using a combination of the NGS and MLPA techniques, revealed two pathogenic mutations in the *DNAI1* gene: c.1612G>A (p.Ala538Thr) and c.180G>A (silent) variants. Both the patient's parents and brother are heterozygous carriers of a pathogenic variant. The patient's family history shows that the mother suffers from chronic sinusitis. The maternal grandfather is known to have dextrocardia, but he did not undergo genetic testing. As potential complications can occur due to recurrent clinical manifestation, a multidisciplinary approach is required to ensure our patient's a better quality of life. Rare genetic disorders and their implications have a significant emotional impact on pediatric patients and their families, so psychological counseling must be included in case management.

Declarations

Ethics approval and Consent to Participate

Not applicable.

Patient Consent for Publication

The patient gave consent to publish the case details and associated images.

Acknowledgments

Not applicable

Competing Interests

The authors declare that they have no competing interests.

Availability of Data and Materials

The datasets used during the present study are available from the corresponding author upon reasonable request.

Authors' Contributions

AC performed genetic consults and counseling; EC has seen and confirmed the raw data's authenticity; MSM interpreted the genetic test results. AFC and CP had clinical and imaging evaluations. All the authors have read and approved the final manuscript.

Funding

No funding was received.

References

1. Kant S, Mishra M, Kumar N, et al. Kartagener′s syndrome: A case series. Lung India 29 (2012): 366.
2. Barbato A, Frischer T, Kuehni C, et al. Primary ciliary dyskinesia: a consensus statement on diagnostic and treatment approaches in children. European Respiratory Journal 34 (2009): 1264-1276.
3. Ciancio N, de Santi MM, Campisi R, et al. Kartagener's syndrome: review of a case series. Multidiscip Respir Med 10 (2015): 18.
4. Özkara B, GOkur G, Kartagener syndrome: A quick glance through a case series misdiagnosed as asthma, European Respiratory Journal 48 (2016): PA3826.
5. Kumar A, Verma AK, Kumar A, et al. Kartagener syndrome: A review of three cases. Sahel Med J 18 (2015): 203-206.
6. Raidt J, Wallmeier J, Hjeij R, et al. Ciliary beat pattern and frequency in genetic variants of primary ciliary dyskinesia. Eur Respir J 44 (2014): 1579-1588.
7. Eliasson R, Mossberg B, Camner P, et al. The Immotile-Cilia Syndrome. New England Journal of Medicine 297 (1977): 1-6.
8. Davis EE, Katsanis N. The ciliopathies: A transitional model into systems biology of human genetic disease. Curr Opin Genet Dev 22 (2012): 290-303.
9. Focșa IO, Budișteanu M, Bălgrădean M. Clinical and genetic heterogeneity of primary ciliopathies (Review). Int J Mol Med 48 (2021): 176.
10. Shoemark A, Rubbo B, Legendre M, et al. Topological data analysis reveals genotype-phenotype relationships in primary ciliary dyskinesia. Eur Respir J 58 (2021): 2002359.
11. Nykamp K, Anderson M, Powers M, et al. Invitae Clinical Genomics Group, Topper S. Sherloc: a comprehensive refinement of the ACMG-AMP variant classification criteria. Genet Med 19 (2017): 1105-1117.
12. Vanaken GJ, Bassinet L, Boon M, et al. MaitreInfertility in an adult cohort with primary ciliary dyskinesia: phenotype–gene association European Respiratory Journal 50 (2017): 1700314.
13. Tanaka A, Nagayoshi M, Tanaka I, et al. Successful treatments for Kartagener syndrome with completely immotile sperm or azoospermia. Fertility and Sterility 106 (2016): e228.
14. Ozkavukcu S, Celik-Ozenci C, Konuk E, et al. Live birth after Laser Assisted Viability Assessment (LAVA) to detect pentoxifylline resistant ejaculated immotile spermatozoa during ICSI in a couple with male Kartagener's syndrome. Reproductive Biology and Endocrinology 16 (2018).
15. Bogart KR, Irvin VL. Health-related Quality of Life among Adults with Diverse Rare Disorders. Orphanet J. Rare Dis 12 (2017): 177-179.
16. Delaye J, Cacciatore P, Kole A. Valuing the "Burden" and Impact of Rare Diseases: A Scoping Review. Front Pharmacol 13 (2022): 914338.
17. Botkin J. Ethical issues in pediatric genetic testing and screening. Current Opinion in Pediatrics 28 (2016): 700-704.