AACE Clinical Case Rep. 8 (2022) 181–182

Contents lists available at ScienceDirect

AACE Clinical Case Reports

journal homepage: www.aaceclinicalcasereports.com

Visual Vignette

Newly Diagnosed Hypoparathyroidism as the Initial Presentation of DiGeorge Syndrome in a 26-Year-Old Man

Anvay Shah, MBBS *, Bridget Sinnott, MD

Division of Endocrinology, Diabetes and Metabolism, Medical College of Georgia, Augusta, Georgia

ARTICLE INFO

Article history:
Received 30 August 2021
Received in revised form
24 January 2022
Accepted 3 February 2022
Available online 7 February 2022

Case Presentation

A 26-year-old African American man was seen for hypocalcemia identified during preoperative assessment for an elective left hemithyroidectomy for a symptomatic multinodular goiter. His albumin-corrected calcium level was 6.1 mg/dL (normal level: 8.7-10.4 mg/dL). He reported some leg cramping and dyspnea. His past history was notable for developmental delay, short stature, delayed eruption of teeth requiring dental surgery in elementary school, scoliosis requiring corrective surgery, and orchidopexy as an infant.

Physical examination was remarkable for thyromegaly, with the left lobe more enlarged than the right lobe, short stature, prominent forehead, thin eyebrows, bulbous enlargement of the nose, abnormal folding of the ear cartilage, displaced trachea, and scoliosis (Figs. 1 and 2). His height was 150 cm (below the midparental height) and weight was 50 kg. Cardiac examination was unremarkable. Chvostek and Trousseau signs were negative.

Laboratory data obtained preoperatively showed serum albumin-corrected calcium level of 6.1 mg/dL (1.57 mmol/L; normal level: 8.7-10.4 mg/dL), ionized calcium level of 2.8 mg/dL (1 mmol/L; normal level: 4.5-5.3 mg/dL), PTH level of 10 pg/mL (normal level: 11.1-79.5 pg/mL), 25-hydroxyvitamin D level of 12.21 ng/mL (normal level: 25- 80 ng/mL), TSH level of 1.758 μIU/mL (normal level: 0.4- 4.7 μIU/mL), free T4 level of 0.73 ng/dL (normal level: 0.58-1.76 ng/dL), creatinine level of 0.82 mg/dL (normal level: 0.60-1.60 mg/dL), and eGFR of >90 mL/min. X-rays of his skull and abdomen were normal, without any basal ganglia calcifications or nephrocalcinosis, respectively. Electrocardiography showed a QT interval of 440 milliseconds (normal: 400-440 milliseconds). He was started on calcium carbonate 1000 mg orally 3 times daily, ergocalciferol 50 000 IU oral capsule once per week, and calcitriol 0.25 μg oral capsule once a day.

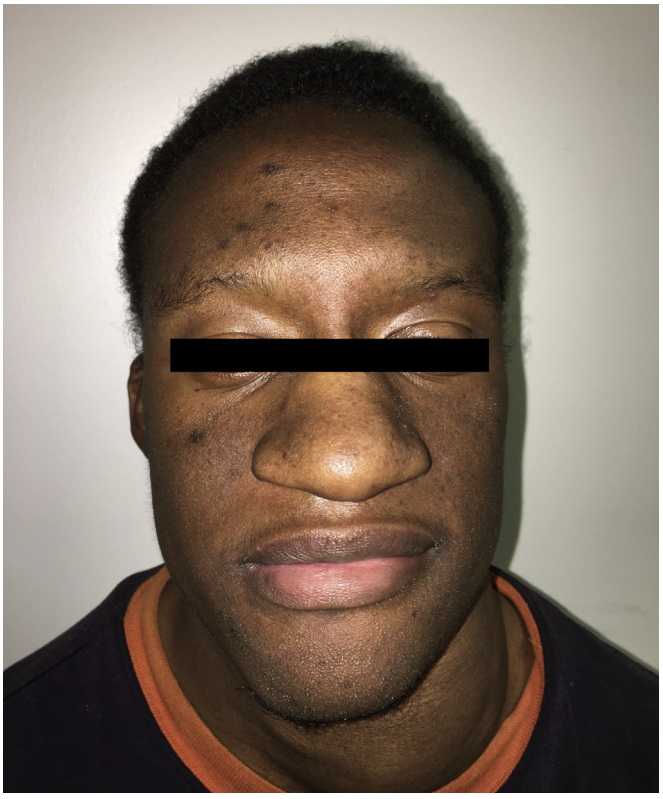

Fig. 1.

