Physical Therapy for Child with Epidermolysis Bullosa
A Case Report

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Abstract The purpose of this paper is to report on the course and physical therapy for epidermolysis bullosa. Epidermolysis bullosa is designated as a specific rare disease by the Ministry of Health and Welfare. The clinical feature is repeated blister formation that occurs following mechanical stimulation in daily living. The blisters may occur without cause especially at an active time of morbid change, although these are actually caused by slight stimulations.

It is absolutely indispensable for sensori-motor learning in the baby that the body surface make contact with the environment. Motor learning is based on having experiences with the interaction between perception and motor. However, it is guessed that no comfort was experienced by the patient via sensory information from the skin. It is probable that he experienced of continuous pain and pruritus, felt as itching of the entire body. Therefore, it is inferred that biased perceptions would be generated and learned. In addition, blisters, erosions, and markedly fragile skin were actually aggravated by sensory inputs made to facilitate developmental intervention. Therapeutic exercise for the patient has been going on since four months from birth, but the patient has been continuing to exhibit retarded motor development. However, the patient was able to enter school normally in spite of having a few problems at present.

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Key words : Epidermolysis bullosa, Blister, Sensori-motor, Motor development, Physical therapy

Introduction

Epidemiological research (Kitamura 1986)[1] indicates that there are 393 epidermolysis bullosa (EB) patients in Japan (estimated prevalence rate: 0.609 per hundred thousand population). There are three major types of EB, classified on the basis of where blisters form ultrastructurally within skin. These three forms have been referred to as epidermolytic, junctional, and dermolytic (Table 1)[2].

Blisters are defined as a condition in which transparent liquid collects and bubbles within the skin, more accurately described as intraepidermal or subepidermal lamina. The clinical feature of EB is repeated blister formation that occurs following mechanical stimulation in daily living. Blisters may occur without cause, especially at an active time of morbid change, although these are actually caused by slight stimulations.

It is absolutely indispensable for sensori-motor development of the baby that the body surface make contact with the environment. In cases where blisters are present from birth, it can be inferred that problems will be caused during motor development. A research group sponsored by the Ministry of Health and Welfare for work on rare disease was founded in 1983. Advances have been made in area of EB, and a number of studies have been made concerning EB, but little has been found in the literature concerning motor development in patients with EB. In a previously reported case, the patient was slow to begin walking due to the morbid state of the feet, finally beginning to walk at the age of four[3]. In a second case, physical and mental retardation were not observed[4]. In a third case, functional disturbance of the hands was observed and was caused by scars or contractures[5].

The current authors have had experience with a case in which the patient was able to enter school normally in spite of having a few problems at

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Table 1. Classification of Clinical Types of Epidermolysis Bullosa[2)]

Intraepidermal
EB simplex, generalized(Koebner)
EB simplex, localized(Weber-Cockayne)
EB herpetiformis(Dowling-Meara)
EB simplex(Ogna)
EB simplex with mottled pigmentation
Junctional(intralamina lucida)
EB atrophicans generalisata gravis(Herlitz;EB letalis)
EB atrophicans generalisata mitis
EB atrophicans localisata
EB atrophicans inversa
EB progressiva
Generalized atrophic benign EB(GABEB)
Cicatricial junctional EB
Dermolytic or dystrophic (sublamina densa)
Dominant forms
Dystrophic EB, hyperplastic variant(Cockayne-Touraine)
Dystrophic EB, albopapuloid variant(Pasini)
Recessive forms
Generalized(gravis or mitis)
Localized
Inverse

present, and it is the purpose of this paper to report on the course and physical therapy.

Presentation of the Case

The case, a male, is presented in October 1994 at the age of seven years and five months. The birth weight was 3750g, and gestational age was 39 weeks. The mother had a spontaneous delivery, and the Apgar Index was nine points. The clinical symptoms that occurred at birth were epidermidosis surroundings the navel and brownish fingernails. Immediately he was admitted for further examination, and a diagnosis of EB was made. (Fig.1)

Blisters arose over the entire body several days after birth, he cried violently, and stridor occurred on the tenth day. Intake of milk decreased after three months due to blister formation on the tongue. With the consequence that body weight also decreased. The baby had no abnormality in the finding of computed tomography and electroencephalogram.

