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| Book Name: | [Medical Science: Trends and Innovations](https://www.bookpi.org/bookstore/product/medical-science-trends-and-innovations-vol-1/) |
| Manuscript Number: | **Ms\_BPR\_4473** |
| Title of the Manuscript: | **A DiGeorge Syndrome Case Report—Challenges of Diagnosis and Management** |
| Type of the Article | **BOOK CHAPTER** |

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| PART 1: Comments | | |
|  | Reviewer’s comment **Artificial Intelligence (AI) generated or assisted review comments are strictly prohibited during peer review.** | Author’s Feedback*(Please correct the manuscript and highlight that part in the manuscript. It is mandatory that authors should write his/her feedback here)* |
| **Please write a few sentences regarding the importance of this manuscript for the scientific community. A minimumof 3-4 sentences may be required for this part.** | DiGeorge syndrome (also known as velo-cardio-facial syndrome) is a rare genetic disorder caused by a microdeletion on chromosome 22q11.2, affecting multiple organ systems, including the heart, immune system, and calcium regulation. Diagnosing DiGeorge syndrome is challenging, especially in regions with limited access to advanced genetic testing, like the Republic of Moldova. In this case, a 6-month-old girl, who had a normal pregnancy and prenatal ultrasounds, presented with cardiac issues and recurrent infections. Despite multiple treatments, her diagnosis was delayed until 9 months when genetic testing revealed a complete 22q11.2 deletion, confirming DiGeorge syndrome. Her condition included cardiac malformations, immune deficiencies, and hypoparathyroidism, requiring extensive treatment and care. The case highlights the importance of a comprehensive diagnostic approach, especially in settings with fewer resources. Early recognition of the syndrome could prevent delays in treatment and improve patient outcomes. |  |
| **Is the title of the article suitable?**  **(If not please suggest an alternative title)** | To make the title more approachable and humanized, here’s a suggestion:  **"A Patient's Journey with DiGeorge Syndrome: Navigating Diagnostic and Treatment Challenges"** |  |
| Is the abstract of the article comprehensive? Do you suggest the addition (or deletion) of some points in this section? Please write your suggestions here. | A few suggestions I’d like to offer to enhance its clarity and impact:  **Abstract**  **Background**: DiGeorge syndrome (also known as velo-cardio-facial syndrome) is a rare genetic condition affecting about 1 in 4,000 to 6,000 live births. While advancements in genetic testing have improved early diagnosis in developed countries, it remains underrecognized in places like Moldova, where access to these tests is limited. Prenatal screening usually includes ultrasounds and routine checks, but the syndrome is hard to detect through ultrasound alone, especially since it often doesn’t show up in clear ways during pregnancy. Genetic testing is usually done when doctors suspect a genetic issue, but this can be missed or delayed, especially when the signs are subtle.  **Purpose**: This study aims to shed light on the diagnostic challenges of DiGeorge syndrome, urging doctors to think about it as a multi-faceted condition, rather than focusing on just one system or symptom at a time.  **Method**: We present the case of a 6-month-old girl, born after an uneventful pregnancy and with a normal prenatal ultrasound, who started showing signs of heart failure. After a series of tests and surgeries, she was eventually diagnosed with DiGeorge syndrome at 9 months.  **Results**: The diagnosis was delayed because there were no clear prenatal markers, and doctors focused on investigating her heart and other individual issues rather than looking at the bigger picture. Despite several treatments to manage her symptoms, the diagnosis was only made after noticing the pattern of various symptoms and conducting genetic tests.  **Conclusions**: This case highlights the need for a more holistic approach to diagnosis, one that looks at the full spectrum of symptoms and integrates multiple diagnostic tests. It also shows how important it is to raise awareness in regions where advanced genetic testing is less accessible, so that we can catch DiGeorge syndrome earlier and ensure timely treatment. |  |
| **Is the manuscript scientifically, correct? Please write here.** | The manuscript provides an accurate and well-rounded look at the challenges of diagnosing DiGeorge syndrome, especially in regions with limited access to genetic testing. It highlights how the condition often goes undetected during pregnancy and the importance of taking a more holistic approach to diagnosis. The case study effectively emphasizes the need for early recognition and better awareness to ensure timely treatment. |  |
| **Are the references sufficient and recent? If you have suggestions of additional references, please mention them in the review form.**  **-** | The references listed are relevant and cover key aspects of DiGeorge syndrome, including its genetic causes, immunological aspects, and clinical management. They span a range of research, from genetic and immunologic studies to population-based studies and clinical protocols. The references are comprehensive but mostly fall within the 2000s and early 2010s, with the most recent being from 2021. However, adding more recent studies on diagnostic advancements and treatment options could make the article even more up-to-date and informative. |  |
| Is the language/English quality of the article suitable for scholarly communications? | The language and English quality of the article seem appropriate for scholarly communication. However, there are several areas where the writing could be more polished for clarity and coherence. |  |
| Optional/Generalcomments | The article provides valuable insights into the clinical presentation, diagnostic challenges, and management of DiGeorge syndrome, highlighting the importance of early recognition and a comprehensive, multidisciplinary approach, especially in resource-limited settings. |  |

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| **PART 2:** | | |
|  | **Reviewer’s comment** | **Author’s comment** *(if agreed with reviewer, correct the manuscript and highlight that part in the manuscript. It is mandatory that authors should write his/her feedback here)* |
| **Are there ethical issues in this manuscript?** | *(If yes, Kindly please write down the ethical issues here in details)* |  |

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| **Reviewer Details:** | |
| Name: | **Ankita R Bhatiya** |
| Department, University & Country | **Smt.L.P.Patel Institute of MLT, India** |