Enteral Nutrition in Children with Recessive Dystrophic Epidermolysis Bullosa

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Nutrition problems in children with RDEB

- **Nutritional risk in children with RDEB:**
  - Decreased feeding capacities
    - Bullae and erosions affect oropharynx/oesophagus
    - Microstomia and teeth dystrophy
    - Oesophageal strictures/stenosis…
  - Increased energy and protein needs
    - Accelerated skin turn-over
    - Blood and plasma losses: proteins, micronutrients
    - Chronic inflammation, infections…
Nutrition problems in children with RDEB

Intakes

Losses

Malnutrition
Nutrition problems in children with RDEB

Nutritional depletion in children with RDEB:

• Negative balance for macronutrients and micronutrients
  – Proteins, carbohydrates, lipids
  – Trace-elements (++ zinc, iron, selenium)
  – Vitamins

• Growth and puberty retardation
  – Impaired weight and height gain
  – Delayed puberty, short adult stature

• Morbidity due to malnutrition

• Force feeding!
Nutrition problems in children with RDEB

« Children with dystrophic forms of EB and oropharyngeal lesions often evidence the features of chronic malnutrition. Nutritional assessment plays a key-role in the long-term management of these patients. »
Gruskay et al. Arch Dermatol, 1988

« While nutrition cannot in itself close their wounds, the development of nutritional deficiencies will not allow patients to successfully heal their wounds and return to a more normal hemodynamic and metabolic profile. »
Gamelli RL. Arch Dermatol, 1988
Nutrition problems in children with RDEB

- Problems related to long-term nasogastric feeding tube in patients with RDEB
  - Placement
  - Day-to-day management
  - Disfigurement

- Fears about gastrostomy: poor healing and infection risk at the insertion site!
Nutrition care in children with RDEB

• The first report on gastrostomy feeding in 18 children with RDEB (1989-1994):

«Our observations suggest that gastrostomy feeding can play a valuable role in severe DEB and is associated with minimal mortality. Such intervention is best undertaken before growth failure is established, and prior to puberty.»

Great Ormond Street Hospital for Children, London
Nutrition care in children with RDEB

• Ten years of experience on gastrostomy feeding and nutrition care in children with RDEB at Necker-Enfants Malades Hospital in Paris

• The collaboration between dermatologists and nutrition specialists, physicians and dieticians
Nutrition care in children with RDEB

• Retrospective study
• Over a 7 year-period (1997-2004)
• 24 patients with RDEB referred to the Dermatology department at Necker-Enfants Malades Hospital in Paris

• 12 patients underwent gastrostomy placement and received enteral nutrition through gastrostomy tube
Nutrition care in children with RDEB

• 11 children (8 girls) : 9.0 ± 5.8 years
• 1 young man : 18 years
• At the date of GT, all children presented with growth retardation
• Age at the time of growth retardation onset :
  – Weight : 3.1 ± 2.7 years
  – Height : 4.9 ± 2 years
• Delay between the first signs of growth retardation and the GT placement : 5.5 ± 4.2 years (0.5 - 14)
Nutrition care in children with RDEB

Growth features at the date of GT

• W : $-2.3 \pm 1.0$ SD
  - loss of $2.0 \pm 1.1$ SD vs best W

• H : $-1.0 \pm 1.2$ SD
  - loss of $1.5 \pm 0.9$ SD vs best H

• W/H : $81 \pm 11\%$

• H/age : $95 \pm 5\%$
Classification of chronic malnutrition in children

<table>
<thead>
<tr>
<th>Malnutrition</th>
<th>Absence</th>
<th>Moderate</th>
<th>Severe</th>
</tr>
</thead>
<tbody>
<tr>
<td>H/age (%)</td>
<td>&gt; 95</td>
<td>85-95</td>
<td>&lt; 85</td>
</tr>
<tr>
<td>W/H (%)</td>
<td>&gt; 90</td>
<td>80-90</td>
<td>&lt; 80</td>
</tr>
</tbody>
</table>

*From Waterlow*
Nutrition care in children with RDEB

Prior to the decision to perform GT:

• All the patients experienced dysphagia
• 9 had undergone oesophageal dilatation
• None had received nasogastric feeding
• All had received regular advices from a dietician
Nutrition care in children with RDEB

