ORIGINAL RESEARCH PAPER

General Medicine

CASE REPORT : PARTIAL DIGEORGE SYNDROME PRESENTING IN ADULTHOOD

KEY WORDS: digeorge syndrome , hypocalcemia, intracerebral calcification

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ABSTRACT

Digeorge syndrome is the most common microdeletion syndrome, and probably underrecognized due to the varied manifestations and variable penetrance. Only a few cases of adult presentation of Digeorge Syndrome have been described in the literature. It is also known as velocardiofacial syndrome or CATCH 22 syndrome. Classically abnormal facies, congenital heart disease , thymus dysplasia, cleft palate , hypocalcemia due to hypoparathyroidism are seen. Hypocalcemia is a strong predictor of digeorge syndrome whenever associated with other clinical features. Patients with chromosome 22q11.2 deletion do not always show all components of DGS. Hypoparathyroidism can be the only abnormality and may exist with no accompanying cardiac or immunologic defects. Here we report a case of 28 year old man presenting in adulthood with hypocalcemia induced tetany and diagnosed as having partial Digeorge syndrome.

INTRODUCTION :

Digeorge syndrome is the most common microdeletion syndrome, and probably underrecognized due to the varied manifestations and variable penetrance. The phenotypic spectrum shows a wide variability. It has an estimated prevalence ranging from 1 in 4000 to 1 in 6395 [1,2]. Most cases (93%) have a de novo deletion, whereas the remaining 7% have inherited the deletion. . Only a few cases of adult presentation of Digeorge Syndrome have been described in the literature[3,4].

CASE :

A 28 year old man presented with a 4 year history of intermittent episodes of carpopedal spasm and tingling sensation over face and limbs. He had hearing loss ,learning and speech difficulty since childhood . On admission he was vitally stable with positive Trousseau's and chvostek's sign . On physical examination he had dysmorphic facial features[Figure 1] with long face , ocular hypertelorism, small mouth . Neurologic examination did not reveal any focal deficits. Other systems examination were unremarkable. An extensive medical and family history was taken. He had no history of seizures, radiation exposure, neck surgery, head trauma and no family history of similar complaints, any automimmune endocrinopathies, immunodeficiencies or congenital defects. Electrocardiogram showed sinus rhythm, t wave flattening and prolonged QTc(520 msec). Chest Xray, ultrasonography of abdomen and 2D echocardiogram were normal. Ophthalmic Evaluation and fundus examination were normal . Laboratory examination revealed low corrected serum calcium level 4.366 mg/dl, high serum phosphorus 5.4 mg/dl , normal Vitamin D3 level 29 ng/ml , low serum parathyroid hormone level 7.70 pg/ml . Complete blood count, liver function, renal function tests, serum magnesium, HIV, HBV, HCV serologies, Thyroid function test, serum magnesium level were normal. CT Brain plain[Figure 2] was suggestive of coarse calcification of bilateral basal ganglia, thalami, and calcific foci in frontal lobes. Pure Tone Audiometry revealed bilateral severe sensorineural hearing loss.

Based on the above he was diagnosed as having hypocalcemia secondary to hypoparathyroidism. Although the classical picture of digeorge syndrome was not entirely present but in the setting of dysmorphic features, hypoparathyroidism and other findings , a final diagnosis of partial digeorge syndrome was suspected and later confirmed on FISH study for 22q11.2 microdeletion using the tuple probe showing heterozygous deletion. He was treated with parenteral calcium correction and oral calcitriol. His serum calcium level normalized over 1 week and subsequently was discharged on oral calcium of 3 gm/day and remained asymptomatic at follow up .

DISCUSSION

Digeorge Syndrome is also known as velocardiofacial syndrome or CATCH 22 syndrome. Classically abnormal facies, congenital heart disease , thymus dysplasia, cleft palate , hypocalcemia due to hypoparathyroidism are seen. Renal abnormalities , skeletal defects, autoimmune disorders, neuropsychiatric disorders may also be seen. In adults , immunodeficiency is rare , but may be present in 70-80% of children with digeorge syndrome. Less than 1% patients have thymus aplasia and need immune reconstitution with bone marrow transplant .22.Hypocalcemia is present in 60 % of patients [5,6]. Hypocalcemia is a strong predictor of digeorge syndrome whenever associated with other clinical features. It commonly presents as tetany, focal or generalized seizures,muscle cramps, prolonged QT interval. Of the many causes of extensive intra cranial calcification as detected in unenhanced CT, hypoparathyroidism is one of the common causes of pathological basal ganglia calcification. Here, the calcifications are coarse or nodular, nearly symmetrical and usually localized to basal ganglia and adjacent structures. This was first noted by Eaton et al. in 1939[7]. Patients with chromosome 22q11.2 deletion do not always show all components of DGS. Hypoparathyroidism can be the only abnormality and may exist with no accompanying cardiac or immunologic defects [8]. Digeorge syndrome should be considered in adults presenting with hypocalcemia due to hypoparathyroidism even in the absence of classical features.

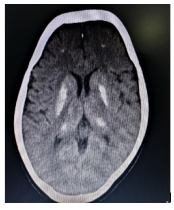

Figure 1 : Patient Photograph
Figure 2 : CT Brain Plain

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