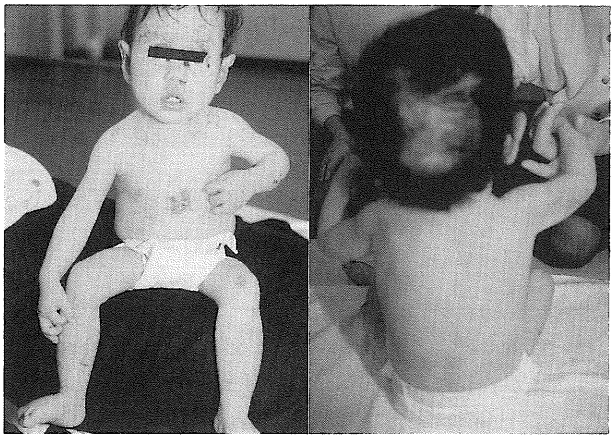

Fig. 1. Blisters, erosions, and, fragile skin were seen in case. (one year and two months)

The majority of the body surface was covered with bandages and gloves, etc. for about six months running. These manipulations are common among EB patients in order to protect blisters and prevent infection. Blister formation has continued through the present, although there have been alternating periods of worsening and remission. The baby was hospitalized for about six months during the first year after birth because of cyanosis and acute bronchitis, etc. He was readmitted as many as seven times for repeated bouts of pneumonia, etc. At two years of age, this patient was diagnosed as having junctional EB from his clinical history, the symptoms, and the findings of electron microscopy.

Course of Physical Therapy

The physical therapist began assessment and therapeutic intervention at four months after the patient's birth. He was initially found to like the satisfying posture of lying on his right side or being held in his mother's arms, and he cried violently in other positions. He could not maintain his head uprightly, and he typically held his head thrown backwards in the midline. He could not hold himself on his elbows, even when placed in that position.

Primitive reflexes were absent in the patient, and he did not have sufficient righting function abilities. The regulation of degree and distribution of muscle tone was nearly normal. It was observed that blisters had arisen over the entire body. Blisters were especially noticable on the surface of the body that made regular contact with the floor.

The physical therapist helped the patient to learn

the antigravitational control of the head at the midline, so as to acquire motor development and righting function. The patient cried frequently during therapeutic handling. Along with the physical therapy program, it was recommended to teach the mother the means to do positioning and handling.

At the age of seven months,the patient could look around with the head raised, supporting himself on his elbows in the prone position. In order to facilitate motor development and righting function, the therapist repeatedly, slowly made him turn over. In additional programs from the age of eight months,he was treated to facilitate the support of lower limbs and parachute reaction with the help of a therapeutic balloon. In this stage, eye and hand movements were frequently observed when lying on his back. At the age of ten months, he kept his feet on the floor, but he did not bounce up and down.

Afterwards there was an interruption of about four months on account of his pneumonia, and physical therapy was restarted. In this stage, the patient could sit alone with slight kyphosis and began to explore his surroundings, but he could not sit up. The physical therapist added programs to facilitate the antigravitational function by playing in both the sitting and standing positions. At the age of one year and four months, he spoke his first words and he could drink liquid with straw. He could sit without being supported in both long sitting and chair sitting positions. The physical therapist went on with the previous programs in order to stimulate the patient to more verticalization.

At the age of one year and six months, the patient could eat normal food and could drink from a cup unaided. He employed shuffling as a means of locomotion. He began to pull himself up by the bars of his play-pen at two years of age, and spontaneous play in the standing position developed. He was still strongly dependent on his mother in this stage. The physical therapist again added programs to strengthen the antigravitational muscles and to facilitate the equilibrium reactions by playing while standing. At the age of two years and four months, he could walk with both hands held; three months later he began to walk without support and to speak two-word sentences. He could walk steadily, falling down only occasionally, but endurance and speed of walking remained low. Along with the physical therapy program, it was recommended to teach mother the means to facilitate the function of vestibular and equilibrium organs, by playing on the slide and seesaw in the park.

