The role of palliative care in advanced muscular dystrophy and spinal muscular atrophy

D PARKER,1 I MADDOCKS1 and LM STERN2

1Palliative Care, Flinders University of South Australia, Daw Park, and 2Department of Child Development and Rehabilitation, Women’s and Children’s Hospital, North Adelaide, South Australia, Australia

Objective: This study examines the potential role for palliative care services in the care of individuals with muscular dystrophy and spinal muscular atrophy, and the support of their families.

Methodology: Semistructured interviews were conducted in South Australia with nine bereaved and four current family members of individuals with muscular dystrophy or spinal muscular atrophy. Issues explored during interview included: (i) the family perceptions of the difficulties in caring; (ii) the psychological and physical resources which were available to assist them; and (iii) family recall of the management of the terminal phase of the illness.

Results: Significant issues identified included: (i) a lack of coordination of care and access to skilled, competent carers; (ii) a lack of support for siblings; (iii) inadequate bereavement care; and (iv) limited discussion of options of ventilatory support and advance directives.

Conclusions: The terminal care for individuals with muscular dystrophy and spinal muscular atrophy and their families requires improvement. Although many individuals with these conditions will die following an acute event, palliative care services may be appropriate for those who require a period of terminal care at home.

Key words: bereavement; muscular dystrophy; palliative care; spinal muscular atrophy.

Palliative care refers to the care of terminally ill people. Where possible, palliative care should be delivered at the site which suits patient and family best. Palliative care encompasses physical, psychological, emotional and spiritual support for the individual and family, including bereavement care of the family after the death of the individual. Although such definitions include all causes of terminal illness, palliative care has focused largely on adults dying of cancer. There are, however, issues of equal concern which pertain to children, adolescents and adults dying as a result of chronic progressive disease, issues that are different from those which affect adults with advanced cancer. The major intent of care for adults with advanced cancer is to maintain normality and strive for the best length and quality of life. The family do not want to discount life, thus it is difficult for the family to adjust to changes which indicate that life is closing. The emphasis for the family is on continued life and function, not discomfort. Knowledge of the care required by adults with advanced cancer will not necessarily assist in seeking to assist the process of dying for individuals with chronic progressive disease.

Commonly, palliative care services for children are poorly developed, resulting in children using adult services. Lack of child palliative care services partially reflects the relative infrequency of child death and the different causes of adult and childhood morbidity and mortality. In the United Kingdom, 50% of child deaths occur suddenly, 38% occur in newborns and 22% occur from progressive illness and primarily degenerative disorders. The symptoms of children with degenerative or genetic disorders differ from those of children with cancer. Most children with cancer have little or no speech or mobility problems or difficulty feeding. The role of the family of a child with a life-threatening illness is different from the family role in adult terminal illness. Parents are the principal caregivers and, they may be young themselves. Other children in the family may feel neglected and there may be more than one child affected by a genetic disorder. Use of generic community-based services may lead to problems due to such services’ inexperience in paediatric care, inadequate understanding of symptom control, and a reluctance by families to work with professionals who are not familiar with their child’s illness.

Only recently in Australia has a specific study been undertaken of the palliative care needs of children with life-threatening conditions other than cancer. In this study, the 28 families interviewed highlighted the importance of being told the truth about the disease and the need for specific information and support services. The author reported that for families to accept palliative care services meant letting go of their dream that their child would live. For some families this transition was not possible. Other issues concerning the appropriateness of the use of adult palliative care services for children include the difficulty of an uncertain prognosis, and knowing when the terminal phase begins. In addition, there was uncertainty among families that adult-based palliative care services would understand the care requirements of their child, and a reluctance by service providers who had been involved in the child’s care, possibly for years, to refer to a palliative care service.

In South Australia, few individuals with muscular dystrophy and spinal muscular atrophy are referred to palliative care services. The focus of this study was to examine the potential role for palliative care services in the care of individuals with muscular dystrophy (MD) and spinal muscular atrophy (SMA) and the support of their families.
METHODS

The project was conducted with the assistance of the Muscular Dystrophy Association (MDA) and the Crippled Children's Association (CCA). These organisations were asked to identify families of children and young adults who had:
1. Died of Duchenne muscular dystrophy, Becker muscular dystrophy or spinal muscular atrophy over the previous 7 year period.
2. Families currently caring for individuals with these conditions.

Due to the small number of affected individuals in South Australia, a convenience/opportunistic sample was used to recruit participants.

Ethical approval

The study was approved by the Social and Behavioural Research and Ethics Committee of the Flinders University of South Australia.

