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CLINICAL FEATURES OF 22Q11.2 DELETION SYNDROME: A LITERATURE REVIEW AND CASE SERIES REPORTS

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The 22q11.2 deletion syndrome is the most common microdeletion syndrome. The clinical features show a broad spectrum of multisystem manifestations and include congenital heart defects, hypoparathyroidism associated with hypocalcemia, hypoplasia of the thymus and subsequent immunodeficiency, distinctive facial features, velopharyngeal insufficiency, and developmental delay or learning disabilities. Retrospective analysis of medical data was conducted and the spectrum of clinical manifestations of case series of five patients with 22q11.2 deletion syndrome is presented. A small series of patients with a 22q11.2 deletion syndrome is described, but still a sufficient number that undisputably displays a recognizable spectrum of manifestations. All patients expressed elements of facial dysmorphism and signs of immune system dysfunction which ranged from lymphopenia and recurrent respiratory infections to congenital defect in T cell immunity. Almost all of the reported patients had associated conotruncal congenital heart defects, and the majority of cases presented with hypocalcemia and elements of motor and developmental delay. Increased awareness of multisystemic features of 22q11.2 deletion syndrome is pivotal for early recognition and early initiation of comprehensive care and treatment.

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