Variety of neuropsychiatric symptoms in a 4-year old girl suffering from Patau Syndrome with translocation

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Abstract

Background: Patau Syndrome is a congenital disorder caused by different abnormalities of the 13th chromosome, such as trisomy, robertsonian translocation or mosaicism. It is a rare disorder, affecting some 1/12.000-1/29.000 newborns. Only 10% of cases are caused by robertsonian translocation and no more than 10% of the newborns live up to 1 year. Our patient is already at preschool age and still stays in relatively good somatic condition. That is why she represents an absolutely unique case of this pathology.

Case report: The patient is a 4-year old girl, born in the 38th week of pregnancy with oligohydramnios. Microcephaly, polydactyly, cleft lift and palate, ureteral stricture and umbilical hernia were diagnosed after birth. Nowadays multiple neurological and psychiatric symptoms are present, overlapping somatic problems. Apart from the intellectual and psychomotor retardation, additional idiopathic epileptic seizures, motor hyperactivity, aggression and auto aggression, sleep disorders and concentration deficits occur. The patients was diagnosed to have autism spectrum.

Conclusions: Despite pharmacotherapy, the patient still presents conduct disorders, which makes her treatment and daily care much more difficult for both parents and medical staff. Due to the congenital etiology of disorder, prognosis is rather pessimistic.

Keywords: Patau syndrome, trisomy 13, 13q2q3q, autism spectrum disorder, epilepsy

Streszczenie

Introduction

Trisomy 13 (Patau syndrome) was first described by Patau in 1960 [1]. Its incidence is 1/12.000 - 1/29.000 live birth [2], usually resulting in a spontaneous abortion. Abortion may occur in early gestation period or be delayed until the 20th week of gestation. The condition may also trigger an early birth [3].

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 abnormalities (cystic kidneys, cryptorchidism). Most infants have orofacial clefts and postaxial polydactyly of the hands or feet [4].

Patau Syndrome is caused by different abnormalities of the 13th chromosome, such as trisomy, robertsonian translocation or mosaicism. Up to 10% of cases are caused by robertsonian translocation and no more than 10% of the patients survive longer than a year [5]. The authors of this study present a unique case of a child suffering from Patau Syndrome with translocation. The child is already at preschool age and still stays in relatively good somatic condition.

Case report

The patient is female, delivered at 38 weeks of gestation. She was born as the first child, with no remarkable incidents in the family history. Due to fetal abnormalities and oligohydramnios detected through an ultrasound examination, a caesarean section was performed. The newborn examination revealed multiple dysmorphic features including microcephaly, cleft lip and palate, congenital talipes equinovarus, polydactyly, scoliosis, ureteral stricture and umbilical hernia. Chromosome analysis revealed trisomy 13 with translocation: 13q2q3q of which there are only around 50 cases all over the world.

Due to numerous somatic issues, the patient had to be hospitalized numerous times. Two surgical procedures were performed, due to her cleft lip and palate. She was also hospitalized in Pediatric Neurological Rehabilitation Unit and Pediatric Neurology Department, where she was diagnosed with unspecified paralytic syndrome (G83.9), unspecified myoneural disorder (G70.9) and unspecified epilepsy (G40.9). Additionally, an MRI scan revealed multiple disseminated ischemic-hypoxemic changes in the brain. The patient is also looked after by clinics of nephrology, cardiology, rehabilitation, neurology, surgery, and orthopedic outpatient units.

Apart from numerous somatic problems, the patient is severely intellectually disabled (F.72). At the age of three, she was already 10 months retarded which means very slow development rate at that age. She is unable to speak or obey any commands. In addition, the girl started to manifest conduct disorders, like hyperactivity, disrupted attentional control, both aggression and autoagression, autostimulation and sleep disorders, like sleepwalking. Interestingly enough, the patient became extremely focused on the shadows that other people cast and she could play with her own shadow all day long provided that she is allowed to do so. Since those symptoms started to intensify, a psychiatric consultation was ordered. Patient was diagnosed with autism spectrum disorder (ASD), which is a neurodevelopmental disorder often associated with chromosomal abnormalities.

ASD symptoms may vary from one child to another, yet overall they fall into two areas: social impairment, including difficulties with social communication and repetitive, stereotyped behaviors. The patient presented both features.

The main goals of treating children with autism are to lessen the deficits associated with the condition, as well as lower the family distress, by improving the quality of life and making them more independent. There is no single cure that can be deemed as the most effective, since the treatment is typically tailored to the child's needs [6]. Since neuropsychological interventions were already introduced, pharmacotherapy was recommended. Patient was prescribed Haloperidol (Haloperidolum guttae 0.2%)

Initially, the patient's behavior and sleep quality improved as a result of taking haloperidol. Unfortunately, the hyperactivity and aggression returned, accompanied by stereotypies. Haloperidol was replaced by chlorpromazine (chlorpromazinum guttae 4%), and then switched to risperidon. No single medication was effective for longer than a few weeks, after which the symptoms returned.

Due to pathological EEG results, firstly valproic acid and then also carbamasepine was implemented which helped to reduce the number of epileptic spasms.

Discussion

Patau syndrome (Trisomy 13 syndrome) is reported to have an incidence rate of 1/12.000-1/29.000 live births. Yet, it is more common in spontaneous abortions than in live births. Its characteristic findings include microophthalmia, cleft lip and palate [6]. Cardinal findings are psychomotor and mental retardation, microcephalus, holoprosencephaly, hypotelorism, and cardiovascular, genitourinary, and/or ocular malformations. The definitive diagnosis is made after a chromosome analysis is conducted. Trisomy 13 may manifest itself with different types of gene variations such as classic trisomy 47, XX +13 (80%), translocation or structural changes (10%), mosaicism (5%) etc [7].

Our patient is already four years old. She presents features typical for Patau syndrome: dysmorphic and somatic issues, like cleft lip and palate or urogenital defects. Although she suffers from mental and psychomotor retardation, her functioning is satisfying, as long as her underlying disease is concerned.

Unfortunately, despite the drug therapy, the patient still presents conduct disorders, which make her treatment and daily care much more difficult for both parents and the medical staff. Due to the congenital etiology of the disorder, the prognosis is rather pessimistic.

Conclusions

Although Patau syndrome is a very serious, incurable disease, with multisystemic signs and symptoms, we can still try to treat some of the patient's ailments. Even a slight improvement in patient's physical and mental health can make their daily life much more convenient, which is the reason why diagnosing of every single manifestation of any comorbid condition is so important. Our patient is a perfect example showing that attentive, multisystemic care can prolong patients' life and improve their condition to a certain degree.

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