Method of approach

The MDA advertised in their newsletter Roundabout that the study was being conducted. Two weeks after the newsletter was circulated, a list of potential participants was made available from the MDA and the CCA to the research team. Contact was made with potential participants by the researcher via a letter of introduction and an information sheet which explained the details of the study. An opportunity was given for potential participants to contact the MDA if they did not wish to be approached by the researcher. If no such notice was received, individuals were approached by phone to participate in an interview.

Interviews

The interviews were semistructured and open ended. Interview questions were developed by members of the research team in collaboration with representatives from the MDA. Questions addressed issues such as: the experience of caring (formal and informal); support that had been available; discussions regarding ventilatory support; recall of the terminal period of care; support during care and in bereavement; the appropriateness of palliative care services; and recommendations for further support.

Data analysis

The interviews were tape-recorded and transcribed verbatim. Transcripts were entered into the NUDIST version 3 software program (Qualitative Solutions and Research, Melbourne, Victoria, Australia) and analysed for recurrent themes.

Participants

Of the 21 potential participants, 13 agreed to an interview. Of the remaining eight participants, five could not be contacted, two declined to participate, and one agreed but, due to the unexpected death of their son with MD, the interview was not conducted. The 13 participants interviewed included nine bereaved families and four current families. Six interviews were conducted with both parents of the individual, four with the mother only, and two with the wives of individuals. For one individual, both parents and grandparents were interviewed.

Of the nine individuals who had died, six died in hospital following an acute respiratory or cardiac event, and three died at home. Age at death ranged from 8 to 31 years. Of the three who died at home, only one individual had used palliative care services. For the four individuals who were still being cared for at the time of the interviews, all lived at home with their parents and were aged 17–21 years. None was receiving palliative care services.

RESULTS

Although each individual’s experience was different, the questions asked during the interview shaped responses around issues that had been identified by the researchers. Lengthy discussions in response to these questions indicated that such issues were relevant to these families. Themes identified from the interview data concern the following: (i) coordination of care; (ii) ventilatory support; (iii) advance directives; (iv) effect of care and death on siblings; (v) parental support; (vi) bereavement care; and (vii) appropriateness of palliative care services.

Coordination of care

Paid carer issues

Paid carers were used by all families and were either from a government-based community service provider or private care agencies receiving home and community care funding. Although the level of paid carer assistance varied for each individual throughout their life, comments regarding the lack of continuity of care and the inadequate level of expertise or education of the carers were raised by most families. At times the wishes of the family and individuals were different from those of the carer agencies. A particular example was the reluctance by care agencies for their carers to become emotionally attached to their clients, leading to rotation of carers. For families, changing carers meant having to explain the routine of what was required and they felt that regular carers who knew the individual provided better care.

Families were concerned that there was no basic skill or training required for carers. Care agencies did not seem to take responsibility in providing information and education about the conditions of the individuals for whom they cared. For the most part the care required was hygiene and the use of untrained carers was appropriate. There were, however, some occasions, particularly during a period of illness or in terminal care, when trained carers were required. The problem of untrained carers was highlighted by one family’s experience of night respite. In this situation the care required included management of a tracheostomy, and the carers had no experience.

MDA arranged for somebody to come two or three nights a week and stay overnight which was good, but the problem there was they weren’t trained nurses or anything. He didn’t want them to suction him, and he didn’t want them to be shown how. I don’t know what it was … it’s trust. I think it’s trust … I needed a nurse (or somebody). I think if it
had been a trained person … because even though these people stayed over, they had to wake me up to suction him.

This example questions the use of limited resources to provide care that was not appropriate and highlights the concern that agencies organising care do not necessarily understand the needs of those requiring it.

**Medical care**

In Adelaide, services for individuals with MD and SMA are provided by the Women's and Children's Hospital and the Crippled Children's Association until the individual reaches the age of 18. Individuals who lived beyond this age had to change from child to adult services. For some, this was a difficult transition, resulting in a delay in establishing a specialist who would undertake ongoing responsibility for care. One parent commented that they had found a very good respiratory physician but that physician was only interested in respiratory problems. They wanted a physician who could provide all the medical care needed. Not all parents experienced this discontinuity and for some the transition from child to adult services had been smooth. One family had organised a network of medical care so the family understood who to contact in an emergency and was confident with the care they would receive.

There were also issues concerning access to adult specialists. The Queen Elizabeth Hospital, located in Adelaide’s western suburbs, has assumed responsibility for the respiratory care of adults with these conditions. Flinders Medical Centre has assumed a similar role in the southern suburbs. For those not geographically located close to these hospitals, routine visits were feasible but in emergency situations the closest hospital was preferred. Unfortunately, if the individual had not been receiving care at that local hospital, medical and nursing staff were unaware of the history and were inexperienced in that individual’s care.

