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CASE REPORT

Malnutrition Despite Adequate Nutrient Intake: A Clue For Diencephalic Syndrome

Yeterli Besin Alımına Karşı Yetersiz Büyüme: Diencefalik Sendrom için İpucu Olabilir

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ABSTRACT

Diencephalic syndrome is characterized by excessive weight loss and subcutaneous fatty tissue loss, hyperactivity, euphoria, and hypervigilance observed in early childhood. It usually accompanies space-occupying neoplastic lesions of the hypothalamic–optochiasmatic region. There is an inverse relationship between mass size and weight gain. In this case report, a patient aged 1 year and 2 months with hypothalamic–optochiasmatic glioma was presented. We aimed to draw attention to diencephalic syndrome, which is a rare cause of weight gain and vomiting.

Keywords: Diencephalic Syndrome, Severe Malnutrition, Tuberosclerosis type 1

ÖZ

Diensefalik sendrom, erken çocukluk döneminde gözlenen aşırı kilo kaybı ve deri altı yağ dokusu kaybı, hiperaktivite, öfori ve hipervijilans ile karakterizedir. Genellikle hipotalamik optokiazmatik bölgenin yer kaplayan neoplastik lezyonlarına eşlik eder. Kitle büyüklüğü ile kilo alımı arasında ters oran vardır. Bu olgu sunumunda hipotalamik-optokiazmatik gliomalı 1 yıl 2 aylık bir hasta sunuldu. Kilo alma ve kusmanın nadir bir nedeni olan diensefalik sendroma dikkat çekmeyi amaçladık.

Anahtar Kelimeler: Diensefalik Sendrom, Ağır Yetersiz Beslenme, Tüberoskleroz tip 1

Introduction

Diencephalic syndrome (DS) is a condition that often develops in infancy and early childhood. DS is associated with space-occupying lesions in the supratentorial midline region of the anterior hypothalamus (1, 2). It is often accompanied by recurrent persistent vomiting and nystagmus. DS is characterized by signs and symptoms caused by hypothalamic insufficiency secondary to space-occupying lesions of the hypothalamic-optic chiasmatic region. Growth hormone (GH) hypersecretion has often been described in patients with DS (2, 3). Despite adequate caloric intake, weight loss and cachexia with hypervigilance and hyperactivity are typical findings for DS (3). We aimed to present a 14-month-old case diagnosed with DS secondary to optic glioma (OG) of neurofibromatosis type 1 (NF type 1) with respect to the rarity of the condition in clinical practice.

Case

A 14-month-old girl with NF type 1 presented with complaints of restlessness, insomnia, swelling in both eyes, vomiting and inability to gain weight. In her family history, the patient's father and uncle were also NF type 1. In the patient's history, she had restlessness, unexplained crying spells, vomiting and weight loss since she was five months old. The daily nutritional requirement was supported as 120 kcal per kilogram of infant enteral product. Her weight, height and head circumference were 5500 gr (0,03 percentile, SD: −5.44), 74 cm (0.03 percentile, SD: −3.4) and 44 cm (0.33 percentile, SD: −2.72), respectively. On physical examination, there were bilateral horizontal nystagmus and proptosis in the eyes (Figure 1). In addition, ophthalmological examination revealed pallor of the optic disk in the fundus and a 1.5 x 1.5 cm nodule due to neurofibroma in the left frontal region. The patient also had a significant decrease in subcutaneous adipose tissue, flattening of the nasal

Table 1: Anthropometric measurements of the patient

Age (month)	Weight (gram)	Weight (percentile)	Weight (SDS)	Height (cm)	Height (percentile)	Height (SDS)	WFA (%)	HFA (%)	HFW (%)
22	5700	<0,02	-5,44	74	0,03	-3,4	45	86	64
28	5600	<0,02	-6,47	74	<0,02	-4,48	43	82	62
32	5500	<0,02	-6,58	76	<0,02	-4,37	40	83	57
37	5,850	<0,02	-6,58	76	<0,02	-4,94	41	79	59
39	6300	<0,02	-6,66	78	<0,02	-4,72	42	78	63
42	6200	<0,02	-7,47	79	<0,02	-4,8	41	80	56
44	7400	<0,02	-6,1	81	<0,02	-4,51	49	79	68
51	8750	<0,02	-5,14	81	<0,02	-5,24	51	76	83

