Malnutrition Despite Adequate Nutrient Intake: A Clue For Diencephalic Syndrome

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

1Anna Carina Ergani, 2Ayşe Kartal, 3Fuat Buğrul, 4Mert Şahinoğlu, 1Meltem Gümüş, 1Hakan Karabağlı, 1Halil Haldun Emiroğlu

Selçuk University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Gastroenterology, Konya, Turkey
Selçuk University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Neurology, Konya, Turkey
Selçuk University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Endocrinology, Konya, Turkey
Selçuk University Faculty of Medicine, Department of Surgery, Neurosurgery Unit, Selçuklu Neurosurgery Center, Konya, Turkey

Correspondence
Anna Carina Ergani, Selçuk University Faculty of Medicine, Department of Pediatrics, Division of Pediatric Gastroenterology, Konya, Turkey
E-Mail: drannaergani@gmail.com

How to cite?
Ergani A. C., Kartal A., Buğrul F., Şahinoğlu M., Gümüş M., Karabağlı H., Emiroğlu H. H. Diencephalic Syndrome in A Patient With Severe Malnutrition. Tuberosclerosis type 1

CASE REPORT

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–optic chiasmatic 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–optic chiasmatic 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


Anahtar Kelimeler: Diencefalık 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
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, the patient underwent enteral feeding via a percutaneous endoscopic gastrostomy (PEG) tube. The patient was followed up with anthropometric measurements (Table I) and cranial MRI examinations.

**Table 1: Anthropometric measurements of the patient**

<table>
<thead>
<tr>
<th>Age (month)</th>
<th>Weight (gram)</th>
<th>Weight (percentile)</th>
<th>Weight (SDS)</th>
<th>Height (cm)</th>
<th>Height (percentile)</th>
<th>Height (SDS)</th>
<th>WFA (%)</th>
<th>HFA (%)</th>
<th>HFW (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>22</td>
<td>5700</td>
<td>&lt;0.02</td>
<td>-5.44</td>
<td>74</td>
<td>0.03</td>
<td>-3.4</td>
<td>45</td>
<td>86</td>
<td>64</td>
</tr>
<tr>
<td>28</td>
<td>5600</td>
<td>&lt;0.02</td>
<td>-6.47</td>
<td>74</td>
<td>&lt;0.02</td>
<td>-4.48</td>
<td>43</td>
<td>82</td>
<td>62</td>
</tr>
<tr>
<td>32</td>
<td>5500</td>
<td>&lt;0.02</td>
<td>-6.58</td>
<td>76</td>
<td>&lt;0.02</td>
<td>-3.37</td>
<td>40</td>
<td>83</td>
<td>57</td>
</tr>
<tr>
<td>37</td>
<td>5850</td>
<td>&lt;0.02</td>
<td>-6.58</td>
<td>76</td>
<td>&lt;0.02</td>
<td>-3.94</td>
<td>41</td>
<td>79</td>
<td>59</td>
</tr>
<tr>
<td>39</td>
<td>6300</td>
<td>&lt;0.02</td>
<td>-6.66</td>
<td>78</td>
<td>&lt;0.02</td>
<td>-3.72</td>
<td>42</td>
<td>78</td>
<td>63</td>
</tr>
<tr>
<td>42</td>
<td>6200</td>
<td>&lt;0.02</td>
<td>-7.47</td>
<td>79</td>
<td>&lt;0.02</td>
<td>-4.8</td>
<td>41</td>
<td>80</td>
<td>56</td>
</tr>
<tr>
<td>44</td>
<td>7400</td>
<td>&lt;0.02</td>
<td>-6.1</td>
<td>81</td>
<td>&lt;0.02</td>
<td>-4.51</td>
<td>49</td>
<td>79</td>
<td>68</td>
</tr>
<tr>
<td>51</td>
<td>8750</td>
<td>&lt;0.02</td>
<td>-5.14</td>
<td>81</td>
<td>&lt;0.02</td>
<td>-5.24</td>
<td>51</td>
<td>76</td>
<td>83</td>
</tr>
</tbody>
</table>

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

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...
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.

References