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"Heroic Medicine" in the Management of a Child with Spinal Muscular Atrophy Type 1: A Fair Choice of Treatment?

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ABSTRACT

Medicine is a rapidly changing field. The use of new drugs has given hope for patients' survival and comfort for the family, especially in cases of children with life-limiting conditions. This has influenced the clinical decision on the options of care and treatment by either the caregivers or healthcare professionals. Medical management for these patients is threaded on many ethical discussions to determine the best choice for these patients. We illustrated a case of a baby with spinal muscular atrophy (SMA) type 1 who was started on a new medication, leading to a renewed hope for the family but a challenging task for the managing team to decide on the highest ceiling of care for the patient.

Keywords: *Spinal muscular atrophy, Ethics, Heroic medicine, Life limiting disease, Novel therapy*

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INTRODUCTION

Spinal muscular atrophy (SMA) type 1 is a severe neuromuscular disorder that affects children and has been considered a life-limiting illness (LLI). The mainstay of the treatment is supportive therapy directed at providing nutrition, respiratory assistance and the prevention of complications. However, novel disease-modifying therapies are now available for infants with SMA. These agents have been reported to have neurologically improved the patients' condition. Most SMA patients are susceptible to various hospitalisations depending on the disease trajectory and status. What is best for the patient may depend on the interpretation by the healthcare professionals or parents. Collective decision making by both parties is an essential consideration in respecting family's wishes and preferences, thus reaching agreed goals of care. In the context of palliation during the patient's care, the focus should be on comfort care, maximising the quality of life and reducing suffering despite the presence and availability of the novel treatment.

CASE HISTORY

A 7-month-old boy with SMA type 1 was referred for follow-up at the clinic. He was

a 39-weeker baby with a birth weight of 3.4 kg and had to be ventilated briefly at birth. The maternal chorionic villous sampling at 16 weeks confirmed the diagnosis of SMA type 1. Investigation was initiated as both parents were known to be SMA gene carrier and the history of the passing of his older sibling at 2-month of age with SMA. He presented at this episode with poor respiratory effort and was suspected to have pneumonia, hence, required intubation and antibiotic therapy. His ventilatory setting remained static leading to a failed extubation and weaning off the ventilator. The discussion among the multi-disciplinary team has led to a consensus agreement on the continuation of his ventilation management and the patient was enrolled on the "Compassionate Use Programme". This was a lottery schedule to start him on a novel medical therapy for the next three years. He also underwent tracheostomy after two months on the ventilator. It was planned that he would have been weaned off onto non-invasive ventilation at home once the drug took effect on him. The parents had hoped for a miracle despite a guarded prognosis for the infant. The discussion on palliation was deferred until a further medical crisis occurred.

There are two important issues that need to be addressed when discussing the shifting goals of care for a life-sustaining intervention among children with LLI.

a. What is the appropriate decision when the LLI status changed and considered curable? How do we discuss parental hope on the new therapies for their child?

b. How do we decide on the ceiling of care when the new promising drug therapies are available for LLI? Should we aggressively go for an all-out treatment?

DISCUSSION

SMA type 1 is known as LLI. The life span for those with this condition is virtually short and prolonging the life would be burdensome to the patients and the caretakers, psychologically and emotionally. The focus of care should be aimed for the patients' best interest and avoidance of potential harm to them. This would indirectly alleviate sufferings, knowing that all heroic effort could be futile. However, the best interest of a patient may change depending on the situation – if the underlying problem is potentially reversed and salvaged, ethically, the patient should be treated equally as other patients (1). In the context where the futility of medical care on the condition is confirmed, the principle of distributive justice could be challenged when the patient require ventilatory support as he could potentially occupy the ventilator or intensive care space for a long time, as beds and resources are largely scarce. Providing ventilatory support for patients with SMA type 1 could lead to ventilator dependency, poor quality of life and prolonging the suffering. Medical futility relates to the pointless intervention when available evidence stated that the patient's condition will not improve despite the intervention. Previous understanding that ventilating SMA patients would be a futile effort as there was no effective intervention previously. It is also important to consider a fair and socially just approach to other patients as well, without neglecting this patient's rights.

The availability of novel drugs (Nusinersen, Zolgensma and Risdiplam) has brought new hope for the search of cure for SMA (2). Unfortunately, these therapies do have limitations including high costs, unknown long-term effects and disregarding of survival of motorneuron protein independent targets. The parents would consider this as a sense of renewing hope in their search for a cure for their child. Discarding parental hope and longing for their child's life would be wrong as well.

DECISION-MAKING

The ethics at this juncture can be challenging. With the knowledge that the disease as LLI and the emergence of new therapeutic drugs that would promise hope, it is also essential to be aware that the extension of life may not fully reverse the clinical condition into normality. Discussion should be focused on what a realistic goal entails (3), in our setting. If the patient continues to improve with the use of the novel drug therapy, it would be wrong to abandon the treatment. However, it is also known that these drugs are costly and are available for free for a limited time frame. The financial cost would be impossible to sustain as the drugs are unavailable in the country and not funded in our healthcare setting. Is it right to enrol a patient to therapy and then withdraw it once it is deemed as financially unsustainable? Even ventilating an SMA child for the rest of his life would not be an appropriate choice. The change of clinical status may be seen when death appears inevitable. Predicting what will happen in the future, especially in non-malignant LLI, would be a difficult proposition.

