

Treatment and Care for Children with Very Severe Mental and Physical Disability

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Abstract: We present two cases of severely disabled children who required tracheotomy and ventilatory management under long-term care at home. Both cases were infants who had survived life-threatening crises during the perinatal period and infancy. Even if their conditions have stabilized, they are always at risk of sudden changes. There are several preconditions for the long-term survival of such children, in which family plays a considerable part. Medical personnel should provide the best supportive care in both acute and chronic phases.

Keywords: Special healthcare needs; Home-based care; Best supportive care

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1. Introduction

Severe mental and physical disability is a combination of severe physical disability and severe intellectual disability. Although the term has long been used in pediatric medical and welfare professions, it is not strictly a medical diagnosis but rather a definition given in the Child Welfare Law (Article 7). Children with severe mental and physical disability have underlying central nervous system (CNS) disorders that developed *in utero* and progressed through infancy, and the disorders vary, ranging from congenital abnormalities of the brain to those caused by head trauma, hypoxic encephalopathy, intracranial hemorrhage, CNS infection, neurometabolic disorders, and degenerative diseases. Depending on the underlying disease, it is not uncommon to encounter a life-threatening crisis at the time of injury or acute deterioration, with each time, the family being aware of the risk of mortality. Furthermore, recently, a group of children with severe mental and physical disability^[1] with particularly high needs for intensive medical care has been classified, mainly on the basis of the need for distinction in terms of medical reimbursement. The cases reported in this article are children with very severe physical and mental disability who constantly need various medical equipment for support, including ventilators, due to difficulties in maintaining life-related functions. Some of them lack the ability to express their intentions clearly or even to move their bodies, and most of them are raised at home by their families. Based on a presentation given at the workshop "Society Living with Disabled Children" at the 33rd Annual Meeting and Congress of the Society for Brain Death and Cerebral Resuscitation, actual cases of children with very severe mental and physical disability are discussed in this paper.

2. Cases

2.1. Case 1

No specific perinatal or medical history. At 6 months of age, the patient was found suffocated at home by

a family member, who subsequently performed chest compressions and made an emergency call. It took approximately 45 minutes for return of spontaneous circulation, and she was admitted to the pediatric intensive care unit (PICU) of Saitama Children's Medical Centre (hereafter referred to as our center) for general and hyperthermia management. On admission, there were no apparent seizures, and continuous electroencephalographic (EEG) monitoring showed no rhythmic or cyclic patterns suggestive of seizures. Even after the patient's general condition had stabilized and vasopressors were no longer required, the Glasgow Coma Scale was E1VTM1, light reflex was only faintly present on one side, and there was minimal spontaneous breathing. Severe residual CNS impairment, including brainstem function, remained, and medium- to long-term airway and respiratory support was required to sustain life. After several conversations with the medical team and the family, a decision was made for tracheostomy and ventilatory management. After the patient was discharged from the PICU, the trunk and the extremities were relaxed, and there were no obvious body movements other than slight movement of the chest (spontaneous breathing). During the recovery period, the EEG showed irregular scattered slow waves, with an amplitude of about 20 μ V and little activity. The auditory brainstem response (ABR) was not unresponsive bilaterally, but V waves, one of the central components, were not observed (**Figure 1**). The patient's nutritional and fluid intake were dependent on nasogastric tube feeding, and catheterization and enemas were required for defecation.

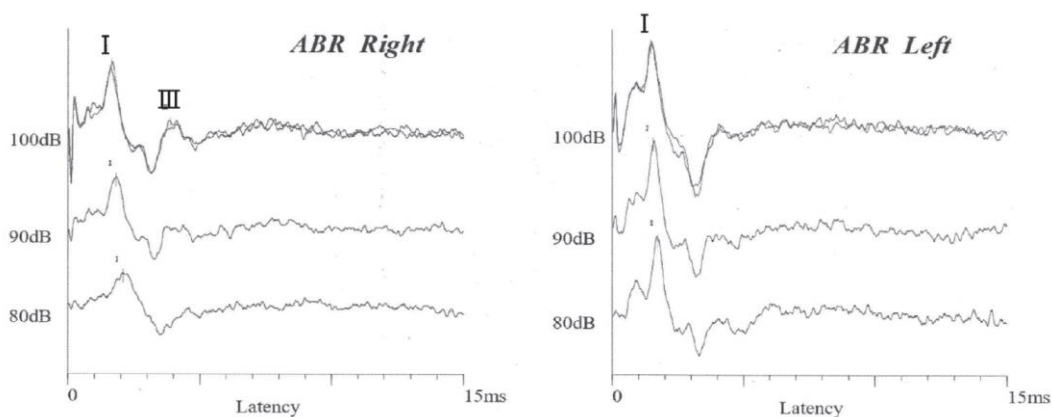


Figure 1. Auditory brainstem response 1 month after the onset showing no V waves when stimulated at 100dB (left and right)

2.2. Case 2

A full-term birth with no perinatal or birth abnormalities. At 2 months of age, the posterior arch recumbent position became increasingly prominent. The patient had decreased sucking reflex, and at 6 months, the patient developed recurrent airway infection and breathing problems, probably due to aspiration. Along with the retention of airway secretions, respiratory depression occurred, which then worsened. At 8 months, the patient was diagnosed with Gaucher disease type 2 (acute neurological type) after a close examination that revealed marked hepatosplenomegaly. Enzyme replacement therapy (intravenous veraglycerase alfa every two weeks) was initiated to treat the underlying congenital hypoactivity of glucocerebrosidase. Dysphagia and hypoventilation were suggested to be irreversible symptoms of the primary disease, upon which the decision for laryngotracheotomy and home ventilation was made at 1 year 2 months. The patient had marked hypertonia predominantly over the trunk muscles on awakening, strong and frequent myoclonus (>100 per minute), and a pulse rate of >180 beats per minute for a period of time. Each symptom was controlled by increasing the dose or number of oral medications, resulting in oversedation (Richmond Agitation-Sedation Scale [RASS] score -4). Since then, the patient has been able to live at home for more than five years without prolonged hospitalization or life-threatening crises.