Nutritional supplies :

• At onset, EN provided $67 \pm 29\%$ of the upper RDA for total energy and and $170 \pm 96\%$ of the upper RDA for proteins (for age and gender)

• At the date of study, the mean EN duration was $53 \pm 20$ mths (12-89 mths)

• At the date of study, EN provided $79 \pm 28\%$ of the upper RDA for total energy and $216\pm 134\%$ of the upper RDA for proteins (for age and gender)
Nutrition care in children with RDEB

Outcome while on EN through GT:

- Oral feeding became impossible in 8 cases because of oesophageal stricture
- Catch-up growth in all the children
- Three girls completed puberty while on GF (age of menarche 13.3, 13.9 and 15.9 y)
- Two girls presented with delayed puberty (aged 16.7 and 17.9 y)
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Outcome while on EN:

- Best W obtained: $-0.6 \pm 1.5$ SD
- W gain: $+1.3 \pm 1.3$ SD
- Best H obtained: $+0.1 \pm 1.3$ SD
- H gain: $1.0 \pm 1.1$ SD
- W/H: $92 \pm 15\%$ at the date of study
- H/A: $98 \pm 5\%$ at the date of study
Antoine

GT performed at 1 y of age
GT performed at 8.5 y of age
Sophie

GT performed at 13.5 y of age
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Outcome while on GT feeding:

• 8 patients never presented with skin complications at the GT insertion site
• Moderate skin erosions in 4 children
• All GT could be used until the date of study
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Children without GT feeding:

- 12 children with RDEB
- 3 lost for follow-up ≥ 1 year at the update
- At the date of study, 9 children (4 girls): 6.6 ± 2.6 year-old
- 7 children underweighted: – 0.9 ± 1.4 SD
  – loss of 1.2 ± 1.0 SD vs the best W
- Growth velocity stable in 3 children for W and H and in 2 other children for H only
Nutrition care in children with RDEB

Children without GT feeding:

• Age at the time of growth retardation onset:
  – W: 2.0 ± 2.4 years (6 children)
  – H: 1.8 ± 1.2 years (4 children)

• W: −0.9 ± 1.4 SD

• H: +0.2± 1.5 SD (loss of 0.5 ± 0.8 SD vs best H)

• W/H: 84 ± 11%

• H/age: 100 ± 5%

• Spontaneous total energy intake # 110 ± 9% of the upper RDA for gender and age
### Nutrition care in children with RDEB

#### Outcome in children with and without GT feeding

<table>
<thead>
<tr>
<th></th>
<th>Patients without GT</th>
<th>Patients with GT at GF onset</th>
<th>Patients with GF at d.o study</th>
<th>+/-</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>W (SD)</strong></td>
<td>- 0.9 ± 1.4</td>
<td>- 2.3 ± 1.0</td>
<td>-1.2 ± 1.5</td>
<td>+ 1.3 ± 1.3</td>
</tr>
<tr>
<td><strong>H (SD)</strong></td>
<td>+ 0.2 ± 1.5</td>
<td>- 1.0 ± 1.2</td>
<td>- 0.2 ± 1.4</td>
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<td><strong>W/H (%)</strong></td>
<td>84 ± 11</td>
<td>81 ± 11</td>
<td>92 ± 15</td>
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<tr>
<td><strong>H/age (%)</strong></td>
<td>100 ± 5</td>
<td>95 ± 5</td>
<td>98 ± 5</td>
<td></td>
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</table>
Nutrition care in children with RDEB

Children with RDEB benefit from long-term GT feeding:

- Catch-up growth and normal growth velocity
- Quality of life
Nutrition care in children with RDEB

The timing to decide and perform GT should be adapted on an individual basis

• Growth velocity
• Feeding behaviour
• Parents’ demand…

But always before the occurrence of severe malnutrition and growth retardation, with special attention given to the puberal period
Nutrition care in children with RDEB

• Nutrition issues should be discussed with the parents as soon as possible

• GT should be presented and considered as a tool which might belong to the treatment if necessary, (in about 50% patients ?), never as a threat which « justify » force feeding!

• Surgical GT procedure is recommended

• Nutrition specialists should belong to the multidisciplinary team in charge of patients with RDEB