Discussing end of life issues with the family should be done in an empathetic and compassionate manner. Decision-making should be shared between the managing team and parents rather than in a paternalistic way. Any decision should not be influenced by the healthcare professionals' values but should also consider the parental values, by exploring family's understanding on the disease process, their future hope and preferences (3). It is essential to work within the parental expectation, acknowledging their emotions and concerns. The presence of novel treatment for SMA has paved a brighter pathway in treating SMA patients which was not previously available. Treatment for life prolongation has now been made possible. Such treatment raises the hope for the survival of the patient beyond the predicted age. On the other hand, being too optimistic about the promising effect of the treatment without extensively researching and discussing the overall effects could be detrimental. Health professionals and parents must work in tandem with the management and expectation of the disease. There is a limitation in what the novel treatment could achieve, and this decision on continuing or ceasing the treatment may be taken when the clinical progression is static. Transparency of information is important in ensuring the optimal care needed by the child.

Is it beneficial to allow the parents to a have high hope for this drug? Managing parental expectation is needed to allow acceptance of realistic goals. These goals should be sensible and achievable rather than ambiguous such as control of symptoms. Once the family members and doctors reached a consensus a clear plan can be devised to guide the family members on the remedial actions and future planning.

CEILING OF CARE

Discussing on the prognosis and end-of-life issues is a challenging task. The timing of the discussion is an important factor to allow better understanding of such information. This information would be vital to help the family in making appropriate and informed decisions. The prognosis, life expectancy, disease trajectory and end-of-life issues should be allowed to be discussed especially in the advanced LLI (3). These topics are considered as culturally taboo topics for discussion in our setting; and the clinicians need to provide accurate information based on the available evidence. Decisions to withhold, withdraw or limit life-sustaining treatment in children with LLI may still be contentious, difficult and emotive, and determining the best interest of an individual child is even more complex (4). Treatments, including palliative care, should be intended to reduce the child's suffering. The outcome of such a process

requires a high-quality plan to ensure comfort care with an appropriate expertise support.

Healthcare providers must have a clear understanding and recognition of the unique and specific influences, such as culture, religion and customs, have on the behaviours, attitudes, choices and decisions around end-of-life care. Trust is the key behind cultural competency, and this would influence the outcome of the decision-making. A cultural assessment provides a systematic way of gathering and documenting information about the cultural beliefs, meanings, values, patterns and expressions as they relate to the patient's perception and his response to an illness (5).

Medicine is not all about curative treatment. In a certain situation, when risks or complications outweigh the benefits of treatment, especially in acute settings, all-out management is not always the best course of managing a SMA patient. Introducing a newer treatment modality would give some hope to the family, helping in the improvement of the quality of life and may potentially prolonging life. The dilemma arises if the patient is admitted in critical condition, that is to what extent of the management that we should provide. The decision to withhold, withdraw or limit life-sustaining treatment among SMA children has proven to be difficult and emotive. While management of serious conditions would be tailored according to the resource availability, parents would try to persuade decisions towards optimising the treatment for their child despite the uncertainty of the benefits and outcome of the treatment to the child. It is fundamental to consider that the best interest of a child with SMA may differ between health practitioners and parents, at a different time and disease progression. It will remain the most complex decision-making to be made. Decisions to limit treatment should be reached after weighing the benefits and quality of life. This would depend on whether the child has a limited quantity of life (brain death or imminent death), limited quality of life (burden of treatment or illness) and lack of ability to derive benefits (persistent vegetative state) (4).

Definitive decision on necessary intervention is based on the ongoing discussion between the relevant parties to resolve any disparity regarding the management of the patient. Advance care planning should be discussed early to prepare all the involved parties when anticipating unexpected death. This is far from the ideal.

ETHICAL ANALYSIS

Jonsen et al. has proposed a four quadrants approach to ethical case analysis: medical indications, patient's preferences, quality of life and contextual features (6), which is summarised in Table 1. The approach has been labelled as casuistical approach, dissecting the important elements before applying the moral principles. It serves to cover the content of the information and facts about the patient from a bird's eye view of the clinical encounter and health care itself. It must also be translated to clinical reality that is applicable to our scenario and locality. This approach is merely a systematic application to identify and analyse the clinical ethics problem but could not offer clear account of right action to the case (7). In our synthesis, we believe that approach decision on each step for ethical view must be related to the time of assessment, principalism theory, balancing and methods of balancing (7). Ideally, all cases which deemed to have ethical dilemma issue should be broadly discussed within the clinical or hospital ethics to achieve unified agreement from all parties.

Table 1: Four box method assessment on decision-making and ceiling of care based on Jonsen et al. (6)

Medical indications	Patient's (parental) preference
SMA type 1 Started on Risdaplam (novel therapy) Improve neurologically Medication needed for life Aware that medication is not curative but halting the disease progression	Known carrier for SMA gene; previous child also had SMA Wanting all out for the child Aware that when medication programme cease, child's condition may deteriorate Has discussed on advance care plan, no clear plan for resuscitation status Would communicate with non-governmental organisation for cough assist and BiPAP machines Aware that no specialised centre and support if needed to use continuous care at home
Quality of life	**Contextual features**
Immobile and bed chair bound Feeding dependent on gastrostomy Needed full time carer Needed cough assist to remove secretion Still require physiotherapy Needed respiratory support i.e., non-invasive ventilation at home	Poor socioeconomic background Living in rural area Has to pay for care, electric bills (use of non-invasive ventilation at home), purchasing equipment for the care Mother hopes to be the primary caregiver for the patient Aware that the "Compassionate Use Programme" has time end

CONCLUSION

In every decision and changes of LLI status, supportive care remains the crucial part of management and this is tailored according to the requirement of the patient. Details on decision-making whether to go all out or to limit treatment would depend on the continuous communication between healthcare personnel and parents to ensure consensus decisions, despite a difficult and thorny road ahead.

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