Edwards syndrome in a 6-year old girl

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Abstract

Trisomy 18 (Edwards syndrome – ES) is the second most common trisomy. It occurs in 1/3 000 to 1/8 000 births. ES is a cause of numerous developmental disorders and malformations. The median life span of children with ES is about 2 weeks and only 5%–10% will survive their first year of life. The report presents a case of a 6-year-old girl with ES.

CASE REPORT

INTRODUCTION

Among autosomal trisomies only trisomies 21, 18 and 13 have led to a live birth. Trisomy 18 (Edwards syndrome – ES) is the second most common trisomy. (Figure 1.) It was first described by John H. Edwards (1928–2007) in 1960. The incidence of the syndrome is estimated as 1/3000 to 1/8000 live births and over 70% of pregnancies end in spontaneous abortion. (Altug-Teber *et al.* 2008; Richmond 2007; Rasmussen *et al.* 2003; Forrester *et al.* 1999; Charrow 2007; Hsiang-Yu *et al.* 2006; Brewer *et al.* 2002; Morris and Savva 2008).

Trisomy 18 is a cause of numerous developmental anomalies and malformations (Table 1). The clinical picture of Edwards syndrome includes: low birth weight resulting from intrauterine dystrophy, malformations of motor system and head, congenital cardiovascular disease and significant mental retardation. The most commonly observed malformations in newborn babies with ES are: clenched hands with overriding fingers (95%), rocker-bottom feet (90%), low-set, malformed ears (90%) as well as genitourinary (51%) and digestive system anomalies (31%, among others atresia, tracheo-oesophageal fistula). A lot of these malformations occur at the same time creating the pentalogy of Cantrell (umbilical hernia, anterior diaphragmatic hernia, sternal cleft, ectopia cordis, ventricular septal defect). Ninety per cent of the cases are associated with delayed /inhibited intrauterine development. In about 85% of the cases thrombocytopenia was detected. Frequent bronchitis, pneumonia, urinary tract infections as well as pulmonary hypertension and seizures also affect child's general condition (Forrester *et al.* 1999; Charrow 2007; Hsiang-Yu *et al.* 2006; Sell 1997; Baty *et al.* 1994; Embleton *et al.* 1996; Goc *et al.* 2006; Kosho *et al.* 2006; Wiedermeier *et al* 2008; Tucker *et al.* 2007).

The median life span of children with ES is about two weeks. Majority of authors have reported that from among live born children with ES 90%–95% die within their first year of life. Longer survival is rare, although there were cases of children born with this syndrome who lived to the age of nineteen and twenty but those were less severe cases of mosaic ES or deleted extra chromosome (Rasmus-

sen *et al.* 2003; Hsiang-Yu *et al.* 2006; Brewer *et al.* 2002; Baty *et al.* 1994; Goc *et al.* 2006; Niedrist *et al.* 2006; Root and Carey 1994; Nembhard *et al.* 2001; Kelly *et al.* 2006; Petek *et al.* 2003).

CASE REPORT

A girl A.P. six years old, born in 2003 at 37 week of gestation. Birth weight was 1 650 g, body length 42 cm, the Apgar score at 1 min was 3, at 3 min was 5 and at 8 min was 8. At birth the child presented cleft palate, narrow mouth, narrow palpebral fissures and very low-set ears (Figure 2). In the motor system the following were detected: clenched hands, particularly the right one, hammer toes and decreased muscle tone. Further examinations concerning cardiovascular system revealed ventricular septal defect and patent ductus arteriosus. Blood cell count demonstrated thrombocytopenia at the level of 68 000/ml. Cytogenetic examination confirmed Trisomy 18 (47, xx, +18). Earlier ultrasonography at 30 week of gestation revealed ventricular septal defect and low weight of the fetus.

Further on, profound mental and physical retardation were diagnosed. Psychological tests performed in 2006 with the use of Brunet-Lézine scale demonstrated significant psychomotor retardation. At that time the girl was 4 years and 3 months old whereas her developmental age was estimated as 9 months (Table 2).

At present the child demonstrates low body weight (height 104 cm, body weight 11.8 kg), muscular atrophy with marked reduction of strength, normal passive mobility of all upper and lower extremities joints with a tendency to excessive extension of limbs. The observed difficulties in unaided sitting and standing are caused by significantly decreased tone of back muscles. Weakening of trunk postural muscles is accompanied by right-sided myopathic scoliosis in Th6–Th12 = 65°dx and L1–L5 = 57°sin. (Figure 3).

Tab. 1. Occurrence of malformations and developmental anomalies (most frequent).

Kind of malformation	%
Delayed intrauterine development	90
Clenched hands	95
Rocker-bottom feet	90
Short breast bone	95
Narrow pelvis	70
Prominent occiput	70
Scoliosis	70
Low-set ears	90
Narrow palpebral fissures	80
Congenital hart diseases including:	95
Ventricular septal defect	90
Patent ductus arteriosus	80
Atrial septal defect	75
Thrombocytopenia	85
CNS defect	80
Mental retardation	100

Tab. 2. Estimation of psychomotor development acc. to Brunet-Lézine scale.

