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Case Report

Long survival case of trisomy 13 mosaicism in a 7-year-old male

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SUMMARY

Trisomy 13 is a complication of various congenital abnormalities of the heart, brain, etc. Regarding the vital prognosis, many die within a year from birth. We herein report on the case of a 7-year 1-month-old boy with mosaicism trisomy 13 with the two considerations mentioned below as the cause for long-term survival in this case. The first is that there were no serious associated abnormalities to the heart, brain, or other organs, and the second is that a tracheotomy was carried out on a repeated respiratory infection with respiratory failure. Long-term in-home care was possible for the child and he was observed playing with toys by touching them. Trisomy 13 has a poor vital prognosis, so some argue that active treatment should be restrained. However, for cases with no severe associated abnormalities, long-term survival may be possible with active treatment.

Key Words : trisomy 13, mosaic, survival

INTRODUCTION

Trisomy 13 syndrome is a congenital anomaly syndrome caused by an excess of chromosome 13 that was first reported by Patau *et al.* in 1960, and may also be referred to as Patau syndrome. Regarding the symptoms and course of the present syndrome, death at an early phase following birth cannot be avoided due to serious congenital heart disease, respiratory illness, and brain malformation. According to reports, approximately 50% die within a month following birth and approximately 90% die within a year following birth. Moreover, the social prognosis is also very poor, with serious psychomotor retardation observed in all survival cases[1～5]. Therefore, some institutes inside and outside of Japan consider the present syndrome applicable to prenatal diagnosis. Moreover, a tendency is observed for active treatment not to be carried out[6]. On the other hand, though dispersive, there are reports of long-term survival along with the development of a medical environment in recent years[7,8]. In this paper, we report on the clinical course of mosaicism trisomy 13 in a 7-year-old boy.

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CASE REPORT

The case pertains to a 7-year one-month-old boy. He had no special family history. He was child of a father who was 47 years old and a mother who was 34 years old. There was no history of drug intake or infectious diseases to the mother during pregnancy. The child was born by spontaneous cephalic delivery on

the 39th week ; he weighed 3,366 g, had a height of 48.5 cm, a head circumference of 33.0 cm, and a chest circumference of 31.5 cm. His apgar score was 4 points at 1 minute, and 8 points at 5 minutes. The child suffered from a complication of meconium aspiration following delivery and management of artificial respiration was carried out in a neonatal intensive care unit. Hearing of the systolic phase and systolic phase and diastolic murmur was carried out regarding heart sounds, and an interatrial septal defect (defect of ostium secundum, or fossa ovalis), a ventricular septum defect (IV type : muscular septal defect), as well as patent truncal arteriosus were observed upon cardiac ultrasound. The abdomen was bloated and a complication of malrotation was observed upon contrast X-ray examination. The child was observed with the following various congenital anomalies. Regarding the skull, the forehead was slanted and deformed. A partial defect of the scalp was observed in the parietal region of the forehead. A cephalhematoma was observed on top of the head. Erythema was observed in the forehead and the posterior region of the neck. Both eyes had microphthalmia and the right eyelid was observed as failing to close in addition to having blepharoptosis. He had a saddle nose. Both auricles were large and observed to be in a lower position. There was no cleft lip but a V-shaped cleft palate was observed in the soft palate. The anteroposterior diameter of the trunk and chest was largely deformed, with an umbilical hernia observed. Bilateral inguinal hernia, hydrocele, cryptorchidism, and a concealed penis were observed in the external genitalia. There was an abduction limitation in flexion at the hip joint, his nails were arched, and his fingers were tapered and overlapped. His heels and soles were cradle — like. Mild spasticity was observed in the limbs.

Trisomy 13 was suspected from congenital defects, G-band chromosome banding (Fig. 1) was carried out upon approval of the parent, and the child was diagnosed with trisomy 13. A FISH method (FISH : fluorescence *in situ* hybridization) test using peripheral blood determined that it was a mosaicism trisomy 13 with 73.2% normal karyotype and 26.8% trisomy 13. We did not test the chromosome analysis by buccal mucosa and skin fibroblast.

He was withdrawn from artificial respiration after 2

Fig. 1 Chromosome analysis
Excess of No. 13 was observed upon G-band chromosome banding.

weeks but tubal feeding was carried out due to a suckling disorder and aspiration pneumonia. Partial convulsion of the extremities repeatedly occurred for 6 months. The oral administration of clonazepam was commenced with an observation of multifocal spike upon electroencephalogram. The parents were instructed on the manipulation of tubal feeding and care of the child, the child left the NICU following 11 months from birth at a weight of 4,849 g and a height of 60.2 cm, and in-home care was carried out following this.

