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A Complex Case of Partial Digeorge Syndrome with Multiple System Involvement: Seizure Disorder, Hypothyroidism and Recurrent Infection

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ABSTRACT

DiGeorge Syndrome (DGS), or 22q11.2 deletion syndrome (22q11DS), is a genetic disorder characterized by a microdeletion on chromosome 22 at band q11.2, presenting with a range of clinical features including immunodeficiency, hypoparathyroidism, and congenital heart disease. Partial DiGeorge Syndrome involves a subset of these features, complicating diagnosis and management.

This case study describes a 45-year-old female with partial DiGeorge Syndrome who presented with a seizure disorder, hypothyroidism, and recurrent infections. Following a fall, she sustained multiple facial bone fractures and developed respiratory symptoms. Her management included intensive care for seizure activity, correction of electrolyte imbalances, and treatment for respiratory complications and infections. The patient required multidisciplinary care involving neurologists, endocrinologists, and pulmonologists.

The patient's complex presentation highlighted the need for comprehensive, coordinated medical care to address the multifaceted impacts of partial DiGeorge Syndrome. Key management strategies included continuous monitoring, endocrine support, and targeted antibiotic therapy.

This case underscores the importance of a multidisciplinary approach in managing partial DiGeorge Syndrome, emphasizing the need for integrated care to improve patient outcomes and quality of life.

KEYWORDS: DiGeorge Syndrome, Seizure Disorder, Hypothyroidism, Recurrent Infection

INTRODUCTION

DiGeorge Syndrome (DGS), or 22q11.2 deletion syndrome (22q11DS), is a genetic disorder caused by a microdeletion on chromosome 22 at band q11.2. Initially recognized in 1828 and later detailed by Dr. Angelo DiGeorge in 1965, this syndrome includes a range of clinical features such as immunodeficiency, hypoparathyroidism, and congenital heart disease. Partial DiGeorge Syndrome refers to cases where only some of the typical features are present, adding complexity to diagnosis and management.[1,2,3]

This case study focuses on a patient with partial DiGeorge Syndrome who presents with a combination of seizure disorder, hypothyroidism, and recurrent infections. These symptoms illustrate the multi-system involvement characteristic of 22q11DS and underscore the importance of comprehensive, multidisciplinary medical care.

DiGeorge Syndrome is highly variable in its presentation, with over 180 documented clinical features affecting multiple organ systems. Key characteristics include physical anomalies such as cardiac defects and thymic hypoplasia, cognitive and behavioral issues, and varying degrees of immunodeficiency. The extent of thymic hypoplasia can influence the severity of immunodeficiency, leading to an increased susceptibility to infections.[1,4]

Our patient's presentation with seizure disorder, hypothyroidism, and recurrent infections highlights the diverse and complex nature of partial DiGeorge Syndrome. The seizure disorder reflects the neurological involvement often seen in 22q11DS, necessitating ongoing neurological assessment and management. Hypothyroidism, due to parathyroid gland hypoplasia, requires regular endocrine evaluations and treatment to manage thyroid hormone deficiency. The recurrent infections point to the underlying immunodeficiency, requiring vigilant monitoring and prompt medical intervention.[1,5]

Effective management of partial DiGeorge Syndrome demands a multidisciplinary approach, with coordinated care from neurologists, endocrinologists, immunologists, and other specialists. This ensures comprehensive monitoring and tailored treatments to address the multifaceted needs of the patient, ultimately aiming to improve their quality of life and clinical outcomes.[1]

CASE REPORT

A 45-year-old female was admitted to the hospital on 16/01/2024 given a history of fall on 15/01/2024, around midnight, sustaining multiple facial bone fractures. The patient also had complaints of cough for the past 1 week, along with fever and breathlessness on 16/01/2024. CT Brain and facial bone revealed B/L nasal bone fracture with minimal displacement, doubtful fracture of right inferior orbital wall. The patient had involuntary movements of both upper and lower limbs on 19/01/2024 (seizure activity) and was shifted to CCU. She is a known case of seizure disorder, DLP, bronchial asthma, and hypothyroidism. History of surgery for VSD (more than 20 years back). On arrival at CCU, the patient was found to have a GCS of 9 9patient given Inj. Lorazepam at ward). The patient was managed in the CCU with IV antiepileptics, IV antibiotics, and other supportive care. S.calcium levels were found to be low, along with elevated phosphorous levels, low magnesium, and hypokalemia. MRI Brain 20/1/2024 showed dense blooming in bilateral basal ganglia SWI-likely calcifications, no evidence of intracranial hemorrhage, infract, or space-occupying lesion. The patient also has a history of symptoms of carpopedal spasm and hypocalcemia. Parathyroid hormone levels were found to be low. Endocrine consultation sought for the same, advised to continue IV calcium. Symptoms of cough aggravated since 22/1/2024, ENT consultation sought for the same, advised to continue medical management serial EEG showed no spikes (only slowing) till 21/1/2024; seizure activity noted in EEG since then and serial EEG taken afterward showed occasional spikes, but of decreasing frequency. Patient GCS showed a fluctuating pattern daily; CT neck and USG abdomen were done to rule out other causes of