The patient entered an ordinary kindergarten in order to improve his independence at three years and five months of age. He had bilateral tightened hamstrings from the age of four, and scoliosis with convex on the left side was diagnosed by a regular medical examination at five years of age. At five years and eleven months, he began to attend swimming school regularly in order to improve his physical strength. He was accepted for entrance to a normal elementary school in April 1994.

Discussion

Many different therapies have been used effectively to promote wound healing in patients with various forms of EB. The research group of the Ministry of Health and Welfare made therapeutic guidelines for EB[6], but there are no certain and specific therapies for EB at present. Therefore, the goals of local skin care and treatment are directed toward reduction of inciting or exacerbating factors and the promotion of healing of previously developed blisters and erosions (Fine 1986)[2].

We have found few references in the literature to motor development in patients with EB. In the present case, the onset of blisters occured at birth and became increasingly severe. In consequence, the patient has been continuing to exhibit retarded motor development. It is absolutely indispensable experience for sensori-motor learning in the baby that the body surface make contact with the environment. Motor learning is based on having experiences with the interaction between perception and motor. It is held that the baby forms body image, recognizes surroundings, and develops space perception rather quickly through motor experiences.

However, it is guessed that no comfort was experienced by the patient via sensory information from the skin. It is probable that he experienced continuous pain and pruritus, felt as itching of the entire body. Therefore, it is inferred that biased perceptions would be generated and learned. In addition, blisters, erosions, and markedly fragile skin were actually aggravated by sensory inputs

made to facilitate developmental intervention. In consequence, it is thought that the patient had increased discomfort and that a vicious cycle was generated.

Shirai et al.[7] made an experiment on giving frictional stimulation to siblings'feet and obtained findings from electron microscopy. They surmised that on the ultrastructural level, cell membrane defects were produced by mechanical stimulations. Yoshikawa et al.[8] reported that the force of gravity to the fibroblast led to blister formation.

However, it is necessary for sensori-motor learning to use tactile sense and pressure sense, and it is necessary for babies to experience safety, pleasantness, and certainty in sensori-motor learning. Therefore, they are dependent on perceptional information, and this perceptional information is indispensable for all motor planning. Adams[9] reported that rapid and slight tactile stimulations to children with central nervous system disturbance improved motor development. Casler[10] reported that the score in Gesell's developmental index was improved by the stimulation of slight tactile sense and pressure sense.

This case was treated along with attention to the motor developmental level throughout the course, and it was possible for the patient to achieve the corresponding motor development, but always cry. With age, his blisters became less severe, but he has been continuing to exhibit retarded motor development. On the other hand, mental activity went well. It is not denied that the stimulation of pressure sense and tactile sense are important for sensori-motor learning. Therefore, it is thought that the means of giving stimulations is especially important, and dermal property is an important point to be considered. Newborns have several morphological, physiological, and functional particularities. Newborns' skin is marked by incompletion, a thin horny layer, a frail structure, and weakness of resistance against physical and chemical stimulations. The implication is that skin damage is predisposed to occur. Pruritus, felt as itching, is a specific sense which generates reflectively intolerable displeasure, and physical and chemical stimulations are responsible for the induction of pruritus.

It is recommended that the physical therapist should make a plan to conduct activities of daily living and to facilitate motor learning in light of the above stated specific conditions. As the patient has problems with motor endurance and rapid motion at present, follow-up will be necessary.

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先天性表皮水疱症に対する理学療法

―症　例　報　告―

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要　旨　先天性表皮水疱症は厚生省による特定疾患治療研究対象疾患で，いわゆる「難病」指定疾患である．表皮水疱症では，日常生活で外力が加わる部位に反復する水疱を臨床症状の主体とし，軽微な外力による水疱発生が基本であるが，病変活発な時期では特に誘因なく発生する場合もあるという．著者らは，7年間にわたって表皮水疱症接合部型の男児に理学療法を実施してきた．体表面が外界と接触するということは乳幼児の感覚－運動学習過程において必要不可欠な体験であるにもかかわらず，水疱発生によってそのことが阻害され結果的に運動発達の遅れを呈した．今回，問題を残しながらも普通小学校への就学ゴールに達したので，これまでの理学療法経過と若干の考察を加えて報告する．

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