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CASE REPORT
OLGU SUNUMU

Unilateral Internal Carotid Artery Hypoplasia Incidentally Found in Di George Syndrome diagnosed in Adolescence

Adölesan Dönemde Di George Sendromunda Rastlantısal Tespit Edilen Tek Taraflı İnternal Karatis Arter Hipoplazisi

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ABSTRACT

A 15-year-old child with mild dysmorphic features presented to the emergency department with a sudden onset loss of consciousness. Chovostek's and Trousseau's sign were positive. Laboratory investigations showed hypocalcemia (7,6 mg/dl normal range 8,4–10,2 mg/dl) and hyperphosphatemia (4,9 mg/dl normal range 2,3–4,7 mg/dl). He was confirmed to have primary hypoparathyroidism with a parathyroid hormone (PTH) level of 18,3 pg/ml (normal range 12–88 pg/ml). According to routine blood investigations he was confirmed to have primary hypoparathyroidism. In the view of his dysmorphic facial features, mental retardation and hypoparathyroidism,the 22q11.2 deletion syndrome was suspected. Genetic study showed heterozygous deletion of 22q11.2 region. Hypophysis and cranial magnetic resonance imaging (MRI) was performed because of growth retardation and seizure history. Unilateral Internal Carotid Artery Hypoplasia were seen incidentally.

Keywords: Di George Syndrome, internal carotid artery hypoplasia, adolescent

ÖZ

15 yaşında dismorfik yüz görünümü olan erkek çocuk acil servise ani bilinç kaybı nedeniyle başvurdu. Chovostek ve Trousseau bulguları pozitif idi. Labaratuar incelemede hipokalsemi ve hiperposfatemi vardı. Düşük parathormon düzeyi ile 18,3 pg/ml (normal aralık 12–88 pg/ml) hipoparatroidi tanısı aldı. tetkikleri prımer hipoparatroidi ile uyumlu idi.Dismorfik yüz görünümü,mental retardasyon, hipoparatroidi bulguları ile hastada 22q11.2 deletion syndromundan süphe edildi. Genetik analizinde 22q11.2 bölgesinde heterozigot delesyon mevcut idi.gelişme geriliği ve nöbet öyüsü nedeniyle hipofiz ve beyin magnetik resonans görüntülemesi yapıldı.Unilateral internal karotis arter hipoplazisi rastlantısal olarak görüldü.

Anahtar kelimeler: Di George sendromu, internal karatis arter hipoplazisi, adölesan

INTRODUCTION

Di George syndrome (DGS) is a constellation of signs and symptoms associated with defective development of the pharyngeal pouch system. Most cases are caused by a heterozygous chromosomal deletion at 22q11.2. The classic triad of features of DGS on presentation is conotruncal cardiac anomalies, hypoplastic thymus, and hypocalcemia (1).

CASE REPORT

A 15-year-old child with mild dysmorphic features presented to the emergency department with a sudden onset loss of consciousness which lasted approximately 1 minute, during which he sustained tonic-clonic movements which self-terminated. The patient reported that he had history of episodic carpopedal spasms and muscle cramps for 6 years. His past medical history revealed febrile seizure at 1 years and also learning disabilities since childhood.

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He was born at term by normal vaginal delivery (birth weight 3200 g) from a healthy mother as first children of the family. There was no consanguinity between the parents. There was no family history of hypocalcemia. On physical examination there was mild facial dysmorphism with a short philtrum. He spoke with a hypernasal speech. On admission he revealed a temperature of 36. 2 C, a pulse of 99 beats per minute, a respiratory rate of 22 per minute, and a blood pressure of 90/60 mm/Hg. The patient's height was 149cm (-2,7 SDS) and his weight was 42 kg (-2.04SDS).Calculated body mass index was 18,5 kg/m2 (-0,48SDS). He was pubertal at Tanner stage 5. Respiratory, cardiovascular and neurological systems' examination was normal. Chovostek's and Trousseau's sign were positive. Laboratory investigations showed hypocalcemia (7,6 mg/dl normal range 8,4–10,2 mg/dl) and hyperphosphatemia (4,9 mg/dl normal range 2,3–4,7 mg/dl). He was confirmed to have primary hypoparathyroidism with a parathyroid hormone (PTH) level of 18,3 pg/ml (normal range 12–88 pg/ml). Other routine blood investigations including magnesium levels were normal.

He was prescribed with intravenous calcium gluconate and then placed on oral calcium lactat (2000 mg two times per day) and calcitriol (0.50 mg daily) In the view of his dysmorphic facial features, mental retardation and hypoparathyroidism, the 22q11.2 deletion syndrome was suspected. Genetic study showed heterozygous deletion of 22q11.2 region. Hypophysis and cranial magnetic resonance imaging (MRI) was performed because of growth retardation and seizure history.

