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Long-term survival of full trisomy 13 in a 14 year old male: a case report

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Abstract. – Long term survival for the cases of trisomy 13 into over a first decade is very rare. We reported here the case of a 14-year-old male karyotype with full type of trisomy 13. In this clinical phenomenon, the case had typical facial, finger and limb anomalies for trisomy 13. Arterial septal defect and patent ductus arteriosus were recognized using ultrasonography after birth. Major cerebral malformation such as holoprosencephaly or cerebellar hypoplasia were also not revealed. After 5 months of his age, artificial ventilation therapy for dyspnea associated with laryngomalacia was required. A tracheotomy was performed at 6 months of his age. After 12 years old, intractable partial epilepsy was recognized. For his partial seizures, a treatment with a combination of two anti-epileptic drugs, valproic acid and levetiracetam, were advised. Now he is alive for 14-years-old and he is the 4th longest surviving patient with full karyotype of trisomy 13.

Key Words:
Survive, Prognosis, Patau syndrome, Epilepsy, Anomaly.

Introduction

Trisomy 13, also known as Patau syndrome, is a congenital genetic disorder caused by an excess of chromosome 13 firstly defined by Patau et al in 1960[1]. The incidence of this rare syndrome has been estimated at about 1:5,000 or 1:20,000 live births[1-2] and the third most viable trisomy syndrome after trisomy 21 (Down syndrome) and trisomy 18 (Edward's syndrome). The clinical spectrum of this syndrome exhibits a variety of anomalies, including central nervous malformation, facial, ocular, limb, finger, lung-cardiac system, uro-genital involvement and others. The prognosis of this syndrome is generally lethal. In addition, the social skill outcome with serious psychomotor retardation were observed in all of the long survival cases of trisomy 13[3]. There have been some cases of long survival individuals, who were reported earlier over 5 years old to the adolescent[4-20]. Here we reported and discussed a long-term survival case of a 14-year-old Japanese male with full karyotype of the trisomy 13.

Case Report

This patient was born cesarean section under the diagnosis of both intrauterine growth retardation and fetus bradycardia after 35 weeks of gestation, with body weight 1950 g and height of 46.0 cm, head circumference of 29.8 cm and chest circumference of 25.5 cm at private obstetrics and gynecology clinic. He was a first child of a father who was 41 years old and mother who was 37 years old. There was no history of infectious diseases and drug intake of the mother during pregnancy. Birth asphyxia was noted and his Apgar scores were 3 points at 1 minute and 6 points at 5 minutes. He had several minor anomalies including capillary hemangioma on forehead, low set ears, cryptorchidism, narrow finger and polydactyly. Cytogenic analysis with G-banding karioytype, performed using a peripheral blood specimen, revealed the presence of full type of trisomy 13. We did not test the chromosomal analysis of fibroblast of the skin and oral buccal mucosa. He had cardiac murmurs and was diagnosed with an arterial septal defect (size of the defect was 6.8 mm) as well as patent ductus arteriosus. Dyspnea due to both vocal cord paralysis with laryngomalacia and heart failure were noted and he required incubation and artificial ventilation therapy. At the 6 months of his age,

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after 5 months period of incubation treatment, had taken tracheotomy operation for difficult to respiratory condition. Respiratory problems were, therefore, controlled after the tracheotomy. At the 1 year and 3 months, the first episode of partial seizures of the upper limbs occurred and gradually increased. Electroencephalogram showed multifocal small spikes predominantly in both frontal areas, so that he was diagnosed as lateralized partial epilepsy. Seizures were well controlled by oral administration of valproic acid at that time. At the 5 years of his age, he revealed a severe motor and intellectual developmental delay. Brain magnetic resonance imaging estimated slightly delayed myelination. Moreover, dilations of lateral ventricles, bifurcated septum pellucid and thin of the corpus callosum were observed in fluid-attenuated inversion recovery sequence (Figure 1). At the age of 7 years, he had an intestinal hernia. After intestinal enhance examination, he was diagnosed intestinal malrotation and was operated. At the age of 12-year-old, his epilepsy with partial seizures of limbs became intractable. At the 13-year-old, sleep induced electroencephalogram showed poly spike with high voltage slow wave complex predominantly in both fronto-temporal areas (Figure 2). He took add on therapy with levetiracetam and his seizures were decreased but not ceased. When he was 14-years-old, his physical stature and weight were 147 cm and 29 kg, respectively