**Parent support**

Families indicated there were opportunities to attend support groups run by either the MDA or other organizations. These were more likely to be used soon after diagnosis for practical information, such as arranging equipment. Some families spoke of a very close network of parent support, and when faced with a problem they preferred to ring another parent who may have been able to help or at least understand what was happening. Families acknowledged that professionals were available for assistance but, because sometimes professionals had been unable to help, confidence had been lost in their ability to do anything. Others mentioned support from a church, or their own families.

Other parents commented that it was easier to be involved in some support groups and activities when the child was younger and did not require as much care. As one mother commented:

I used to think when he was little, why didn’t we have any of the older mums … why don’t they come and tell us what happens? But then you get to the stage when they were most probably at then. I don’t think that I would have wanted to, or could have or would have had the time. I think most of your time is taken up with coping, day to day, and that takes all your energy and all your resources too.

**Ventilatory support issues**

One of the most significant but sensitive issues to be faced by affected individuals is management of inevitable respiratory failure. It is preferable that a decision regarding ventilatory support is not made in an emergency situation, but has been discussed in advance with the individual, family and respiratory physicians.

For the individuals who died, none used full-time positive-pressure ventilation, but one had a tracheostomy and two had used bi-level ventilatory support (bi-pap). For some, the choice regarding further respiratory support had been offered on a number of occasions. This was usually precipitated by an episode of illness due to a respiratory infection. For others, the issue had not been discussed fully, particularly for those who died suddenly.

Families interviewed who were currently caring for their sons did not like to discuss issues concerning ventilatory support with them. The parents indicated they had been made aware, by health professionals, of the available choices in ventilatory support. It appeared to be an individual matter whether discussion about end-of-life care and decisions were encouraged within a family.

The way in which issues such as ventilatory support were approached by professionals was important. One parent commented that when their son had been seen by a respiratory specialist regarding breathing difficulties and options of care had been discussed, the specialist had been very blunt. Later, seeing another specialist who was much more gentle and less confronting, she felt her son was able to understand and make informed decisions regarding his future management.

**Advance directives**

Despite MD and SMA having no cure, only one individual who died at home had specifically written an advance directive. The discomfort of health professionals with advance directives for these individuals was indicated when this family went to casualty with their son for treatment of a respiratory infection. Before their regular doctor arrived, the mother overheard doctors questioning why the family had come to emergency if they didn’t want treatment for the son. Of the two other individuals who died at home, although no advance directive had been completed, discussions concerning resuscitation had occurred. For those who died in hospital, their philosophy had been ‘live for the moment’ and there was a reluctance to discuss end-of-life care. Some parents took the opportunity when one of the other boys died, to discuss end-of-life decisions and consequently felt they understood what measures were acceptable to their son. Four individuals were resuscitated because there was no clear directive or adequate record of discussion regarding emergency care.

For the families who were still caring for their sons, both the individual and the parents wanted whatever treatment was available. Discussions concerning emergency care and treatment decisions were difficult for parents to initiate. Completion of an advanced directive indicates that discussion and agreement on a course of action has been undertaken. Parents’ difficulty in discussing issues of emergency treatment was interpreted as an implicit rejection of the use of advance directives. As one father commented:

We never talk much about the future, especially with him, he never asks for it. Sometimes I probe a little but he doesn’t really want to, so it is an unwritten law: we never
Sibling issues

Parents commented that living with a sibling who has a chronic but terminal disease is difficult both when the individual is alive, and also after death. Some siblings had become quite involved with the friends of their affected brother, and when a friend died it distressed the sibling as well as the affected brother. Parents commented on reports from school of daydreaming or poor performance by siblings, and felt that this was related to how they were dealing with what was happening in the family. Some siblings had good support from school and had accessed school counsellors, both before and after the individual’s death.

Grief reactions of siblings differed, depending on the relationship to the individual before he died, the age of the sibling, and how they were able to express their feelings. Some grieved openly and were able to talk and cry, but others did not appear to grieve until triggered by some other event. One parent felt quite strongly about setting up a support group specifically for the siblings of children with progressive muscular diseases, so these children could talk to someone who could understand the experience.

Bereavement care

Not all families had accessed bereavement care, and there was mixed satisfaction from those who had. Establishment of a good rapport with the bereavement worker prior to bereavement was important. The best bereavement support was felt to be from someone who knew the deceased.

The worker, whoever it would be, knows a bit about that child, not just appearing on the scene when the child has died, to try and pick up the pieces. It’s got to be an ongoing thing that they’re familiar with that family situation and then they’re there. Because you’ve got the confidence in that person to open up, you’re not going to open up to a stranger.

