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EXTENDED ABSTRACT OF A CASE REPORT

PECULIAR PROPERTIES OF HEMIHYPERTROPHY IN PATIENT WITH BECKWITH-WIEDEMANN SYNDROM. POSSIBILITY OF ITS CONSERVATIVE TREATMENT

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Summary

Beckwith-Wiedemann syndrome (BWS) is characterized by a combination of three classic signs (macrosomia, omphalocele, macroglossia) with a predisposition to the formation of malignant tumors and hemihypertrophy. It should be noted that unilateral increase in the longitudinal dimensions of one of the lower limbs leads to significant and persistent changes in locomotor system biomechanics on the whole. The incidence of BWS is 1: 15,000 of newborns.

Keywords: Beckwith-Wiedemann syndrome, hemihypertrophy, different length of the lower limbs, magnetic therapy

Introduction

Currently, it is believed that BWS, first described by J.B, Beckwith in 1963 and H.R. Wiedemann in 1964 [3,7], is inherited in an autosomal dominant manner with incomplete penetrance [2,5]. Such an opinion was formed due to development of molecular diagnostic methods with high-resolution, that allowed to discover a partial trisomy of the chromosome 11 short arm distal portion [4,6]. Structural abnormalities of the chromosome 11 short arm distal portion can be inherited from both parents.

The minimum diagnostic criteria for BWS, considered today, are the combination of macrosomia, omphalocele, macroglossia with normal karyotype [2,5]. The vast majority of patients has a hemihypertrophy [2].

During three years we observed a BWS patient with a complete set of minimal diagnostic signs listed above. The main orthopedic disorder in this patient is the progressing unilateral hemihypertrophy. At the time of initial treatment in our clinic the child was 5 months old.

Case report

The child was born on the seventh pregnancy, third premature childbirth at 33 weeks. Although prematurity, the child was born with signs of macrosomia (birth weight - 3510 g, body length – 52 cm). The hemihyperplasia of right half of the body and limbs, mycrogyria, partial atrophy of the optic nerves, delayed psychomotor development, macroglossia and hypertrophy of the right kidney were diagnosed just after the birth. The BWS with karyotype 46, XY was diagnosed in the Medical-genetic center of the city Ufa.

This work is the result of observation and treatment of BWS patient during 3 years. As the main diagnostic methods the anthropometry and roentgenometry were used. The obtained results were subjected to statistical processing and mathematical modelling.

Results

At the time of initial treatment in our clinic (age of patient 5 months) the relative difference in lower limbs length was 2.0-2.5 cm (according to anthropometry) and the absolute difference in the length of thighs with shins - 1.2 cm (according to roentgenometry). The left thigh circumference was greater then the right one by 3 cm. The difference in feet length was 1.5 cm. As the precise measurements and roentgenometry at the place of patient residence have not been made, assessment of the hemihypertrophy progression was impossible at the initial examination. The dynamic observation was recommended.

During re-examination after 4 months (age of patient 9 months) along with increase of body length in 7 cm the following dynamics was found: the relative difference in lower limbs length increased from 2.0 cm to 4.0 cm (according to anthropometry) and the absolute difference - from 1.2 cm to 2.4 cm (according to roentgenometry).

Because of hemihypertrophy progression and the beginning of self-walking, a course of comprehensive treatment was performed. The essence of this treatment was impact on growth plates of limb bones based on low-intensity magnetotherapy (Patent of Russian Federation №2212258). It allowed to inhibit the functional activity of "giant" limb growth plates and to stimulate it of contralateral one [1]. To enhance the treatment efficiency the additional procedures were performed improving bloodstream in stunted limbs growth plates. The control examination and diagnosis were carried out 4 months later after the first course of treatment. The further intensive progression of the defect (approximately twice) could be expected due to growth increasing by 6 cm during this period. However, the relative difference in lower limbs length according anthropometry does not exceed 3 cm (4 months ago it was 4 cm). The absolute difference in lower limbs length according roentgenometry does not exceed 2.5 cm. Thus, a significant progression of the defect has not occurred during the observed period contrary to the results of previous surveys. To date, the patient has received 6 similar treatment courses. He grew up in 3 years by 24 cm. The relative difference in lower limbs length according anthropometry is 3.5 cm, the absolute difference according to roentgenometry - 2.8 cm.

Discussion

The expected difference in lower limbs length was estimated at more than 8 cm in mathematical modelling of the rate of progression of the defect, taking into account the history.

As a result of treatment the progression of left-sided hemihypertrophy was slowed down. Currently a difference in lower limbs length is 3.5 cm (2.8 cm according to roentgenometry), wile the growth has increased during this period by 24 cm. The treatment continues.

Conclusion

Hemihypretrophy in BWS patients has a progressing character

The rate of progression of the defect (the difference in leg length) depends on the intensity of growth

Conservative treatment, based on non-invasive management of the functional activity of the lower limbs growth plates, slows the progression of hemihypertrophy.

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