficit.

Radiological investigations suggestive of situs in versus total is with HRCT suggestive of mucus secretion within segmental bronchi and mild bronchiectasis of left middle lobe and posterobasal segment of left lung lobe forming consolidation likely secondary to mucoid impaction. BERA suggestive of moderate loss of hearing at high frequency. Considering child's presentation and investigation possibility of PCD suspected. Genetic study for PCD was sent and child was confirmed to have PCD With mutation in gene CCDC103/gene with pathogenic variant C461A>C.

Child had difficulty in concentrating, aggressive behavior which subsided after initiation of treatment. Along with difficulty in learning mathematics and science leading to poor scholastic performance, detailed evaluation of the child was done and was started on behavioral therapy for learning disability and ADHD.

DISCUSSION

Primary ciliary dyskinesia is a recessive genetic disease with more than 30 different genetic mutations that affect ciliary development have been identified.[8] Males and females are equally affected. Typically, first symptoms occur at a very early age. Newborns with PCD often suffer from respiratory distress and require oxygen therapy for multiple days.[9] As the child gets older, they have frequent respiratory tract infections.[10] Detailed history and clinical examination along with imaging test are helpful for diagnosing PCD but most helpful being the genetic testing.

Treatment for PCD usually focuses on improving lung function and limiting disease progression.

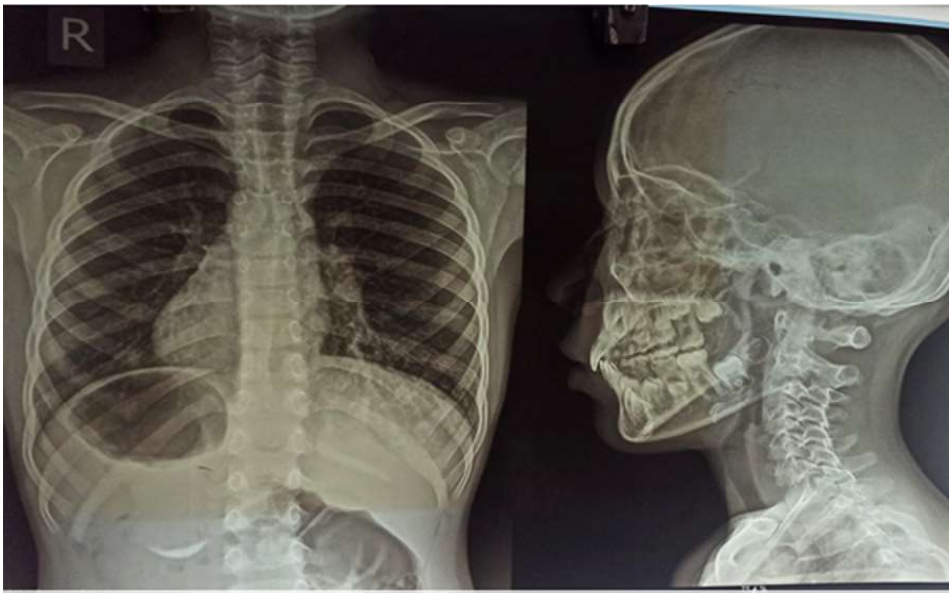

Fig. 1: Chest X ray suggestive of Situs inversus Totalis.

Antibiotics can be used for lung or sinus infections.[11]Airway clearance methods including coughing and breathing techniques along with chest physical therapy can help to loosen the mucus from the lungs.[12]

PCD may lead to severe lung disease requiring oxygen treatment or breathing assist devices during strenuous activities, and if lungs are severely affected can require lung transplantation.[13-15]

The etiologies of Learning disorders can be influenced by hereditary and or environmental factors and are not precipitated by deficit in sensorimotor functioning. They are nearly twice common in the children with chronic illness as compared to healthy children. Risk factors include family history of learning disorder, poverty, prematurity and other neuro developmental disorders.[16] Most common being Dyslexia. They often coexist with disorders like oppositional defiant disorder, attention deficit hyperactive disorder, anxiety and obsessive compulsive disorder.[17]

Evaluation of learning disorders done by first excluding the organic and functional differential diagnosis. Once the other differentials are ruled out diagnostic guidelines set out by the Diagnostic and Statistical Manual of Mental Disorders (DSM) along with relevant history of patient taken and wide varieties of test.

The most common treatment for learning disability is special education, detailed diagnostic evaluation assessing the child's academic and intellectual potential with basic approach is to teach learning skill by building on child's abilities and strength while correcting and compensating for disabilities. Multidisciplinary approach by educators, physician, speech therapist, psychologists, special services helps in the Most cases. Also it is important to monitor developmental progression, child's scholastic performance and school behavior. Prognosis of the it will depend on the severity of the learning disabilities and subsequent intervention and compliance to therapy.