3. Considerations

3.1. Severe mental and physical disability is different from brain death

Both patients, as described above, were dependent on ventilator and intensive care to sustain their lives. Given its similarity with brain death, it was once confused with prolonged disorders of consciousness and chronic brain death ^[2]. In an editorial published in the Journal of the Japanese Society of Paediatric Neurology, in response to the implementation of the revised Organ Transplant Law in 2010, Sasaki ^[3] stated that the medical care for severely retarded children is not curative, but rather supportive. The concept of brain death should not be confused with severely disabled children. Emphasis is placed on support rather than cure, and that the child is alive, not dead.

In Case 1, the patient's general condition was maintained without any active therapeutic intervention other than securing her airway and breathing (tracheostomy and ventilatory management). The patient's family decided to look after the patient at home, since the patient's condition had been stable for several months. In addition, no premorbid internal organ damage was observed. Although physical growth and changes are expected in the future, there is little evidence that prognostic improvement in the patient's CNS function can be expected. When preparing and guiding the transition from PICU to the general ward, and eventually back home, the medical staff were reminded to treat each parent and child as a person rather than a condition.

Cardiac arrest is rarely the cause of cardiopulmonary arrest in children and infants. It is often followed by respiratory arrest. Although the outcome is poor in children or infants who had suffered from cardiopulmonary arrest, the survival rate has been reported to be more than 70% if they are found only with respiratory arrest and treatment is initiated before cardiac arrest ^[4]. Even in life-saving cases, lifelong dependence on medical devices and home medical care is expected in cases of severe CNS disorders such as the present case. Rehabilitation is no longer primarily aimed at restoring function, but at maintaining range of motion of joints and optimal positioning, becoming more care based as the patient's physique changes.

3.2. There are several preconditions for long-term survival of children with very severe mental and physical disability

In Case 2, the patient was born with inborn errors of metabolism, resulting in the rapid deterioration of functions of multiple organs, including the brain, during the first year of life. In Japan, the replacement of defective enzymes in Gaucher disease is covered by insurance and can be performed regardless of the clinical form, but in Europe and the U.S.A., where insurance does not cover the neurological form, including type 2, the prognosis is not promising. As stated in a well-known pediatric textbook, children die from respiratory symptoms in the first few years of life ^[5]. The majority of respiratory symptoms referred to in the cases are due to aspiration. After explaining the situation to the family and respecting their willingness to live together with the child at home, we proceeded with tracheostomy, aspiration prevention surgery, and ventilator management.

It must be stressed that there are several preconditions for a child with Gaucher disease type 2 with a poor prognosis to be able to stay at home for more than five years. Each of the preconditions listed in **Table 1** is necessary to be repeated many times on a daily basis, and the many types, doses, and frequency of oral medications would place an additional burden on the family. It is gratifying that advances in medical technology and knowledge have greatly reduced the hurdles to long-term survival and home transfers for the patients in the aforementioned two cases. However, their families who have to accept them as a matter of course would be under enormous pressure. Healthcare providers should respect the family's desire to look after their child at home and provide long-term support.

Table 1. Preconditions for a child with Gaucher disease type 2 to be under long-term home care

(1)	Continuation of enzyme replacement therapy (every 2 weeks visit)
(2)	Tracheotomy management (10 or more tracheal suctionings per day)
(3)	Continuous mechanical ventilation
(4)	Liquid nourishment through gastrostomy
(5)	Catheterization, enema, and disimpaction
(6)	Management of hypertonia and myoclonus

For muscle hypertonia: oral tizanidine (high and frequent dosing), *etc.*
For sympathetic hypertonia: risperidone, phenobarbital (high dose), *etc.*
For myoclonus: clonazepam, perampanel, *etc.*

3.3. Awareness that the possibility of a sudden change is always around the corner

The children described in this paper are not in a state of chronic brain death^[6], but they are totally dependent on ventilators for life support. When a child in such a state stays at home with his/her family, the medical personnel will need to be involved in a way that is different from the standpoint of the end of life or death. There is always a possibility that the condition could change at any moment, considering that there are multiple risks of sudden change, including airway problems.

In regular outpatient care, primary care physicians should check for changes in these risks and support the child and family to maintain quality of life in “moderation.” In the event of hospitalization following a change in condition, care should be taken to share an understanding of the child’s living environment, daily care, and developmental progress along with the child’s condition in a situation where relationships with staff members are inadequate. This is to ensure that best supportive care^[7], which is the axis of daily care for many children with very severe mental and physical disability, is maintained even when the child is placed in an unusual environment (*e.g.*, PICU) in the event of a sudden change.

4. Conclusion

Although children with very severe mental and physical disability are dependent on medical equipment and others for survival, their abode is their home, and their existence is with their families. The involvement of family is as close as possible to nursing care, but medical practitioners must bear in mind that the parent-child relationship is the foundation of treatment and care.

Disclosure statement

The author declares no conflict of interest.

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