Hypoplastic left internal carotid artery and also narrowing of the left carotid canal were seen on hypophysis and cranial MRI (**Figure 1A**). On additional, contrast enhanced-MR angiography demonstrated that origins of main arteries were normal but common carotid artery was hypoplastic on the left side and the hypoplastic left İCA was extending to become thinner distally (**Figure 1B**).

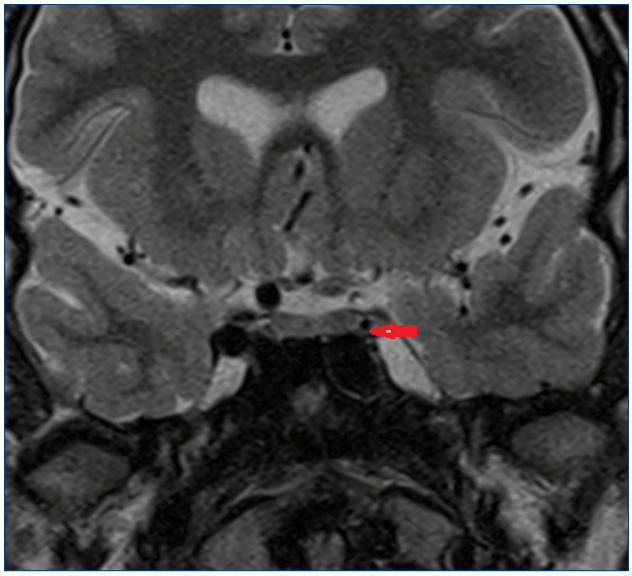

Figure 1A. Hypoplastic left internal carotid artery and narrow left carotid canal

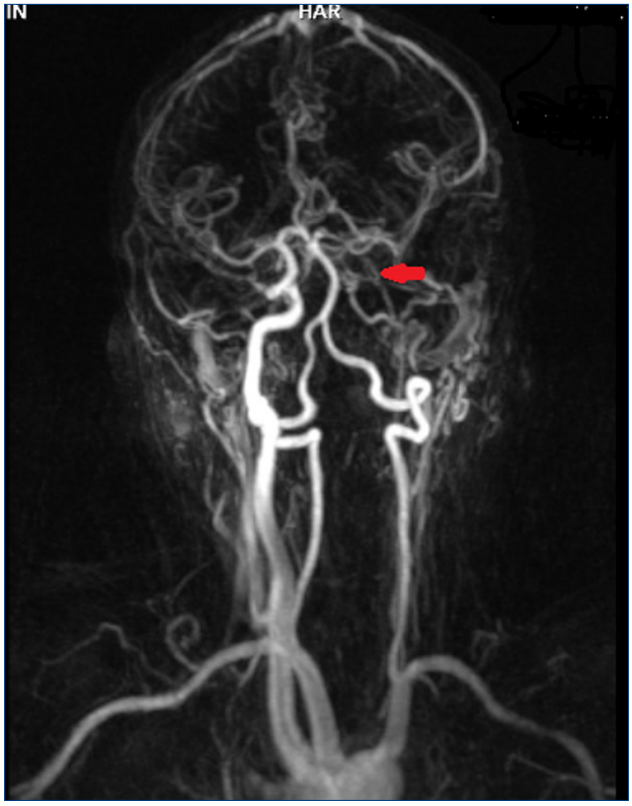

Figure 1B. Hypoplastic left İCA extending to become thinner distally

DISCUSSION

DiGeorge syndrome, which was originally described in 1967 by Di George et al. (1), is associated with microdeletions of chromosome 22q11.2 and less commonly chromosome 10p13. The syndrome is associated with failure of development of the third and fourth branchial pouches (2). There are variable findings include congenital heart diseases (74%), palatal abnormalities (69%), learning difficulties (80%) (3), hypoplasia or aplasia of the parathyroid glands and thymus glands, which cause hypocalcaemia (50%) and immune deficiency (70%) (4).

Multiple cardiac and vascular anomalies have been previously well described in 22q11 deletion syndrome, but but hypoplasia of the internal carotid is rare. In our review of the literature, we found only two case of ICA hypoplasia with DiGeorge syndrome (5,6)

We report a rare case of Di George syndrome with hypoplasia of internal caratid artery and presented as hypocalcemia-induced seizure.

This case was reported because association of DiGeorge syndrome and hypoplasia of internal caratid arter is rare. Also our patient was delayed diagnosis of DiGeorge Syndrome presented as hypocalcaemia-induced seizure.

CONCLUSION

DiGeorge syndrome should be considered in the differential diagnosis of hypocalcaemia presenting in adolescent. Although rare, DiGeorge syndrome may present with hypoplasia of the ICA in this age group.

ETHICAL DECLARATIONS

Informed Consent: Written informed consent was obtained from all participants who participated in this study.

Referee Evaluation Process: Externally peer-reviewed.

Conflict of Interest Statement: The authors have no conflicts of interest to declare.

Financial Disclosure: The authors declared that this study has received no financial support.

Author Contributions: All of the authors declare that they have all participated in the design, execution, and analysis of the paper, and that they have approved the final version.

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