Abbreviation: WFA, weight for age; HFA, height for age; HFW, height for weight

root and multiple café-au-lait spots on the whole body's skin (Figure 2). Upon neurological examination, no object tracking was observed, and the patient was found to be hyperactive. After the exclusion of other reasons explaining the weight loss of the patient with a neurofibromatosis type 1 diagnosis, cranial magnetic resonance imaging (MRI) showed bilateral optic glioma and a mass lesion with intense contrast. In the glucagon stimulation test, the growth hormone peak was insufficient (GH peak: 2.73 µg/L). The lesions filled the suprasellar cistern and caused compression at the base of the third ventricle (Figures 3, 4); therefore, gross total mass excision was performed with a left frontotemporal craniotomy. Histopathological examination of the surgically resected mass revealed neurofibroma. The patient was diagnosed with DS due to the presence of a mass in the hypothalamic region and severe malnutrition. During the patient's follow-up, a third ventriculostomy was performed due to the development of hydrocephalus in the fifth month after the first operation.

Since there was excessive weight loss despite having an adequate nutritional intake via the oral route, she underwent enteral feeding via a percutaneous endoscopic gastrostomy (PEG) tube. The patient was followed up with anthropometric measurements (Table I) and cranial MRI examinations.

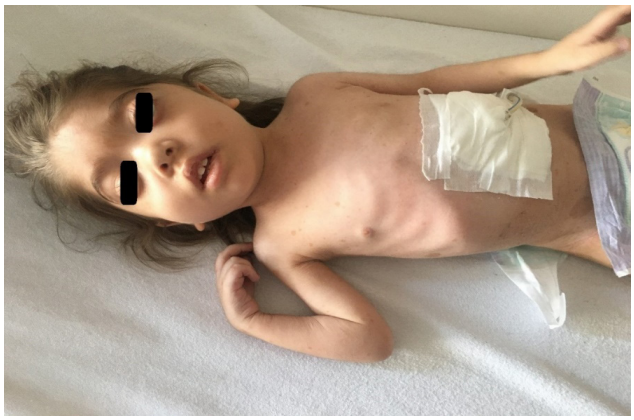

Figure 1: A decreased amount of subcutaneous adipose tissue, cachectic appearance, and proptosis in the left eye

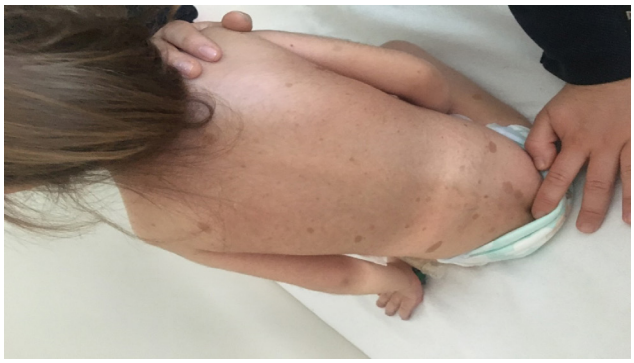

Figure 2: > 6 café-au-lait spots on the entire back

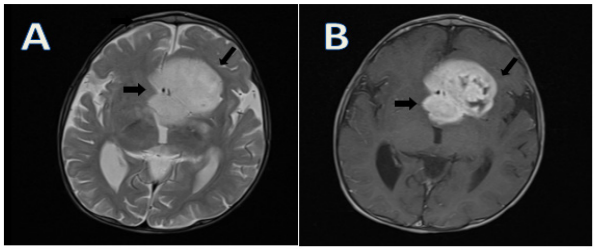

Figure 3: Cranial magnetic resonance imaging (MRI) showing a mass filling the suprasellar cistern and causing compression at the base of the third ventricle (A), and a mass lesion with intense contrast enhancement (B)