CONCLUSION

The clinical exome for PCD group helped in confirming the clinical diagnosis and in providing better understanding of relationship between gene mutations and diagnostic phenotype. Irrespective of underlying genetic issue developmental delay, behavioral issues and learning disability has to be addressed. The child with PCD with proper management and life style modifications can have better life style with less complications.

The PCD can affect the central nervous system by various ways and affect the proper development leading to learning disability in the affected individual. Early intervention and compliance to therapy leads to better outcomes in cases of learning disabilities.

REFERENCES

1. Ellerman A, Bisgaard H. Longitudinal study of lung function in a cohort of primary ciliary dyskinesia. Eur Respir J 1997; 10:2376–2379 [PubMed] [Google Scholar].
2. Valente EM, Rosti RO, Gibbs E, Gleeson JG. Primary cilia in neurodevelopmental disorders. Nat Rev Neurol. 2014 Jan;10(1):27-36. doi: 10.1038/nrneurol.2013.247. Epub 2013 Dec 3. PMID: 24296655; PMCID: PMC3989897.
3. Higginbotham H, et al. ARL13B in primary cilia regulates the migration and placement of interneurons in the developing cerebral cortex. Dev. Cell. 2012;23:925–938. [PMC free article] [PubMed] [Google Scholar].
4. Cantagrel V, et al. Mutations in the cilia gene ARL13B lead to the classical form of Joubert syndrome. Am. J. Hum. Genet. 2008;83:170–179. [PMCfreearticle] [PubMed] [Google Scholar].
5. Basu B, Brueckner M. Cilia multifunctional organelles at the center of vertebrate left-right asymmetry. Curr Top Dev Biol. 2008;85:151–174. [PubMed] [Google Scholar] [Ref list].
6. Al-Mahrezi A,Al-Futaisi A,Al-Mamari W, Learning Disabilities: Opportunities and challenges in Oman. Sultan Qaboos University medical journal. 2016 May; [PubMed PMID: 27226902].
7. Ministry of Social Justice and Empowerment. Notification, 2018, Gazette of India (Extra-Ordinary) Department of Department of Empowerment of Persons with Disabilities (Divyangjan) 2018. Jan 4, [Last accessed on 2018 Mar 26]. Available from: https://groups.google.com/d/topic/wethepwd/XuRiT0VdWsg .
8. Zariwala MA, Knowles MR, Leigh MW. Primary Ciliary Dyskinesia. In: Pagon RA, Adam MP, Ardinger HH, Wallace SE, Amemiya A, Bean LJH, Bird TD, Fong CT, Mefford HC, Smith RJH, Stephens K, editors. Gene Reviews(R); Seattle (WA): 1993. (updated Sept 2015) [Google Scholar] [Ref list].
9. Meeks M, Bush A. Primary ciliary dyskinesia (PCD). Pediatr Pulmonol 2000; 29:307–316 [PubMed] [Google Scholar].
10. Bush A, Chodhari R, Collins N, Copeland

F, Hall P, Harcourt J, et al. Primary ciliary dyskinesia: current state of the art. Arch Dis Child 2007; 92:1136–1140 [PMC free article] [PubMed] [Google Scholar] [Ref list].

11. King PT, Holmes PW. Use of antibiotics in bronchiectasis. Rev Recent Clin Trials. 2012;7:24–30. [PubMed] [Google Scholar].
12. Bradley J, Moran F, Greenstone M. Physical training for bronchiectasis. Cochrane Database Syst Rev (2002) 3: CD002166. doi:10.1002/14651858.CD002166PubMed Abstract | CrossRef Full Text | Google Scholar.
13. Gokdemir Y, Karadag-Saygi E, Erdem E, Bayindir O, Ersu R, Karadag B, et al. Comparison of conventional pulmonary rehabilitation and high-frequency chest wall oscillation in primary ciliary dyskinesia. Pediatr Pulmonol (2014) 49:611–6. doi:10.1002/ppul.22861 PubMed Abstract| CrossRef Full Text |.
14. Deuse T, Reitz BA. Heart-lung transplantation in situs inversus totalis. Ann Thorac Surg (2009) 88:1002–3. doi:10.1016/j.athoracsur.2009.01.060 PubMed Abstract | CrossRef Full Text | Google Scholar.
15. Shapiro AJ, Zariwala MA, Ferkol T, Davis SD, Sagel SD, Dell SD, et al. Diagnosis, monitoring, and treatment of primary ciliary dyskinesia: PCD foundation consensus recommendations based on state of the art review. Pediatr Pulmonol (2016) 51:115–32. doi: 10.1002/ppul.23304.
16. Snowling MJ,Gallagher A,Frith U, Family risk of dyslexia is continuous: individual differences in the precursors of reading skill. Child development. 2003 Mar-Apr [PubMed PMID: 12705560].
17. Kohli A, Sharma S, Padhy SK, Specific Learning Disabilities: Issues that Remain Unanswered. Indian journal of psychological medicine. 2018 Sep-Oct; [PubMed PMID: 30275613].