

Clinical Images/Spotters

Tall Stature with Bumpy Tongue: A Rare Polyendocrine Neoplasia Syndrome

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Received : 02 August 2021
Accepted : 30 August 2021
Published : 08 October 2021

DOI
10.25259/JPED_11_2021

Quick Response Code:



A 15-year-old girl presented with multiple nodular swellings over the tongue, lips, angle of the mouth, and bilateral upper eyelids. She was comparatively tall (height 160 cm, +0.6 Z) when compared to her mid-parental height (146 cm, -2.12 Z) and had a marfanoid habitus with bilateral ectropion on examination [Figure 1]. Due to the clinical phenotype and suspicion of the polyendocrine syndrome, investigations for medullary carcinoma thyroid (MTC) were done which showed high serum calcitonin 956 pg/mL (normal range < 10 pg/mL). Ultrasound thyroid revealed two intrathyroidal lesions with calcification [Figure 2] and biopsy of the lesion confirmed the diagnosis of MTC. The clinical features and lab abnormalities established the diagnosis of multiple endocrine neoplasia type 2B (MEN2B). MEN2B is a polyendocrine syndrome caused by activating mutations in the *RET* proto-oncogene and is characterized by MTC (100% cases), pheochromocytoma (50%), and a classic clinical phenotype which



Figure 1: (a) Nodular swellings over the lips, buccal mucosa, and tongue. (b) Demonstration of wrist sign in the same child. (c) Bilateral ectropion and nodular swellings in bilateral upper eyelids. (d) Marfanoid habitus with arm span longer than length. (e) Another image of nodular swellings on both the lips.

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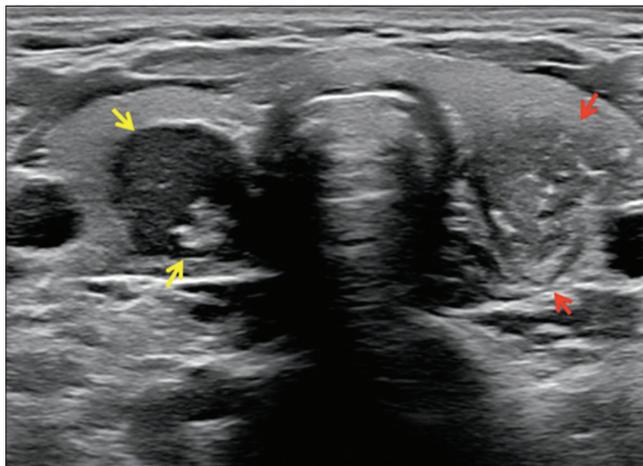


Figure 2: Axial ultrasound image of the thyroid gland showing a very hypoechoic nodule in the right lobe (yellow arrows) and slightly isoechoic to hypoechoic nodule in the left lobe (red arrows). Echogenic foci seen within both nodules represent calcification.

includes the presence of mucosal neuromas on the lips, tongue, or conjunctiva, marfanoid habitus, pes cavus, pectus

excavatum, high-arched palate, scoliosis, slipped capital femoral epiphysis, joint laxity, proximal muscle weakness, and thickened lips. Alacrima, ectropion, and ptosis can also be seen. Surgical treatment for thyroid and adrenal lesions offers the best chances of survival.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

How to cite this article: Yadav J, Gupta S, Sodhi K. Tall stature with bumpy tongue: A rare polyendocrine neoplasia syndrome. *J Pediatr Endocrinol Diabetes* 2021;1:30-1.