spontaneous diuresis and renal function. Treatment duration ranged between 16 and 155 h, with a median of 27 h. We did not detect any complications associated with the technique, such as infection or catheter malfunction (Table 1).

In this sample of patients, we found that CFPD with a dual catheter system for RRT was easy to implement, efficient, and safe due to the absence of associated complications.

References


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Ketogenic diet as a therapeutic option in refractory epilepsy

Dieta cetogénica como opción terapéutica en la epilepsia refractaria

Dear Editor:

Refractory epilepsy (RE) is defined as epilepsy with poor seizure control despite treatment with anticonvulsant medication. It accounts for 25%–30% of epilepsy cases, and poses a significant therapeutic challenge from a medical perspective.1–6 One of the effective therapeutic alternatives for RE is the ketogenic diet (KD).1–6 The KD consists of a diet rich in fat and low in protein and carbohydrate aimed at increasing the levels of ketone bodies.1–6 Several theories have been proposed to explain its mechanism of action, none of them conclusive.1–3,6

Despite the popularity the diet has achieved in the past twenty years, there is still no consensus on how the KD must be managed to achieve maximum efficacy.

Table 1 Characteristics of the patients under study.

<table>
<thead>
<tr>
<th>Patient (number)</th>
<th>Genetic cause</th>
<th>Sex</th>
<th>Age of onset</th>
<th>No. seizures/day</th>
<th>Status epilepticus before diet</th>
<th>Total number of AEDs used before diet</th>
<th>Other previous non-AED treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Dravet syndrome</td>
<td>♂</td>
<td>3–6 months</td>
<td>&gt;50</td>
<td>Yes</td>
<td>5</td>
<td>γ globulin</td>
</tr>
<tr>
<td>2</td>
<td>Dravet syndrome</td>
<td>♂</td>
<td>3–6 months</td>
<td>&lt;10</td>
<td>Yes</td>
<td>7</td>
<td>None</td>
</tr>
<tr>
<td>3</td>
<td>Rett syndrome</td>
<td>♂</td>
<td>3–6 months</td>
<td>&gt;50</td>
<td>No</td>
<td>9</td>
<td>ACTH</td>
</tr>
<tr>
<td>4</td>
<td>SCNZA encephalopathy</td>
<td>♂</td>
<td>&lt;1 month</td>
<td>&gt;50</td>
<td>No</td>
<td>7</td>
<td>None</td>
</tr>
<tr>
<td>5</td>
<td>Dup 15q13.3 multifocal encephalopathy</td>
<td>♂</td>
<td>4 years</td>
<td>&lt;10</td>
<td>Yes</td>
<td>3</td>
<td>Prednisolone</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Patient (number)</th>
<th>Structural cause</th>
<th>Sex</th>
<th>Age of onset</th>
<th>No. seizures/day</th>
<th>Status epilepticus before diet</th>
<th>Total number of AEDs used before diet</th>
<th>Other previous non-AED treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>6</td>
<td>Thalamic infarction</td>
<td>♂</td>
<td>4 years</td>
<td>20–50</td>
<td>No</td>
<td>7</td>
<td>γ globulin</td>
</tr>
<tr>
<td>7</td>
<td>Extensive malformation of cortical development in both hemispheres</td>
<td>♂</td>
<td>3–6 months</td>
<td>20–50</td>
<td>No</td>
<td>5</td>
<td>ACTH</td>
</tr>
</tbody>
</table>

♂, male; ♂, female; ACTH, adrenocorticotropic hormone; Dup, duplication; AED, antiepileptic drug.

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Table 2  Summary of study outcomes.

