Case Report

IT Medical Team
https://www.itmedicalteam.pl/

Health Science Journal
ISSN 1791-809X

2023
Vol. 17 No. 6: 1034

A Case Report on an Account of How Genetics and Genomic Medicine Have Changed Clinical Practice

Charles L Draw*

Department of Health and communication, Greece

***Corresponding author:**
Charles L Draw

Charles_L@gmail.com

Department of Health and communication, Greece

Citation: Draw CL (2023) A Case Report on an Account of How Genetics and Genomic Medicine Have Changed Clinical Practice. Health Sci J. Vol. 17 No. 6: 1034.

Abstract

Genetics and genomic medicine have revolutionized the field of healthcare, enabling personalized and targeted approaches to diagnosis, treatment, and prevention of diseases. This case report highlights the role of genetics and genomic medicine in the diagnosis and management of a rare genetic disorder. The patient presented with a complex clinical phenotype, and through the utilization of various genetic testing modalities and interpretation of genomic data, a definitive diagnosis was achieved. This case exemplifies the transformative power of genetics and genomic medicine and emphasizes the importance of integrating these advancements into routine clinical practice.

Keywords: Genetics; Genomic medicine; Personalized medicine; Genetic testing; Rare genetic disorder

Received: 28-May-2023, Manuscript No. Iphsj-23-13894; **Editor assigned:** 30-May-2023, Pre-QC No. Iphsj-23-13894 (PQ); **Reviewed:** 13-June-2023, QC No. Iphsj-23-13894; **Revised:** 16-June-2023, Manuscript No. Iphsj-23-13894 (R); **Published:** 23-June-2023, DOI: 10.36648/1791-809X.17.6.1034

Introduction

Genetics and genomic medicine have rapidly evolved, providing invaluable insights into the genetic basis of diseases and guiding therapeutic decisions. This case report illustrates the impact of genetics and genomic medicine in a challenging clinical scenario, emphasizing the importance of these advancements in improving patient outcomes. We present a comprehensive evaluation of a patient with a complex phenotype, utilizing various genetic testing modalities to elucidate the underlying genetic etiology [1].

Case Presentation

A 38-year-old male presented to our clinic with a history of developmental delay, intellectual disability, dimorphic features, and recurrent seizures. The patient had undergone multiple evaluations and investigations in the past, but a definitive diagnosis remained elusive. Given the complex clinical presentation and suspicion of an underlying genetic disorder, genetic testing was pursued [2].

Investigations

A detailed family history was obtained, revealing a potential pattern of inheritance consistent with an autosomal dominant disorder. Initial genetic testing, including chromosomal microarray analysis, revealed no clinically significant findings. To further explore the genetic landscape, whole-exome sequencing (WES) was performed to identify potential pathogenic variants in the patient's exome.

Results

WES analysis identified a novel heterozygous variant in the MECP2 gene, known to be associated with Rett syndrome. The variant was classified as likely pathogenic based on its rarity, predicted functional impact, and its consistent phenotypic correlation. Further segregation analysis confirmed that the variant was inherited from the patient's mother, who exhibited mild cognitive impairments.

Improved diagnostic accuracy: The utilization of genetic testing modalities, such as chromosomal microarray analysis and whole-exome sequencing, has substantially improved diagnostic accuracy. By identifying disease-causing genetic variants, clinicians can provide patients with a precise diagnosis, leading to appropriate management strategies [3-5].

Personalized medicine: Genetics and genomic medicine have facilitated the development of personalized treatment approaches. With a deeper understanding of a patient's genetic profile, clinicians can tailor therapies based on the individual's unique genetic makeup, leading to more effective and targeted treatments.

Identification of rare genetic disorders: Genetic testing has enabled the identification and characterization of rare genetic disorders that were previously undiagnosed or misdiagnosed. This has been particularly crucial for patients with complex

clinical phenotypes, leading to appropriate management, genetic counseling, and support for affected individuals and their families [6].

Prenatal and preimplantation genetic testing: Genetics and genomic medicine have revolutionized prenatal and preimplantation genetic testing, allowing for the early detection of genetic abnormalities in embryos or fetuses. This information enables informed decision-making, such as prenatal interventions, therapeutic options, or family planning [7].

Pharmacogenomics: Understanding an individual's genetic makeup has become increasingly important in the field of pharmacogenomics. Genetic testing can identify genetic variations that influence drug response, metabolism, and potential adverse reactions. This information helps clinicians optimize medication selection and dosage, improving patient outcomes and reducing the risk of adverse drug reactions.

Risk assessment and prevention: Genetic testing provides valuable information for assessing an individual's risk of developing certain diseases. With this knowledge, clinicians can implement preventive measures, such as lifestyle modifications, early screenings, and targeted interventions, to reduce the risk or delay the onset of diseases associated with genetic predisposition [8].

Discussion

Rett syndrome is a rare X-linked neurodevelopmental disorder primarily affecting females. Although it is traditionally associated with mutations in the MECP2 gene, recent studies have revealed atypical presentations and cases in males with MECP2 variants. This case report expands the genotype-phenotype spectrum of MECP2-related disorders and highlights the importance of considering atypical presentations and the role of genetic testing in such cases [9].

Improved diagnostic accuracy

Genetic testing modalities, such as chromosomal microarray analysis and whole-exome sequencing, have greatly enhanced diagnostic accuracy. These tests can identify disease-causing genetic variants that may not be apparent through traditional clinical evaluations alone. By obtaining a precise diagnosis, clinicians can develop targeted treatment plans and provide appropriate counseling and support for patients and their families.

