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A CASE WITH SILVER – RUSSELL SINDROME: THE EFFICACY OT TREATMENT WITH GROWTH HORMONE, Case report

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Abstract

Silver –Russell Syndrome (SRS) is rare syndrome, described in a separate way from Silver and Russel as a syndrome of "intrauterine dwarfism"with so many clinical phenotypes ranging from mild to classic (1,2). Different studies estimate the incidence of SRS that varies widely from 1:3000 to 1:100,000 people (2,3,4).

SRS is a heterogeneous disorder and the basic of the underlying defect is not known jet (3,5,6). Various molecular defects have been reported, mostly involving chromosomes 7 and 17 (7,8,9).

Here we present our first case in our Pediatric – Endocrinology Clinic,a 5 years old boy with a classic form of the syndrome. He had a severe deficit of height and weight (< -4DS) at presantation.GH deficiency, with no other hormonal deficits is established. He started treatment with growth hormone and had a good catch-up growth. After 11 years of treatment he achived a final height up to 159.7cm (-1.8DS), but although it was 0.5 SD less than his predicted height. The weight improvement was more slowly, not helping growth hormone therapy to have the best results.

Keywords: Silver –Russell syndrome, growth hormone.

Abbreviations: GH - Growth Hormone, SD – standard deviations

Introduction

Silver- Russell Syndrome (SRS), is both clinically and genetically a heterogeneous disorder and the basic underlying defect for this is not known (10). Except of other dismorphic features, this syndrome is also characterized by a slow growth before and after birth and is one of the reasons of the definitive short stature in males and females. Males with RSS can arrive a finale stature up to 151cm, and women with RSS up to 140cm (11,12,13).

There are over 400 case described in the literature with this syndrome, from the moment of its discovery, either classical or mild forms (14).

We are presenting our first case diagnosed with SRS in our clinic, followed for 11 years, and the results of his treatment with growth hormone (GH).

Case report

The child F.B., born 21.01 .1995, originates from the north of Albania was presented in our clinic at 21 .03. 2000, and was admitted in our hospital with the diagnosis: Short stature for exploration. In this moment he was 5 2/12 years old. His weight was 8 kg (<<- 4 DS aged matched), the height 92 cm (<< - 4 DS aged matched), bone age was -18mo-2yrs. His statural age at five yrs. was like a child of 30 months.

He was born in terme, from a normal pregnancy, but with a very low birth weight – 1300gr. Later on, he did not gain weight properly. He was very weak, lethargic in first days of life, did not eat well. He continued this way in the first 2 years of life mainly, with failure to thrive and in the same way even in coming years with a bad appetite, weak and without energy as a child.

He did not have infectious disease during infancy, besides some common viral infections.

In the physical examination at age of five years, you could see:

1. A very short child, but with a deeper deficit in weight,
2. A clear asymmetry of the body, with the right side shorter and thinner then the left side.
3. The head looks relatively bigger compared to the face and the whole body.
4. A small and triangle-looking face with expressed