<table>
<thead>
<tr>
<th>Patient</th>
<th>Diet ratio</th>
<th>Cognitive outcome</th>
<th>Sleep</th>
<th>Reduction in number of seizures</th>
<th>EEG</th>
<th>Status epilepticus after TX</th>
<th>Time seizure-free</th>
<th>Side effects</th>
<th>Duration of TX (months)</th>
<th>Reason for discontinuation</th>
<th>General satisfaction</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>3:1</td>
<td>Mild improvement</td>
<td>Worsening</td>
<td>&gt;75%</td>
<td>No changes</td>
<td>No</td>
<td>1 month</td>
<td>Vomiting, irritability, insomnia</td>
<td>13</td>
<td>Subsequent ineffectiveness</td>
<td>Very satisfied</td>
</tr>
<tr>
<td>2</td>
<td>3:1</td>
<td>Marked improvement</td>
<td>Marked improvement</td>
<td>No changes</td>
<td>No changes</td>
<td>No</td>
<td>1 month</td>
<td>Vomiting</td>
<td>9</td>
<td>Not completed</td>
<td>Satisfied</td>
</tr>
<tr>
<td>3</td>
<td>3:1</td>
<td>Mild improvement</td>
<td>Mild improvement</td>
<td>No changes</td>
<td>No changes</td>
<td>No</td>
<td>1 week</td>
<td>None</td>
<td>12.5</td>
<td>Subsequent ineffectiveness</td>
<td>Moderately satisfied</td>
</tr>
<tr>
<td>4</td>
<td>4:1</td>
<td>No changes</td>
<td>No changes</td>
<td>&gt;75%</td>
<td>No changes</td>
<td>No</td>
<td>3 months</td>
<td>None</td>
<td>4</td>
<td>Subsequent ineffectiveness</td>
<td>Moderately satisfied</td>
</tr>
<tr>
<td>5</td>
<td>4:1</td>
<td>No changes</td>
<td>Worsening</td>
<td>&gt;75%</td>
<td>No changes</td>
<td>No</td>
<td>None</td>
<td>Vomiting, constipation, changes in eating patterns</td>
<td>2</td>
<td>Family rejection</td>
<td>Dissatisfied</td>
</tr>
<tr>
<td>6</td>
<td>3:1</td>
<td>No changes</td>
<td>No changes</td>
<td>&lt;50%</td>
<td>No changes</td>
<td>No</td>
<td>None</td>
<td>None</td>
<td>3</td>
<td>Family rejection</td>
<td>Dissatisfied</td>
</tr>
<tr>
<td>7</td>
<td>3:1</td>
<td>Marked improvement</td>
<td>No changes</td>
<td>50–75%</td>
<td>No changes</td>
<td>No</td>
<td>None</td>
<td>None</td>
<td>12</td>
<td>Subsequent ineffectiveness</td>
<td>Satisfied</td>
</tr>
</tbody>
</table>

EEG, electroencephalogram; TX, treatment.

* Patients numbered consistently with the previous table.

b Initial effectiveness with recurrence/worsening of seizures over time.
epilepsy and after trying multiple antiepileptic drugs (>6 AEDs).

The side effects were similar to those described in the literature, with vomiting being the most frequent complication. None of the side effects led to discontinuation of the diet. We found fewer side effects in association with the 3:1 diet ratio.

Although this is a retrospective study with a small sample, we can conclude that the KD is efficacious for the treatment of RE. However, more randomised studies are needed to improve its implementation and prevent complications.

Acknowledgments

We want to thank our patients, who motivate us day after day. For their smiles.

References


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Gaucher disease: Enzyme replacement treatment initiated at pediatric age; 20-year experience∗

Enfermedad de gaucher: tratamiento enzimático sustitutivo iniciado en la edad pediátrica. Experiencia de 20 años

Dear Editor:

Gaucher disease (GD) is caused by deficiency of the enzyme acid beta-glucosidase (GBA). The currently accepted classification is: type 1 (non-neuropathic) (GD-1), type 2 (acute neuropathic) (GD-2) and type 3 (chronic neuropathic). Since 1994, enzyme replacement treatment (ERT) has been available in Spain. Due to the limited long-term safety and efficacy experience with ERT, it was of interest to describe the experience in a tertiary pediatric hospital in which, this treatment started being used as soon as it was available.

The evolution of 7 patients with GD (5 GD-1 and 2 GD-2) treated within the past 20 years in Hospital Infantil La Fe (Valencia) was retrospectively described. The diagnosis was confirmed by glucocerebrosidase activity in leukocytes (GAL) and by mutations in the GBA gene. At diagnosis, and yearly thereafter, weight, height, BMI, hepatomegaly–splenomegaly degree, hemogram, hemostasis, chitotriosidase, chemokine ligand 18 (CCL18) and spine densitometry were assessed. Abdominal magnetic resonance imaging (MRI) was performed to assess liver and spleen volumes, calculating the multiples of their normal volumes (MNV). In patients with bone involvement, MRI of long bones was performed. ERT was administered by iv infusion for 2 h, every 2 weeks at a 60UI/kg dose in patients with GD-1.

Table 1 shows evolution of the 5 patients with GD-1. Age at diagnosis ranged between 22 months and 11 years. In every case, bone marrow aspiration showed Gaucher cells. Genetic analysis showed that 3 of the patients carried the mutation N370S in heterozygosis and 2 the mutation L444P. Every patient showed hepatosplenomegaly and anemia. The initial hepatomegaly was 1.9–3.5 times MNV, and splenomegaly was 9–65 times MNV. One patient presented with lesions of bone infarction and two had Erlenmeyer-flask shaped femurs. GAL ranged from 0.26–2.87 nM/mg prot.h (lower than 8% of control GAL in every case). Patients 1 and 2 received treatment with alglucerase from 1995 to 1998 and with imiglucerase thereafter. Every patient improved his/her weight and height, hemoglobin and platelets count. ERT reduced the hepatic volume up to 3 times and that of the spleen 4–15 times. These improvements occurred mainly during the first 12–24 months of treatment. The patients did not need blood transfusions. In 2009, due to the 6-month shortage of imiglucerase, 2 patients started taking miglustat. Both patients showed an increase in chitotriosidase and CCL18; both biomarkers returned to normal values when ERT was reintroduced at previous doses. These patients did not show hematologic or bone involvement and no modification