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Title	Delayed Diagnosis of 22q1 1.2 Deletion Syndrome in a Teenager with Refractory Epilepsy and Hypocalcemia
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INTRODUCTION

DiGeorge syndrome (22q11.2 deletion syndrome) is a congenital disorder resulting from a microdeletion on chromosome 22. It presents with highly variable features, including cardiac defects, hypocalcemia, immunodeficiency, and neuropsychiatric involvement. While often diagnosed in infancy, cases with atypical features may be identified later in life.

AIM

To highlight the diagnostic challenges and multisystem involvement of DiGeorge syndrome in a pediatric patient with delayed diagnosis, and to emphasize the importance of clinical suspicion beyond infancy.

METHODS

We report the case of a 14-year-old girl with:- History of ventricular septal defect, recurrent sinusitis, and anal atresia- Refractory epilepsy, intellectual disability, anxiety, and bilateral basal ganglia calcifications- Notable dysmorphic facial features She presented with symptoms of hypocalcemia. Laboratory results showed low calcium and parathyroid hormone levels. Diagnosis was confirmed by fluorescence in situ hybridization (FISH) demonstrating a 22q11.2 microdeletion.

RESULTS

- Initiation of calcium and vitamin D supplementation led to clinical improvement- Genetic counseling was provided to the family- The case revealed a syndromic picture involving cardiac, gastrointestinal, neurologic, and endocrine systems

DISCUSSION

This case underscores the broad spectrum of DiGeorge syndrome and the potential for delayed diagnosis in patients with subtle or atypical features. The co-occurrence of congenital anomalies, neuropsychiatric symptoms, and hypocalcemia should prompt evaluation for 22q11.2 deletion even in adolescence. Bilateral basal ganglia calcifications and epilepsy add to the expanding neurological phenotype of the syndrome.

CONCLUSION

DiGeorge syndrome should be considered in pediatric patients with hypocalcemia and multisystem manifestations, particularly in the presence of congenital anomalies and neurodevelopmental disorders. Early recognition and genetic testing are crucial for management and counseling, even beyond the typical diagnostic age.

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