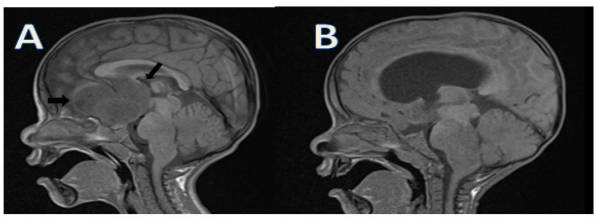

Figure 4: Cranial MR images of the patient before (A) and after treatment (B)

Discussion

NF type 1 is an autosomal dominant genetic disorder that causes a predisposition to OCG (4). NF type 1 affects nearly every organ system in the body with

broad clinical ramifications, such that children and adults with this condition may exhibit pigmentary abnormalities (café-au-lait macules, skinfold freckling, Lisch nodules), tumors of the peripheral and central nervous system (neurofibromas and gliomas), learning and attention problems, bone abnormalities (long bone dysplasias, scoliosis), seizures, sleep disturbances, vasculopathies and non-nervous system cancers (breast cancer, pheochromocytoma). In our patient, the presence of more than six café-au-lait spots on the skin and optic nerve tumor, together with her family history, confirmed the diagnosis of NF type 1 (5). DS has rarely been reported in children with NF type 1 and OCG. The development of hypothalamic-optochiasmatic-space occupying lesions in early childhood can lead to DS with extreme weight loss and restlessness (6). The most important symptom of the syndrome is excessive weight loss, despite having an adequate nutritional intake; this symptom is observed in almost all cases (3). Our case presented with complaints of weight loss and extreme restlessness despite having adequate nutritional intake. Tumors located in the third ventricle most frequently cause DS, followed by tumors in the optic nerves and chiasm, fourth ventricle and hypothalamus region, in that order (6). Our case had a neurofibroma in the hypothalamus and bilateral OCG.

It has been reported that the mean age of DS development in patients with NF type 1 is older. Only one patient younger than 12 months has been reported in the literature (6). Our case was one year and two months old at the time of admission. Although patients usually present with severe vomiting, severe weight loss, hydrocephalus and nystagmus, they may sometimes present with only growth percentile loss (7). Our case presented with severe vomiting and severe weight loss. The cause of weight loss in these patients remains unknown. However, increased GH levels and related pathways due to GH resistance are blamed in endocrinological examinations (8, 9). Fleishman et al. (10) showed that baseline GH levels were high in 9 of their 11-case series; there was insufficient suppression of the response to the GH in all cases that underwent an oral glucose tolerance test, and IGF-1 levels were within the normal limit. However, an insufficient response to the glucagon stimulation test performed during the endocrinological follow-up was observed in our case (peak GH: 2.72 µg/L). In these patients, a careful ophthalmologic examination is required to detect nystagmus, strabismus and optic atrophy, among others. Additionally, a neuroradiological examination should be performed based on the results of the ophthalmological examination. A careful ophthalmologic examination is crucial to detect symptoms such as nystagmus, strabismus and optic atrophy with the support of neuroradiological investigation. Neuroradiology, especially MRI, can describe the location, extension and association of hydrocephalus or other orbital pathologies.

Since these tumors in the optic chiasma of children under three years of age are usually very aggressive, neurosurgical intervention is necessary in children with symptomatic increases in intracranial pressure caused by the mass effect and in whom chemotherapy has failed. The cause of the symptomatic increase in intracranial pressure in these cases is often obstructive hydrocephalus. Although excising the giant neurofibroma mass in our case was sufficient to prevent hydrocephalus in the first step, hydrocephalus that developed in the fifth postoperative month also had to be performed because the tumor had an aggressive course.

Consequently, the appearance of DS syndrome symptoms should prompt the clinician to perform cranial MRI to rule out OCG with hypothalamic involvement. For this reason, intracranial pathologies should be considered in children with normal appetites who show decreased weight gain and growth-percentage losses.

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