Nusinersen and children with Spinal Muscular Atrophy type 1: do they still need Paediatric Palliative Care?

Rome - October 26, 2018

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Spinal Muscular Atrophy (SMA)

SMA
Neuromuscular disorder
Autosomal recessive: deletion in SMN1 gene
Weakness and paralysis

Same GENOTYPE different PHENOTYPE

SMA 1
NON-SITTERS

SMA 2
SITTERS

SMA 3-4
AMBULANTS

www.togetherinsma-hcp.com
Spinal Muscular Atrophy (SMA)

Most severe form

Classification:
- SMA 1A: onset < 1 months
- SMA 1B: onset < 4 months
- SMA 1C: onset < 6 months
Consensus statement for standard of care in SMA 2017


Finkel, R. S. et al. «Diagnosis and management of spinal muscular atrophy: Part 2: Pulmonary and acute care; medications, supplements and immunizations; other organ systems; and ethics.» Neuromuscul.Disord. (2018).
Innovative Therapies

D. Kariyawasam, K. Carey, K.J. Jones, M.A. Farrar, New and developing therapies in spinal muscular atrophy, Paediatric Respiratory Reviews (2018)
Aims and Methods

Evaluate if a curative treatment alters children’s and families’ needs and if they could still benefit from Pediatric Palliative Care.
Aims and Methods

Evaluate if a curative treatment alters children’s and families’ needs and so if they could benefit from Pediatric Palliative Care

Retrospective medical records review

Population

Children with SMA 1 referred to our Hospital for Nusinersen (October 2017-June 2018)

Data collected:

- Age
- Sex
- Respiratory status
- Feeding status
- Basal motor ability with CHOP INTEND
- Anticipatory plans/DNR
- PICU
- Death
## Results

### Characteristics of Population

<table>
<thead>
<tr>
<th>Sex</th>
<th>n. (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Male</td>
<td>7 (47%)</td>
</tr>
<tr>
<td>Female</td>
<td>8 (53%)</td>
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</tbody>
</table>

<table>
<thead>
<tr>
<th>Age values</th>
<th></th>
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<tbody>
<tr>
<td>Median</td>
<td>22 mo</td>
</tr>
<tr>
<td>Range</td>
<td>3 mo – 15 yrs</td>
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<table>
<thead>
<tr>
<th>Type of SMA 1</th>
<th>n. (%)</th>
</tr>
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<tbody>
<tr>
<td>SMA 1A</td>
<td>3/15 (20%)</td>
</tr>
<tr>
<td>SMA 1B</td>
<td>7/15 (47%)</td>
</tr>
<tr>
<td>SMA 1C</td>
<td>5/15 (33%)</td>
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</tbody>
</table>

**Child’s death**
- None

![CHOP INTEND](chart.png)
Results

Respiratory assistance

- Suctioning: 15
- Cough Assist: 13
- NIV: 10
- IMV: 4
- No Ventilation: 1

Nutritional management

- PEG: 7
- NGT: 3
- PO: 5
Results

14/15 Consent to Nusinersen Therapy

4/15 started loading doses
10/15 maintenance doses after enrolment in trials

12/15 completed loading doses and proceeded in maintenance doses

1/15 Refusal for severity

1/14 Refusal for futility
1/14 Refusal for suffering
Results

Ethical Considerations

- Pediatric Palliative Care Consult: 15
- PICU Admission: 4
- Do Not Resuscitate Order: 3
- Ethic Committee: 1
# Limits

<table>
<thead>
<tr>
<th></th>
<th>Limit</th>
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<tbody>
<tr>
<td>1</td>
<td>Limited number</td>
</tr>
<tr>
<td>2</td>
<td>Limited time</td>
</tr>
<tr>
<td>3</td>
<td>Information to parents before consent</td>
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<tr>
<td>4</td>
<td>High pressure decision-making</td>
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</table>
Conclusions: PPC still essential

- **Multi-disciplinary approach**
  - Paediatric Palliative Care ➔ COORDINATION

- **Anticipatory Plans**
  - Paediatric Palliative Care ➔ DECISION-MAKING
  - MANAGING EXPECTATIONS

- **Change Of Culture**
  - Paediatric Palliative Care ➔ MODEL OF COMPLEMENTARY