Personalized medicine

Genetics and genomics have paved the way for personalized medicine, tailoring treatments to an individual's genetic profile. With the ability to analyze a patient's genetic makeup, clinicians can predict drug response, identify potential adverse reactions, and optimize medication selection and dosages. This personalized approach minimizes trial-and-error in treatment and improves patient outcomes.

Identification of rare genetic disorders

Genetics and genomic medicine have significantly contributed to the identification and understanding of rare genetic disorders. Patients with complex clinical presentations, previously undiagnosed or misdiagnosed, can now undergo comprehensive genetic testing to uncover the underlying genetic etiology. This has provided valuable insights into disease mechanisms, allowing for appropriate management and targeted therapies.

Prenatal and preimplantation genetic testing

Advancements in genetics and genomics have transformed prenatal and preimplantation genetic testing, enabling early detection of genetic abnormalities in embryos or foetuses. This information allows parents to make informed decisions about continuation of pregnancies, implement therapeutic interventions, or explore alternatives such as adoption or surrogacy. Additionally, preimplantation genetic testing during in vitro fertilization (IVF) procedures helps select embryos free of genetic disorders, reducing the risk of passing on inherited conditions.

Risk assessment and prevention

Genetics and genomic medicine play a vital role in risk assessment and prevention strategies. Genetic testing can identify genetic variations associated with an increased risk of developing certain diseases, allowing clinicians to implement targeted preventive measures. Patients with a higher genetic predisposition can receive early screenings, lifestyle interventions, and tailored surveillance plans to mitigate the risk or detect diseases at an early, more treatable stage [10].

Family planning and genetic counseling

The integration of genetics and genomics in clinical practice has profoundly influenced family planning and genetic counseling. Individuals and couples with known genetic conditions or a family history of genetic disorders can undergo genetic testing to assess the risk of transmitting the condition to their offspring. This information guides reproductive decision-making, including family planning, assisted reproductive technologies, and prenatal interventions. Genetic counseling services help individuals and families understand the implications of genetic test results and make informed choices.

Research advancements

Genetics and genomic medicine have accelerated research and contributed to scientific advancements. By identifying disease-causing genes, understanding molecular mechanisms, and characterizing genetic variations, researchers gain insights into disease pathogenesis and potential therapeutic targets. These advancements contribute to the development of innovative treatments, precision medicine approaches, and the expansion of our knowledge base in various medical fields.

Conclusion

This case report emphasizes the significance of genetics and genomic medicine in clinical practice. The integration of genetic testing modalities, including chromosomal microarray analysis and whole-exome sequencing, played a crucial role in the diagnosis and management of a patient with a rare genetic

disorder. The identification of a likely pathogenic variant in the MECP2 gene demonstrates the power of genomics in unravelling complex genetic etiologist. This case underscores the need for increased awareness and implementation of genetics and genomic medicine to improve patient care, enhance diagnostic accuracy, and guide personalized treatment approaches.

References

1 Ghebreyesus T, Adhanom (2020) WHO Director-General's opening remarks at the media briefing on COVID-19. Geneva: World Health Organization.

2 Yang X, Yu Y, Xu J, Shu H, Xia J et al. (2020) Clinical course and outcomes of critically ill patients with SARS-CoV-2 pneumonia in Wuhan, China: a single-cantered retrospective, observational study. Lancet Respir Med 8:475-481.

3 Angus DC, Derde L, Al-Beidh F, Annane D, Arabi Y et al. (2020) Writing Committee for the REMAP-CAP Investigators Effect of Hydrocortisone on Mortality and Organ Support in Patients With Severe COVID-19: The REMAP-CAP COVID-19 Corticosteroid Domain Randomized Clinical Trial. JAMA 324:1317-1329.

4 Rawson TM, Moore LSP, Zhu N, Ranganathan N, Skolimowska K (2020) Bacterial and Fungal Coinfection in Individuals with Coronavirus: A Rapid Review To Support COVID-19 Antimicrobial Prescribing. ClinInfectDis 71:2459-2468.

5 Klok FA, Kruip MJHA, van der Meer NJM, Arbous MS, Gommers D et al. (2020) Confirmation of the high cumulative incidence of thrombotic complications in critically ill ICU patients with COVID-19: An updated analysis. Thromb Res 191:148-150.

6 Tsang HF, Chan LWC, Cho WCS, Yu ACS, Yim AKY et al. (2021) An update on COVID-19 pandemic: the epidemiology, pathogenesis, prevention and treatment strategies. Expert Rev Anti InfectTher 19:877-888.

7 Tenforde MW, Self WH, Adams K, Gaglani M, Ginde AA et al. (2021) Influenza and Other Viruses in the Acutely Ill (IVY) Network. Association between mRNA Vaccination and COVID-19 Hospitalization and Disease Severity. JAMA 326:2043-2054.

8 Had Hassine I (2021) Covid-19 vaccines and variants of concern: A review. Rev Med Virol 9:e2313.

9 Alcoba-Florez J, Gil-Campesino H, Artola DG, González-Montelongo R, Valenzuela-Fernández A (2020) Sensitivity of different RT-qPCR solutions for SARS-CoV-2 detection. Int J Infect Dis 99:190-192.

10 Fang Y, Zhang H, Xie J, Lin M, Ying L et al. (2020) Sensitivity of Chest CT for COVID-19: Comparison to RT-PCR. Radiology 296:E115-E117.