CASE REPORT

A newborn with trisomy 13 who had tetralogy of Fallot and metopic synostosis: Case report

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Abstract

Background and Aim: Trisomy 13 (Patau syndrome) was first described by Patau et al in 1960. It is characterized by serious head, facial, and extremity anomalies, congenital heart defects, and mental abnormalities. The incidence rate of Trisomy 13 is 1/10.000 live births. Accompanying symptoms and findings vary in rate and severity among the cases. Tetralogy of Fallot and metopic synostosis are very rare abnormalities in patients with Trisomy 13. In this study, we aimed to present a newborn girl with trisomy 13 who had multiple congenital malformations accompanied by tetralogy of Fallot and metopic synostosis.

Description of the case: The patient was delivered at 40 weeks of gestation, and admitted to the neonatal intensive care unit due to respiratory distress and physical abnormalities. The newborn examination revealed multiple dysmorphic features. She had boot-shaped appearance on the chest radiograph. Chromosome analysis demonstrated mosaic trisomy 13.

Conclusion: Patients with trisomy 13 may have different type of gene variations and malformations; however, the most common type of gene variation is classic trisomy 47, XX +13, and the most common malformations are facial anomalies and congenital heart defects. In addition, tetralogy of Fallot and metopic synostosis may accompany trisomy 13**.** Hippokratia 2013; 17 (3): 268-270

Keywords: Trisomy 13, tetralogy of Fallot, metopic synostosis

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Introduction

Trisomy 13 (Patau syndrome) was first described by Patau in 1960[1]. Its incidence is 1/10,000 live birth, usually resulting in spontaneous abortion. Abortion may occur in early gestation period or be delayed until the 20th week of gestation. There may also be early birth[2].

The most frequent clinical features involve the central nervous system (mainly holoprocencephaly, microcephaly, severe psychomotor delay), ocular system (microphtalmia/anophthalmia), cardiovascular system (mainly septal defects or patent ductus arteriosus), and urogenital system (cystic kidneys, cryptorchidism). The majority of infants have orofacial clefts and postaxial polydactyly of the hands or feet[3]. Most of the patients with Patau syndrome have trisomic chromosome (47, XX+13), and most die within the first year of age.

Tetralogy of Fallot (TOF) is the most common type of cyanotic congenital heart disease, and consists of a right ventricular outflow tract obstruction, an overriding aorta, a malaligned ventricular septal defect, and right ventricular hypertrophy. It can have the classic boot-shaped appearance on chest radiograph, and it is very common in the patients with chromosomal abnormalities. TOF and metopic synostosis are rare malformations in Trisomy 13. In this study, we aimed to present a mature newborn girl with trisomy 13 who had TOF, metopic synostosis, and multiple congenital anomalies.

Case report

The patient was delivered at 40 weeks of gestation by normal spontaneous vaginal delivery. She was the eighth child born to non-consanguineous marriage from a healthy 42-year-old mother and 45-year-old father. Her sisters and brothers were normal and the family history was unremarkable. The birth weight of the patient was 2,460 gr (3rd - 5th percentile), length was 48 cm (10th - 25th percentile), and occipitofrontal circumference was 30 cm (< 3rd percentile). APGAR score was 7 at 5 min. She was admitted to the neonatal intensive care unit due to respiratory distress and physical abnormalities. The newborn examination revealed multiple dysmorphic features including metopic synostosis, broad nasal bridge, short nose, low-set ears, cyanosis, sloping forehead, pectus excavatum, hypoplasia of distal phalange of the fifth finger of the right hand, polydactyly of fingers (six finger), telangiectatic nevus, upturned nares, depressed

