

Providing supportive ventilation for a child with SMA type 1

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K

- Aged 4 months
- Severe respiratory distress
- Intubated, ventilated and retrieved to PICU

- “Extremely floppy”
- Absent deep tendon reflexes
- No head control

K

- Muscle biopsy
- Genetic testing
- Extubated on day 6 onto BiPAP
- Ongoing requirement for ventilatory support

K

- Transferred to the ward on BiPAP
- Discharged home aged 6 months
- Ongoing supportive ventilation at home

- Photo of K with her family

SMA(s)

- Autosomal recessive inheritance
- Mutation of the SMN gene at 5q11-13
- Types I II and III
- Type I – 80% die within their first year
- “The prognosis for each child must be defined not by the subtype of the disease but by individual disease progression”

Options for management

- Palliative care without ventilatory support
- Non-invasive ventilation
 - Nocturnal
 - With infective exacerbations
 - Long term day and night time use not felt to be appropriate
- Tracheostomy with invasive ventilation

Respiratory management of the infant with type 1 SMA, Bush et al ADC 2005

Outcomes

- 65 Patients with SMA1
- Tracheostomy (16)
 - Fewer hospitalisation until 3 yrs but more after 5yrs
 - 15 unable to breathe spontaneously or speak post tracheostomisation
 - Survival 73.8 ± 57 months
- NIV & Assisted Coughing (33)
 - 3 required continuous BiPAP
 - 4 unable to communicate verbally
 - Survival 41.8 ± 26 months

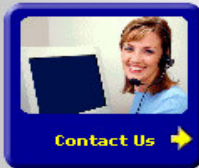
Spinal Muscular Atrophy Type 1: Management and Outcomes, Bach et al Ped Pulm 2002



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Welcome to our Web site

The Mission of the Jennifer Trust for Spinal Muscular Atrophy (JTSMA) is to be there for everyone affected by or involved with Spinal Muscular Atrophy (SMA), providing support and information, and investing in research. The JTSMA is the only British organisation dedicated to providing support, care, information and the funding of research into SMA.

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YOUR CHARITY



Your Help Needed

The JTSMA would like parents of an SMA child to complete a care survey to help identify and improve standards of care world-wide.

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We offer a wide range of services and information for people who are affected by SMA. By clicking on the buttons you can easily access our support and care services, information on research and how you can support our work.



The Key Issues

- Effective planning
- Early multidisciplinary assessment
- Medical assessment
- Maintain open channels of communication

Ethical Dilemmas

- Providing supportive ventilation for children with SMA type 1 is futile and wasteful of resources
- Futile? It depends what you are trying to achieve..

Ethical Dilemmas

- The quality of life of a child with SMA1 is so poor that by prolonging that life we are doing harm
 - Historically, physicians have been felt to underestimate the QOL of ventilated patients

MB's Quality of Life

- “MB enjoys lengthy visits from us, likes having his brother and sister sing to him and play tickling games with him, watching videos, and listening to music CDs”
- “MB's life is helpless and sad with almost relentless discomfort, periods of distress and relatively short episodes of pain”

Beneficence

Non-Maleficence

Autonomy

Justice

Conclusions

- Introducing NIV has improved B's quality of life
- This case sets a standard for future management
- We should consider NIV for children with SMA -1
- Where now?

Thank you