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EDITORIAL

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Spinal muscular atrophy with respiratory disease (SMARD): an ethical dilemma

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The classic riposte of the countryman when asked by a stranger for directions was 'Well I wouldn't start from here, if I were you'! He would probably have said much the same after reading the contribution by Giannini et al. in *Intensive Care Medicine* reporting two babies with spinal muscular atrophy with respiratory disease (SMARD) who were diagnosed only when they were irrevocably ventilator dependent [1]. There can be no more difficult question to address in a paediatric intensive care unit than what is in the best interests of a baby with completely normal central neurological function but virtual complete and untreatable peripheral and respiratory muscle paralysis who is totally ventilator dependent. What lessons can be learned from this report? The ideal is of course prevention, and this starts with general paediatricians and paediatric neurologists. Faced with the as yet unventilated, weak and floppy baby, it is first both crucial and urgent to reach a correct diagnosis. It has only recently been appreciated that SMARD is a genetically separate entity [2], and hence much of the previous literature may unwittingly have classified SMARD babies as having spinal muscular atrophy type 1 (SMA-1). Indeed, since the two are completely distinct conditions, the nomenclature is extremely confusing. Furthermore, it is likely that there are other conditions which present with an almost indistinguishable clinical picture, but in which the underlying genetic abnormalities have not been discovered [2]. Nevertheless, the literature gives us useful information about likely outcomes, which should inform discussions with the parents. We have previously tried to address some of these issues, in the context both of unventilated and of already ventilated SMA-1 and SMARD babies [3]. Key is a culture of openness: whether physicians like it or not, parents will obtain information from the internet and other readily accessible sources. It is therefore much better to discuss possible therapeutic interventions such as non-invasive ventilation, cough in-exsufflation, and tracheostomy in an open manner, setting out the advantages and disadvantages of each, while the child is still well. This should ideally lead to a plan for the care of the child being formulated and written in advance, including whether it is in the child's best interests to have short or prolonged periods of intubation, or to proceed to a tracheostomy. This may prevent inappropriate intubation during crisis management, if the parents first, supported by the multidisciplinary team, feel that this is not in the best interests of the child.

In the case of the children reported by Giannini et al. [1] it appears that these discussions did not take place until the children were already intubated and ventilated. At this point such children are usually alert and well, and the question of withdrawing treatment in an alert, otherwise well child as opposed to not instituting it in a sick, deteriorating one is much more difficult. It may be appropriate in some to extubate them to nasal mask

ventilation with a decision not to re-intubate, but in some babies this may not be possible or be rejected by the parents, who by this time may be demanding that their child undergo tracheostomy. Lack of a prospectively agreed plan frequently leads to a breakdown of trust between heath care team and parents and to the well intentioned but usually unhelpful involvement of politicians or the media or to attempts to fund-raise to send a child abroad for a 'miracle cure'.

In general, many European paediatricians and Intensivists, including this author, are dubious about the rightness of tracheostomy in children with SMARD and other really severe neuromuscular diseases. This is based in part on important papers on the long-term consequences of tracheostomy [4, 5]. Tracheostomised SMA children may have multiple admissions to hospital, and progress to loss of all muscle power, including in the facial muscles, becoming unable to communicate and thus being in a 'locked-in' state. Death comes early to these unfortunate children, irrespective of tracheostomy. Whereas non-invasive respiratory support, with nasal mask ventilation at night, and augmented cough [6] offers a quality of life which largely does not involve hospital admissions after the first 3–5 years of life [4], the quality of life with a tracheostomy has been seriously questioned. Despite this some parents opt for tracheostomy.

It should be noted that in a recent High Court case in the United Kingdom the parents of a severely affected SMA-1 baby won the right for a tracheostomy to be performed, even despite the contrary advice from all the professionals involved in the care of the child. The Court decided that the child had a sufficient quality of life, and that this life should be prolonged at the behest of the parents. This ruling is not of course binding outside this country, but it may well be replicated in other countries in the future.

It is not the purpose of this annotation to question the rightness or otherwise of the decision of the Court in the particular case addressed. However, perhaps we need to ask whether the conventional reluctance (shared by myself) to proceed to tracheostomy is always correct. It should be noted that it is difficult and dangerous for an able person to second guess the quality of life of a disabled person. At least two papers have documented that teenagers and adults with Duchenne muscular dystrophy, wheel chair bound and nasal ventilator dependent, assess their own quality of life as very much higher than would be anticipated by health professionals [7, 8]. Perhaps we need to ask more often 'who has the problem?' Is it the child, or is it the health care professional looking after the child? This is an unanswerable question in the case of small babies, but it is at least worth considering, and setting in the context of the literature about those who can communicate. In intensive care the focus is usually and rightly on rapid decisions in a fast-changing situation, with recovery or death the outcomes, within a short timeframe. Perhaps professionals need to be ready to re-focus these priorities, which may not necessarily be appropriate in the context of a chronic neuromuscular disorder.

If parents are determined that tracheostomy is the best way for their child, what then? Firstly, they need to be given a realistic picture of life at home with a tracheostomy-dependent child: the equipment which will be moved in, the loss of privacy for the long hours when carers are responsible for the child, the lifestyle implications for the parents and other siblings for themselves in terms of the time investment looking after the ventilated child, and the likely 6 month wait in hospital for the care package to be put in place. These are only some of the important issues to be considered [9, 10]. If at all possible, there needs to be agreement with the family about the limits of intensive care (should the child have arterial or venous lines placed if there is deterioration, what about inotropes and resuscitation drugs) and also an exit strategy—if the child deteriorates to a 'locked in' state, how do we recognise it and manage the child humanely? Finally, the likely outcome of death at a young age, even despite a tracheostomy, must be discussed and acknowledged by parents.

This area is clearly a legal and ethical minefield. Even if there is agreement between parents and professionals, in my view it is wise to seek legal opinion as to the lawfulness of any proposed treatment withdrawal from the intubated SMA-1 or SMARD baby. This is even more important if agreement cannot be reached by those concerned. The medical team, at least in the United Kingdom, are protected far better by the Courts than by the General Medical Council (the statutory body responsible for the registration and determination of fitness to practice of physicians). The recent record of the latter body in 'supporting' physicians is disgraceful. Best, if there is doubt, is for the family and professionals to seek guidance from the Courts as joint applicants, rather than as adversaries, if this can be achieved.

It is likely that the dilemmas posed by the two babies reported by Giannini et al. will become more, not less frequent. There is clearly no one right way of proceeding, and it might be wise for those units which have not established protocols to deal with such scenarios to do so promptly. Protocols should be underpinned by the principles of dignity and respect for the family and baby, complete openness with all information, a readiness to listen and not to hurry decisions, and an acknowledgement that choice implies the right of parents to decide differently than professionals.

Ultimately, who generally is best at determining the quality of life of a baby in situations of great uncertainty? Professors? Ethicists? Or the parents of the child?

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