At the 2-week follow-up visit, the albumin-corrected calcium level was 7.1 mg/dL. Calcitriol was increased to 0.25 μg twice a day. The patient was continued on calcium carbonate and ergocalciferol, and repeat laboratory investigation showed albumin-corrected

Editor's Note: Submissions to "Visual Vignettes" are welcomed. Please submit online via the Journal's Editorial Manager site.

* Address correspondence to Dr Anvay Shah, Division of Endocrinology, Diabetes and Metabolism, Medical College of Georgia, Faculty Office Building, 1120 15th Street, HB-5025, Augusta, GA 30912.

E-mail address: anvayshah4299@gmail.com (A. Shah).

https://doi.org/10.1016/j.aace.2022.02.001

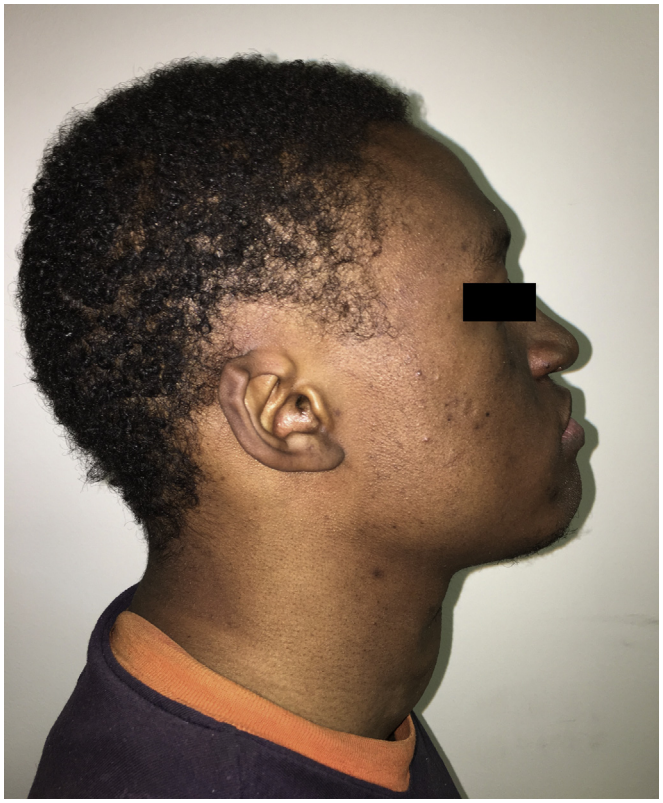

Fig. 2.

total calcium level of 9.1 mg/dL 1 month later and 9.4 mg/dL 2 months later. The patient subsequently underwent a hemithyroidectomy for his multinodular goiter, which showed an incidental 1.9-cm unifocal papillary carcinoma without any vascular invasion or lymphatic involvement.

What is the diagnosis?

Answer

Chromosome 22q11.2 deletion syndrome, also known as the DiGeorge syndrome.

The spectrum of this disorder includes, but is not limited to, aortic arch abnormalities, hypoparathyroidism, cleft lip/palate abnormalities, abnormal facial features, immunodeficiency, and intellectual disability. Although typically diagnosed in the early childhood, few patients may be diagnosed late because of subtle clinical findings.[1]

Our case was unique as the patient presented with hypocalcemia in adulthood, had no relevant cardiac findings, had some facial features, short stature, and scoliosis, and was later diagnosed to have papillary thyroid cancer. The facial features made us suspect the 22q11 microdeletion syndrome, and the diagnosis was later confirmed by fluorescence in situ hybridization. To our knowledge, this is the third reported case of papillary thyroid cancer with chromosome 22q11 deletion syndrome.[2,3]

Disclosure

The authors have no multiplicity of interest to disclose.

References

1. Cohen E, Chow EW, Weksberg R, Bassett AS. Phenotype of adults with the 22q11 deletion syndrome: a review. *Am J Med Genet*. 1999;86(4):359–365.
2. McDonald-McGinn DM, Reilly A, Wallgren-Pettersson C, et al. Malignancy in chromosome 22q11.2 deletion syndrome (DiGeorge syndrome/velocardiofacial syndrome). *Am J Med Genet A*. 2006;140(8):906–909.
3. Lambert MP, Arulselvan A, Schott A, et al. The 22q11.2 deletion syndrome: cancer predisposition, platelet abnormalities and cytopenias. *Am J Med Genet A*. 2018;176(10):2121–2127.