After leaving the NICU, in-home tubal feeding was continued with the support of regional visiting hygienists. The interatrial septal defect was found to have closed upon ultrasonic waves at 2 years from birth. Medical treatment by an artificial respirator was required with a recurrence of lower respiratory tract infection. Tracheotomy was carried out after 2 months of artificial respiration management at 3 years old and respiratory infection declined. In-home oxygen therapy was introduced. Urinary tract infection that accompanied the concealed penis recurred at 5 years old, and vulvaplasty was carried out. Regarding motor development at 6 years old, the child was observed with an ability to turn over in bed, repeatedly touched toys that make noise, and smiled at his mother.

Epileptic seizures accompanying partial convulsions of the upper left limb were aggravated from 6 years old. He was observed as having a repeated spike in the central parietal area of right brain waves and carbamazepine was increased with a diagnosis of locally related epilepsy. Regarding the extremities, lower limb-dominant spastic paralysis was observed and deep tendon reflex generally accelerated. Regarding the cranial nervous system, the activity of facial muscles for expressions declined and disappearance of the eyelash reflex as well as attenuation of the vomiting reflex

were observed. Delayed contraction of the pupil response was observed regarding bilateral light reflexes. Atrophic changes in the bilateral optic nerves were observed upon funduscopy. There was no brain malformation upon brain MRI, with a relatively good myelinization. The left and right lateral ventricles expanded, hypoplasia of the transparent septum was observed. He entered a special elementary school for handicapped children at 7 years old.

DISCUSSION

We herein report on the course and various test results regarding a mosaicism trisomy 13 case with long-term survival. Among trisomy 13 cases, in which long-term survival is rare, we will discuss why the present case experienced long-term survival.

First, the therapeutic strategy towards trisomy 13 of our hospital is mentioned[8]. Our institute does not commit to consistently carrying out active treatment including surgical treatment and/or resuscitation simply due to the disease name of trisomy 13 alone. We persistently carry out therapeutic strategies in accordance with the severity and/or requests of family members. The apgar score at birth of the child in question did not correspond to severe asphyxia. In addition, a prenatal diagnosis was not carried out. Therefore, initially, the same resuscitation as for a normal child was carried out without clinically diagnosing as trisomy 13 from the multiple abnormalities alone. Consultation was repeated with the patient following diagnosis for mosaicism trisomy 13 at a later date, and the concurrence of respiratory complications was significantly reduced by carrying out artificial respiration management and tracheotomy. Moreover, urinary tract infections were also reduced by a combination of urological surgery. The child was not observed with serious heart failure, brain malformation such as holoprosencephaly, or status epilepticus, which are observed in trisomy 13 with early death. Furthermore, due to the request of the parents for in-home care, he was able to grow up with abundant love from his family by tubal feeing while repeatedly entering and leaving the hospital. It was discussed that the long-term survival of the child was possible due to the factors mentioned above.

We would like to consider a therapeutic strategy for trisomy 13 syndrome based on the present case. It was considered that if there is no serious cardiac insufficiency, brain malformation, or status epilepticus at an early postnatal period, the vital prognosis may improve by carrying out active management including surgical treatment with the approval of the parents. Kaneko et al reported an operation on the congenital cardiac anomalies with 9 cases of trisomy 13 from 2000 to 2005. This article shows that improvement of predominance for the duration of survival period of trisomy 13 which caught intensive cardiac management[9]. There are no multi-person comparative reports that include differences in medical environments regarding whether mosaicism trisomy 13 has long-term survival compared to normal complete trisomy 13[7,10]. Therefore, a possibility that the present case coincidentally had long-term survival cannot be ruled out. Similar reports may additionally increase in the future due to development of the medical environment in recent years[8]. Additional reports in the future are awaited regarding whether or not the findings of the present case are findings that are peculiar to mosaicism trisomy 13 syndrome.

In the near future, it is predicted that the day will come in which the vital prognosis or duration of life of trisomy 13 syndrome is further improved. Therefore, it is advisable for health care providers to ascertain the prognosis of each case regarding the possibility of how much a child with a disability will socially grow and provide the maximum QOL. The most important point in the medical care of a child with a chromosome abnormality is to absolutely avoid unnecessary life-prolonging treatment. That is to say, what is important is "how to live" and not "how long to live." Kosho expresses the problem of the genetic ethics of the child of the congenital chromosomal abnormalities such as trisomy 18 with "best interests of the babies"[11]. It is important to develop medical care for disabled children with this viewpoint as the core, with medical professionals and family members supporting each other.

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