Families grieved in different ways. Some had a period of time when neither partner spoke about the death of their child because they were frightened of upsetting each other or other family members. Because mothers played differing roles in the care of their children, so they expressed grief differently. Women appeared to be able to access help more openly. Men spoke of support from work, but this seemed to be for a limited time and was dependent on the existing relationships within that workplace. A good support network among parents was considered particularly important, as was ongoing family support. Many parents commented that coping without the help of extended family members would have been difficult.

Involvement of palliative care services

Only two individuals had been referred to a palliative care service, with one using extensive services. For some families, the deterioration of the affected person was so sudden that they did not think a palliative care service would have had much to offer. One mother, whose son was admitted to hospital in acute respiratory failure, was given the option of taking her son home with palliative care support. As he only lived for 2 days after hospital admission, she did not feel that period of time was long enough to explore the option of palliative care.

Families who knew about palliative care indicated a reluctance to use hospice for respite. For one family, it was because of the age and level of physical illness of the other patients in the hospice. For the other family, where the individual concerned was older, the age of the other patients was not the problem, but rather the image of what a hospice represented. For this individual his wife commented: ‘When you’re in a hospice, you know what a hospice is don’t you? They’re reminding you that you’re on your way out, and you don’t need that’.

One family thought that palliative care services could be involved in talking with the family about what to expect as things deteriorated. One family commented that it is difficult for those who care for the individual on a daily basis to see the deterioration. Reflecting back after their son had died, they realized the signs of their son’s deterioration had been there. It was difficult for some carers to consider that palliative care would have been useful. Although MD and SMA are progressive conditions, death came very unexpectedly for some.

DISCUSSION

Coordination of care

Perhaps the most significant problem identified in this study was the lack of coordination of care and access to skilled competent carers. For individuals with MD or SMA, care needs can span the life cycle, from early childhood through adolescence and into adult life. The responsibility of coordination of care by one agency throughout this period is undoubtedly difficult and may not be feasible. However, as indicated by this study, many families indicated better coordination between services and more consistent and reliable information would enable families to make effective use of all the care that was available. The establishment of a ‘key worker’ as advocated by Woolley et al. may assist families in awareness of services available and improve ability to overcome administrative hurdles in the negotiation of care. Hilton et al. argued that fragmentation of care has resulted from the development of technology and medical specialization. Physicians responsible for long-term care in the community may be different from those called upon to provide care in acute settings. Care for individuals with chronic progressive disease also needs to incorporate a plan of long-term growth and development of the child.

The majority of care for individuals with MD or SMA is predominantly from untrained carers working on a casual basis who are able to provide many of the day-to-day caring tasks. In circumstances of acute illness or terminal care, more experience may be required. For those families who provide terminal care at home, the support and assistance of a palliative care service may be helpful in facilitating this care.

Ventilatory support

Specialised respiratory management clinics for individuals with MD exist in other countries but so far not in Australia. In South Australia, either a paediatric physician or adult respiratory specialist provides discussions on the management of respiratory
failure. Families reported different experiences regarding discussions of respiratory failure and the treatment options available. For some families, discussion of respiratory support or advance directives of care only took place when respiratory distress occurred. Open communication between the individual and other family members regarding these issues was rare. However, families indicated that when necessary they would make a decision based on what the individual had indicated at some time.

Advance directives

In South Australia the Consent to Medical Treatment and Palliative Care Act was introduced in 1995. This provides an opportunity for individuals over the age of 16 years to either appoint a medical agent to make medical decisions on their behalf, or to write an anticipatory directive. Anticipatory directives enable an individual to set out their wishes regarding resuscitation, ventilation, and maintenance of life support systems. Only one individual in this study had completed an anticipatory directive.

As medical technology continues to prolong life, more individuals with chronic progressive disease will survive beyond the age of 16, when they legally can decide their own care. Appointment of a medical agent, or the completion of an anticipatory directive should be encouraged, as it is an ideal opportunity to confirm the wishes of an individual in the event of cardiac and respiratory failure. Approached openly and sensitively, this should not be distressing for the individual or family members and can confirm that, when necessary, the appropriate care will be ensured. From the present study, it is clear that the existence of advance directives may have spared some families the trauma of experiencing a futile resuscitation attempt of their son at home or in hospital.

Sibling issues

The issue of sibling support is something families have had to deal with regularly during terminal care and bereavement. Sibling issues however, have long been overshadowed by a focus on spouse or parent support and bereavement care. There appears to be a need for a sibling support group both during a chronic illness and in bereavement. This was felt by families to be important, as it would facilitate an avenue for siblings to be able to speak in a supported environment about issues that they may not be able to discuss with their parents. In contrast to individuals with cancer, those with MD and SMA require care for many years, and the family may become centred around that individual.