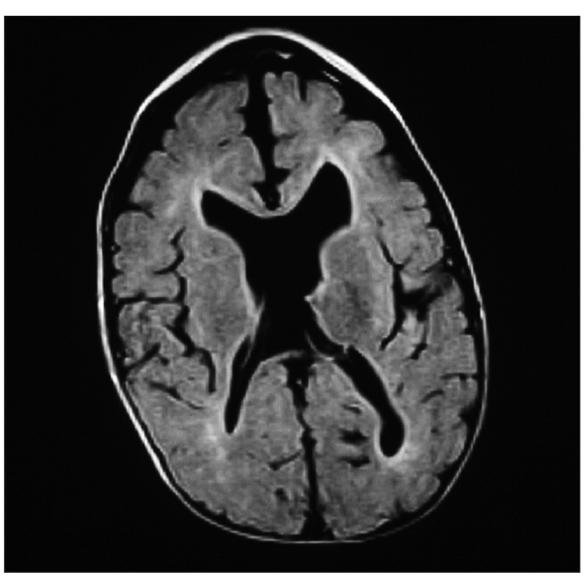

Figure 1. Brain fluid-attenuated inversion recovery (spin echo: echo time, 105.0 msec; repetition time, 9000 msec) magnetic resonance imaging of the brain at 5 years old.

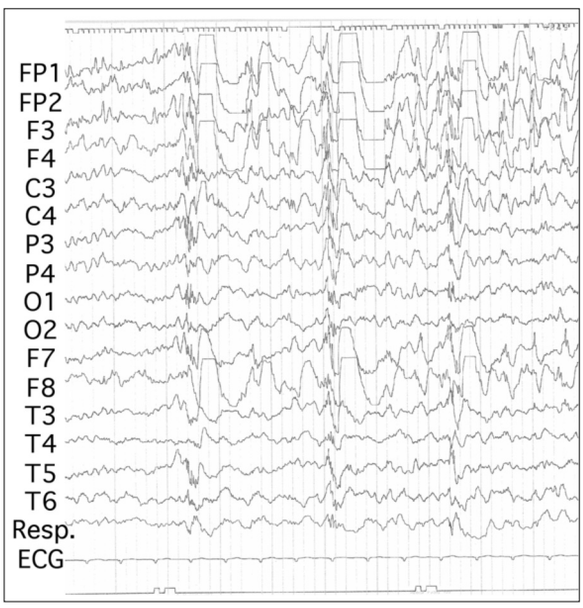

Figure 2. Sleep induced electroencephalogram record at 14 years of his age.

(Figure 3). He enjoyed touching the toy, smiling to hear his parent voice and he could roll over his body.

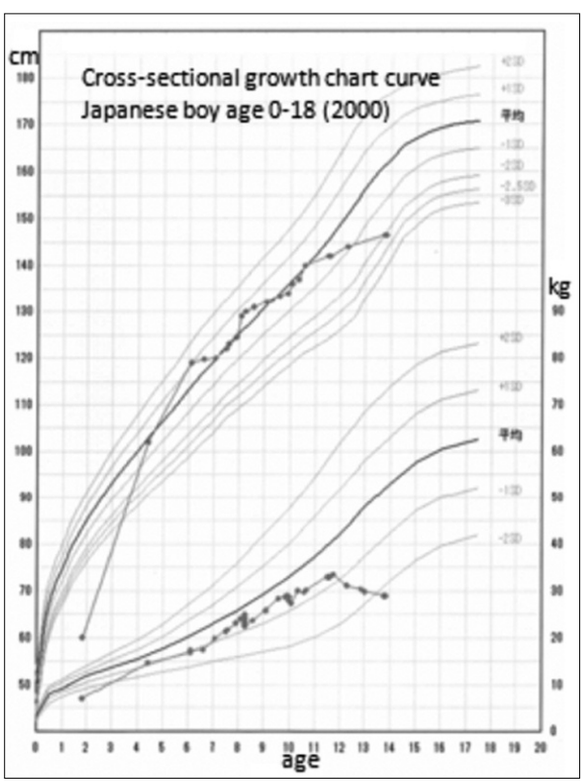

Figure 3. Cross-sectional growth chart curve of the patient (Japanese, boy, age 0-18, 2000 version).