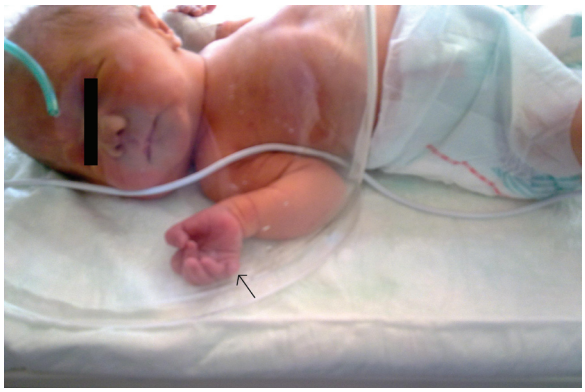

Figure 1: An image of the facial and physical features of the reported patient; metopic synostosis, broad and depressed nasal bridge, short nose, upturned nares, low-set ears, sloping forehead, pectus excavatum, hypoplasia of distal phalange of the fifth finger of the right hand and sixth finger in hands, upslanting palpebral fissures, short neck, high forehead, micrognathia.

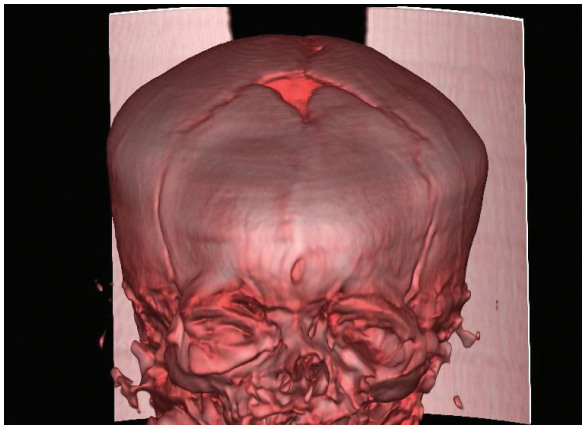

Figure 2: A section of the cranial 3D-computerized tomography revealed metopic synostosis.

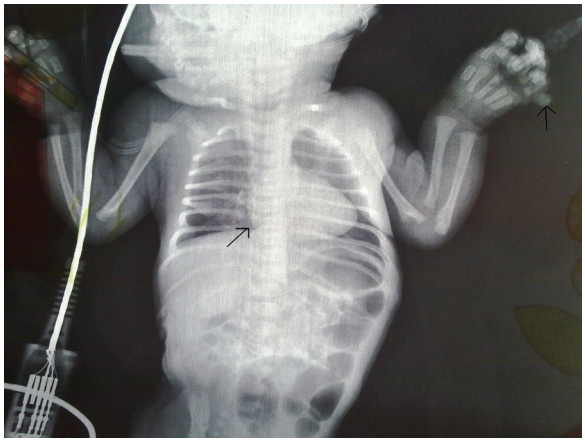

Figure 3: A section of the chest radiograph; boot-shaped appearance and sixth finger in hands.

nasal bridge, upslanting palpebral fissures, microtia, hort neck, high forehead, widely spaced nipples, micrognathia, and high palate (Figure 1).

The results of the complete blood count tests, biochemical laboratory studies, thyroid function tests and urine metabolic studies were normal. The cranial 3D-computerized tomography revealed metopic synostosis (Figure 2), the echocardiographic examination revealed TOF (including a right ventricular outflow tract obstruction, an overriding aorta, a malalignment ventricular septal defect, and right ventricular hypertrophy). In addition, the patient had boot-shaped appearance on the chest radiograph (Figure 3). Chromosome analysis demonstrated Mosaic trisomy 13 (47, XX +13) from peripheral blood leukocytes using GTG banding technique at 100 metaphase. The patient was transferred to the department of pediatric cardiac surgery on the sixth day of life. The patient is 2 months old and her general state remains well.

Discussion

Patau syndrome (Trisomy 13 syndrome) is seen at an incidence rate of 1/12.000-1/29.000 live births. It is, however, more common in spontaneous abortions than in live births[4]. Its characteristic findings are microophthalmia, and cleft lip and palate[5]. Cardinal findings are motor and mental retardation, microcephalus, holoprosencephaly, hypotelorism, and cardiovascular, genitourinary, and/or ocular malformations. The definitive diagnosis is established through chromosome analysis. Trisomy 13 may present with different type of gene variations such as classic trisomy 47, XX +13 (80%), translocation or structural changes (10%), mosaicism (5%) etc[5]. Phenotypically, the caryotypes of some cases with trisomy 13 symptoms are normal. Some of these cases may be Meckel syndrome; some, unrecognized trisomy 13/normal mosaic, and some may be the phenotypes of genetic or environmental origin. The examination of our patient revealed multiple dysmorphic features and in the caryotype analysis, classic trisomy 47, XX +13 was determined.