Category	Estimation
Age/acc.to birth certificate	4 years 3 months
Developmental age	9 months
Locomotion	9 months
Visual and motor coordination	7 months
Communication	6 months

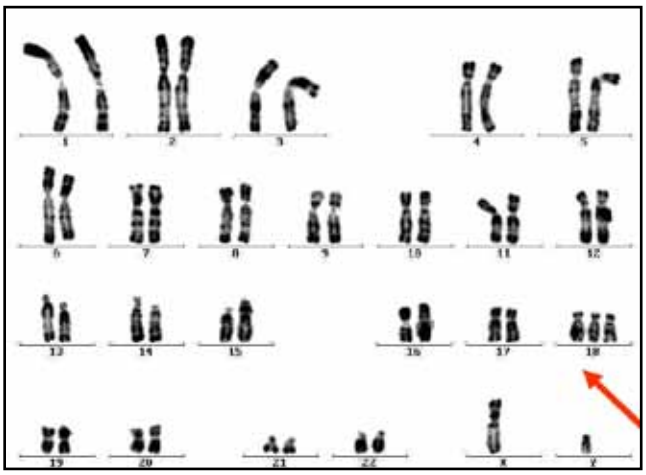

Fig. 1. Kariogram - trisomy 18.

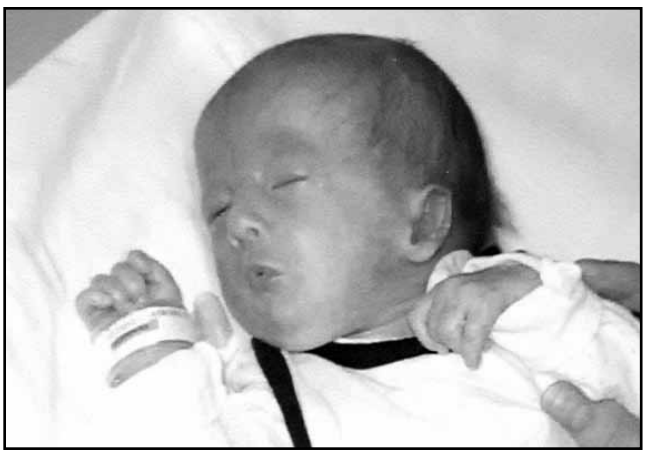

Fig. 2. A. P. in 8 week of life.

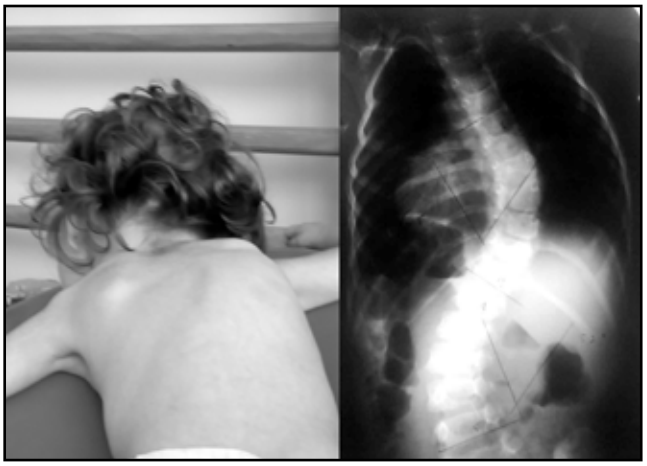

Fig. 3. Right-sided myopathic scoliosis.

Fig. 5. Learning motor coordination.

Fig. 4. Assuming erect position.

Fig. 6. SpineCore brace.

Rehabilitation is performed according to PNF method. Elements of educational kinesiology acc. to Dennison are also introduced. The aim of the carried out therapeutic rehabilitation is to prevent joint malformation and to maintain full range of their movement, improvement of muscle strength, formation of proper coordination of motor function as well as assuming erect position and learning locomotion (Figures 4 and 5).

SpineCor dynamic brace and corrective exercises are applied to provide progressive correction of scoliosis. The brace facilitates assuming erect position (Figure 6).

DISCUSION

A child with Edwards syndrome is a serious medical and ethical problem (Rasmussen *et al.* 2003; Hsiang-Yu *et al.* 2006; McGraw and Perlman 2008; Simón-Bautista *et al.* 2008; Bos *et al.* 1992; Hammerman *et al.* 1998; Piastra *et al.* 2003). Death among these children results from increasing, difficult to control, heart and respiratory failure developed due to organic heart diseases, pulmonary hypertension with accompanying pulmonary haemorrhage, arterial hypertension and respiratory failure of central origin (Embleton *et al.* 1996; Goc *et al.* 2006; Kosho *et al.* 2006).

Children with ES require treatment immediately after birth in intensive neonatal care units and surgical correction of life threatening malformations. Owing to greater and greater possibilities in these fields, the percentage of children who survived one month has clearly increased. However, only 5%–10% survive their first year of life (Brewer *et al.* 2002; Goc *et al.* 2006; Bos *et al.* 1992). Thus, some authors raise a question: whether undertaking the invasive methods of treatment is in

agreement with the ethics and the decision is made with parents (Hsiang-Yu *et al.* 2006; Kosho *et al.* 2006; Piastra *et al.* 2003; Walker *et al.* 2008).

CONCLUSION

Profound physical retardation has been observed in children who survived longer than 12 months. That is why intensive programme of therapeutic rehabilitation should be introduced acc. to neurophysiological methods. Rehabilitation allows to improve muscle strength, heart and respiratory efficiency, motor coordination and malformation of joints and body posture (Simón-Bautista *et al.* 2008).

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