Table I. Prolonged survival reported cases of trisomy 13.

Study (reference)	Patients age	Gender	Karyotype of trisomy 13
Marden PM et al 1967 (4)	10 years	Female	Full
Mankinen et al 1976 (5)	5 years	Female	Full
Cowen JM et al 1979 (6)	9 years	Male	Mosaic (normal/full)
Reardon PC et al 1981 (7)	10 years	Male	Mosaic (full/partial)
Reheendran R et al 1981 (8)	11 years	Male	Full
Reheendran R et al 1981 (8)	19 years	Female	Full
de Toni T et al 1983 (8)	7 years and 8 months	Male	Full
Singh KS 1990 (10)	22 years	Male	Mosaic (normal/full)
Zoll B et al 1993 (11)	11 years	Female	Full
Petit P et al 1994 (12)	38 years	Male	Mosaic (Normal/Full)
Delatycki M et al 1997 (13)	5 years and 1 month	Female	Mosaic (normal/robertsonian translocation)
Delatycki M et al 1997 (13)	9 years and 5 months	Male	Mosaic (normal/ full)
Tunca Y et al 2001 (14)	32 years	Female	Full
Iliopoulos D et al 2006 (15)	12 years and 2 months	Female	Full
Hsu HF et al 2007 (16)	7 years and 0 month	Male	Full
Fogu G et al 2008 (17)	12 years	Female	Mosaic (full/partial trisomy/ partial monosomy/ isochromosome)
Jacob FD et al 2010 (18)	19 years	Female	Full
Peroos S et al 2012 (19)	8 years and 5months	Female	Full
Imataka et al 2013 (20)	7 years and 4 months	Male	Mosaic (normal/full)
Our case	14 years and 0 month (alive)	Male	Full

Discussion

Rasmussen et al[21] reported that life prognosis of median survival time for trisomy 13 is between 7 and 10 days. Moreover, between 86% and 91% of a live-born infant with trisomy 13 do not survive beyond 1 year of their life. To best of our knowledge, there have only been twelve cases of patients with trisomy13 including four cases of trisomy 13 mosaicism, who survived over the first decade (Table I). An individual survived longest with full trisomy 13 was reported by Tunca et al[14], who was 32-years-old female. Tunca et al[14] described that non-lethal congenital anomalies such as brain, cardiac, uro-genital, etc., and the aggressive medical care may contribute to the long survival of patients affected by trisomy 13. Our case also had no brain, along with severe cardiovascular anomalies, so was treated with surgical tracheotomy approach. That case was the fourth longest survival individual with full karyotype of trisomy 13 in the previous literature.

In addition, as shown in Table I, longest survival case of trisomy 13 mosaicism was a 38-year-old female[12]. Some case reports showed the possibility of mosaic type trisomy 13 for longer average life expectancy than patient with full type of trisomy 13[16]. But others reported that survival prognosis is not at all related to the trisomy 13 mosaicism, with no reported evidence[12]. Another unique point observed in our case was the late onset epilepsy. Jacob et al[18] also reported a similar case which had late-onset seizures in an adolescent with full trisomy 13. When the period became long, the life survival of the patient with trisomy 13 in the future, epileptic's complication[22] may increase.

Conclusions

The cases of trisomy 13 have usually a poor life prognosis. However, for the cases with no severe vital associated organ anomalies, physician should recognize that long-term survival may be possible.

Conflict of Interest

The Authors declare that there are no conflicts of interest.

References

1) Patau K, Smith DW, Therman E, Inhorn SL, Wagner HP. Multiple congenital anomaly caused by extra chromosome. Lancet 1960; 1: 790-793.

2) Jones KL. Trisomy 13 syndrome. In: Smith's recognized patterns of human malformation. 6th Ed. Philadelphia, Elsevier Saunders, 2006; pp. 18-21.