Advanced maternal age is associated with the occurrence of the syndrome since the advanced age of the mother (over 35 years of age) leads to nondisjunction of chromosomes[6]. The mother of our patient was 42 years old, and father, 45 years old. The congenital irregularities in patients with Patau syndrome are life-threatening and almost 50% of the cases die in the first month and 90%, within the first year of life. All the cases suffer motor and mental deficits. However, mosaics may improve in time[7]. Although our case had classical trisomy 13 syndrome, the control examination in the 2nd month of age showed no significant health problems. The most common cause of death is cardiopulmonary complications such as TOF. In three reviews of published cases with trisomy 13, the most common malformations were facial anomalies and congenital heart defects, and TOF and metopic synostosis are rare malformations, with incidence of TOF at 15 % [3,8-11]. Our patient had facial anomalies, extremity anomalies, and TOF. Comparing of physical examination findings in

Table 1: Physical examination findings in the reported patients[8-11] and our patient with Trisomy 13.

Physical examination findings	Frequency	% of cases	Reported case
Broad nasal bridge	15/16	94	+
Short nose	4/4*	100	+
Sloping forehead	10/14	71	+
Telangiectatic nevus	5/5	100	+
Upturned nares	5/5	100	+
Uterine anomalies	3/4	75	-
Depressed nasal bridge	8/9	89	+
Cryptorchidism	6/8	75	-
Upslanting palpebral fissures	7/8	88	+
Low-set ears	26/30	87	-
Epicanthal folds	6/7	86	-
Microtia	5/7	71	+
Skin redundancy	6/7	86	-
Apnea	4/6	67	-
Finger clinodactyly	8/11	73	-
Frequent respiratory infections	4/6	67	-
Low posterior hairline	4/6	67	-
Pigmentary abnormalities	4/6	67	-
Short neck	9/11	82	+
Small forehead	5/6	83	-
Tapered fingers	4/6	67	-
Patent ductus arteriosus	7/10	70	-
Cleft palate	27/36	74	-
Facial asymmetry	3/5	60	-
Flat occiput	4/5	80	-
Hearing loss	6/9	67	-
High forehead	3/5	60	+
Poor suck/swallow	4/5	80	-
Malformed ears	12/17	76	-
Seizures	14/20	70	-
Hypotelorism	11/21	52	-
Joint contractures	8/12	67	-
Vertebral anomalies	9/21	43	-
Widely spaced nipples	5/8	63	+
Ventricular septal defect	20/24	83	+
Micrognathia	14/18	78	+
Clenched hands	5/7	71	-
Finger camptodactyly	7/12	58	-
High palate	8/12	67	+
Umbilical hernia	6/22	27	-
Cleft lip	16/27	59	-
Downslanting palpebral fissures	3/6	50	-
Hypoplastic nose	4/6	67	-
Pterygium	4/6	67	-
Microcephaly	8/13	62	+
Two-vessel umbilical cord	3/5	60	-
Atrial septal defect	17/23	74	+
Rib anomalies	8/20	40	-
Finger brachydactyly	3/7	43	+
Polydactyly of fingers	7/13	54	+
Polydactyly of toes	6/13	46	-
Thin upper lip	4/7	57	+
Tetralogy of Fallot	2/13	15	+

the reported patients[8-11] and our patient are presented in Table 1.

In conclusion, patients with trisomy 13 may have different type of gene variations and malformations; however, the most common type of gene variation is classic trisomy 47, XX +13, and the most common malformations are facial anomalies and congenital heart defects. In addition, TOF and and metopic synostosis may accompany trisomy 13.

Conflict of interest

There is no conflict of interest.

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