Adjustment of siblings in bereavement may also be influenced by the place of death of the individual. Lauer et al. found that the siblings of children who died from cancer at home felt better by the place of death of the individual. Lauer et al. found that families of children with cancer, who had cared for their child at home, adapted better in bereavement. The authors cited the opportunity for anticipatory mourning, which enabled families to adjust to the death of their child. These authors did not elaborate about whether access to bereavement care was more likely for families caring at home than those in hospital. In Adelaide, no bereavement follow-up is provided for the family by a hospital following death.

Place of death

For three individuals, death occurred at home, and for six, death occurred in a hospital setting, in some cases in the emergency department. Families who experienced home death, seemed to have come to terms with the individual’s impending death. Acceptance of death seemed less likely for those admitted to hospital with acute respiratory or cardiac failure. In some circumstances, this reflected the uncertain nature of the disease where admission to hospital is something families had to deal with regularly, and where there was always hope of effective treatment. However, whether active treatment in hospital is accepted or refused, it is questionable whether a hospital setting is the most appropriate place of death. Families indicated that hospital staff were inexperienced in care and lacked the time required to provide the care that the individual and family felt they needed. In some circumstances, staff were not familiar with either the individual or the family, and no support or follow-up was available.

For individuals with chronic progressive disease, there have been no comparative studies undertaken of family coping in relation to place of death. Lauer et al. found that families of children with cancer, who had cared for their child at home, adapted better in bereavement. The authors cited the opportunity for anticipatory mourning, which enabled families to adjust to the death of their child. These authors did not elaborate about whether access to bereavement care was more likely for families caring at home than those in hospital. In Adelaide, no bereavement follow-up is provided for the family by a hospital following death.

Bereavement

No formal coordinated bereavement services were evident and it was difficult for some families to identify their own needs for support. Families who have cared for someone with a chronic progressive disease with specific needs want a counsellor who understands their particular situation. Sharing grief with others is an important factor in the bereavement process, but not all professionals may be skilled in bereavement care.

For families not involved with a palliative care service, formal bereavement support, coordinated by a professional, is unlikely in any setting. In these cases the organization of appropriate care should be the responsibility of a designated service or organization. Bereavement support for these families should begin before the death of the individual, to enable the professional to establish a rapport with the family. The timing of this contact will be unique to each situation, depending on the assessment of an experienced professional.

Palliative care services

Hospital and hospice services did not understand or manage the needs for care well. Families indicated a need for increased nursing care to be provided as well as better understanding of MD and SMA among hospital staff. There are many principles of care of individuals with chronic progressive disease which could be improved in institutional settings.

The reluctance of families to use hospice inpatient facilities for respite or terminal care is difficult to overcome. Hospice is well regarded in the community as a place of providing terminal care, but most hospice patients have cancer and are over the age of 60. For adolescents or adults who have lived with a progressive terminal condition such as MD or SMA,
the acceptance of palliative care and hospice care is more difficult.

Many individuals with MD and SMA die quickly, and the use of hospice or home palliative care services may not be appropriate. For those whose decline is slower, and for whom the need for nursing care increases toward the end of life, referral to a palliative care service may be warranted. If so, increased awareness and education of these diseases will be required by palliative care services. This has been possible for other conditions such as motor neurone disease.13

CONCLUSION

Families and individuals with MD and SMA know they have a life-limiting disease. As the condition deteriorates and the day-to-day caring tasks increase, the acceptance of palliative care may be in conflict with the hopes of individuals and families. Palliative care services are also viewed as being for adults with cancer. Families with little experience of cancer may not be aware of palliative care services or, if they are, may not be confident that these health professionals can provide appropriate care for someone with MD or SMA. Palliative care services may be appropriate for some individuals with MD or SMA, but these services should complement a coordinated, existing service, and not seek to replace them.

The Muscular Dystrophy Association of South Australia provides assistance with equipment, disease-specific information, and social, sporting and fund-raising activities. As this association is largely funded by donation and is not a state responsibility, only limited support is possible. Such organizations, however, having the expertise and early contact with individuals and their families, are the obvious link to coordinate services.

This study recommends: (i) an increased awareness by muscular dystrophy associations of the care provided by palliative care services; (ii) the trial of a ‘key worker’ to improve support and care for individuals and families; (iii) a trial of support groups for siblings; (iv) increased training, information and competency standards for care workers; and (v) increased awareness in acute care settings and hospices of the needs of individuals with MD and SMA.

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