hypoparathyroidism, but the report showed no significant abnormality. MRI Brain on 23/01/2024 showed no significant abnormality. CT chest repeated on 23/01/2024 revealed (L) upper lobe collapse and no findings to explain chronic cough. LP study was done on 23/01/2024 to rule out infection causes, (TC showed 1 cell) and CSF was sent for an autoimmune encephalitis panel. On 25/01/2024 given low GCS patient got intubated and mechanically ventilated (difficult airway) following blood C&S sent. Pulmonology consultation sought advised bronchoscopy, BAL from (L) upper lobe taken and sent. 25/01/2024 The patient developed hypotension and started on a single IV ionotropic. BAL revealed growth of pseudomonas aeruginosa and blood culture revealed growth of Enterobacter cloacae and antibiotics changed accordingly. Given prolonged ventilation tracheostomy was done on 27/01/2024.The autoimmune encephalitis panel came negative on 29/01/2024. Repeat EEG, on 31/01/2024 showed no epileptic abnormalities. Repeat blood culture came sterile on 31/01/2024. Persistingly patient serum calcium, magnesium, and potassium were found to be low.

DISCUSSION

DiGeorge Syndrome (DGS), or 22q11.2 deletion syndrome (22q11DS), is characterized by a wide range of clinical manifestations due to a microdeletion on chromosome 22 at band q11.2. This case study of a 45-year-old female patient with partial DiGeorge Syndrome exemplifies the complexity and variability of this genetic disorder. The patient presented with a multifaceted clinical picture, including seizure disorder, hypothyroidism, and recurrent infections, each adding layers of complexity to her medical management.

The patient's history of facial bone fractures following a fall, coupled with respiratory symptoms, underscores the need for comprehensive care in trauma cases involving patients with underlying complex syndromes. The identification of bilateral nasal bone fractures and a doubtful fracture of the right inferior orbital wall via CT imaging required careful monitoring to prevent complications.

Seizure activity, a significant concern in this case, was managed with IV antiepileptics and supportive care in the CCU. The presence of dense blooming in bilateral basal ganglia on MRI, likely due to calcifications, alongside low parathyroid hormone levels, highlights the interconnected nature of neurological and endocrine abnormalities in DGS. The patient's history of carpopedal spasms and hypocalcemia further indicates underlying hypoparathyroidism, necessitating ongoing endocrine consultation and IV calcium administration.

The persistent respiratory symptoms, coupled with a left upper lobe collapse noted on the CT chest, required ENT and pulmonology consultations. The growth of Pseudomonas aeruginosa from BAL and Enterobacter cloacae from blood cultures pointed to significant infections requiring targeted antibiotic therapy. The patient’s need for intubation and subsequent tracheostomy due to prolonged ventilation highlights the critical nature of her respiratory complications.

The fluctuating GCS scores and the presence of seizure activity on EEG underscore the need for continuous neurological monitoring. The negative autoimmune encephalitis panel and sterile repeat blood cultures are reassuring but highlight the need for vigilance in monitoring for new or recurrent infections.

CONCLUSION

This case illustrates the multifaceted challenges in managing partial DiGeorge Syndrome with multi-system involvement. The complexity of this case, involving seizure disorder, hypothyroidism, and recurrent infections, underscores the importance of a multidisciplinary approach. Effective management requires coordinated efforts from neurologists, endocrinologists, pulmonologists, ENT specialists, and critical care teams.

This case emphasizes the need for ongoing, comprehensive care in patients with partial DiGeorge Syndrome. The patient's course highlights the potential for significant complications and the necessity for a proactive, collaborative approach to improve clinical outcomes and quality of life. The successful management of such a complex case hinges on the integration of expertise across multiple medical disciplines, tailored to the individual patient's evolving needs.

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