3) Wyllie JP, Wright MJ, Burn J, Hunter S. Natural history of trisomy 13. Arch Dis Child 1994; 71: 343-345.

4) Marden PM, Yunis JJ. Trisomy D1 in a 10-year-old girl. Normal neutrophils and fetal hemoglobin. Am J Dis Child 1967; 114: 662-664.

5) Mankinnen CB, Sears JW. Trisomy 13 in a female over 5 years of age. J Med Genet 1976; 13: 157-161.

6) Cowen JM, Walker S, Harris F. Trisomy 13 and extended survival. J Med Genet 1973; 16: 155-157.

7) Reardon PC, Greenstein RM, Howard RO, Gianacopolos EK, Breg WR. Unusual mosaicism of de novo structural abnormalities and ocular anomalies in a male with 13 trisomy syndrome. Am J Med Genet 1981; 10: 113-118.

8) Redheendran R, Neu RL, Bannerman RM. Long survival in trisomy-13-syndrome: 21 cases including prolonged survival in two patients 11 and 19 years old. Am J Med Genet 1981; 8: 167-172.

9) de Toni T, Duillo MT, Cafiero V, Cavaliere G, Lazzaroni Fossati F. Long-term study (7 year and 8 months) of a case of trisomy 13 with long survival. Minerva Pediatr 1983; 35: 951-954. (in Italian)

10) Sigh KS. Trisomy 13 (Patau's syndrome): a rare cases of survival into adulthood. J Ment Defic Res 1990; 34: 91-93.

11) Zoll B, Wolf J, Lensing-Hebben D, Pruggmeyer M, Thorpe B. Trisomy 13 (Patau's syndrome) with an 11-year survival. Clin Genet 1993; 43: 46-50.

12) Petit P, Fryns JP. Normal/trisomy 13 mosaicism in a 38-year-old male. Genet Couns 1994; 5: 311-314.

13) Delatycki M, Gardner RJ. Three cases of trisomy 13 mosaicism and a review of the literature. Clin Genet 1997; 51: 403-407.

14) Tunca Y, Kadandale JS, Pivnick EK. Long-term survival in Patau syndrome. Clin Dysmorphol 2001; 10: 149-150.

15) Iliopoulos D, Sekerli E, Vassiliou G, Sidliropoulou V, Topalidis A, Dimopoulou D, Voyiatzis N. Patau syndrome with a long survival (146 months): a clinical report and review of literature. Am J Med Genet A 2006; 140: 92-93.

16) Hsu HF, Hou JW. Variable expressivity in Patau syndrome is not all related to trisomy 13 mosaicism. Am J Med Genet A 2007; 143A: 1739-1748.

17) Fogu G, Maserati E, Cambosu F, Moro MA, Poddie F, Soro G, Bandiera P, Serra G, Tusacciu G, Sanna G, Mazzarello V, Montella A. Patau syndrome with long survival in a case of unusual mosaic trisomy 13. Eur J Med Genet 2008; 51: 303-314.

18) Jacob FD, Ramaswamy V, Kolski H. Long-term survival and late anset seizures in an adolescent with trisomy 13. Can J Neurol Sci 2010; 37: 694-696.

19) Peroos S, Forsythe E, Pugh JH, Arthur-Farraj P, Hodes D. Longevity and Patau syndrome: what determines survival? BMJ Case Rep 2012; 2012. pii: bcr0620114381.

20) Imataka G, Yamanouchi H, Hirato J, Eguchi M, Kojima M, Honma K, Arisaka O. Autopsy report of a 7-year old patient with the mosaic trisomy 13. Cell Biochem Biophys 2013; 67: 813-817.

21) Rasmussen SA, Wong LY, Yang Q, May KM, Friedman JM. Population-based analyses of mortality in trisomy 13 and trisomy 18. Pediatrics 2003; 111: 777-784.

22) Imataka G, Tsuboi Y, Kano Y, Ogino K, Tsuchioka T, Ohnishi T, Kaji Y, Wake K, Ichikawa G, Suzumura H, Arisaka O. Treatment with mild brain hypothermia for cardiopulmonary resuscitation after myoclonic seizures in infant with robertsonian type of trisomy 13. Eur Rev Med Pharmacol Sci 2015; 19: 2852-2855.