EVALUATION OF GROWTH PATTERN IN *PRADER-WILLI SYNDROME*

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Background

The main reason of decrement of growth in children with Prader-Willi Syndrome (PWS) is dysfunction of hypothalamo-hypophseal axis (HHA) and a decrease in the capacity of secretion of growth hormone (GH). In fact, in some cases, GH levels are normal, so there may be other factors in the etiology.

Figure 1. There are many different organisations for children with PWS.

Case

13 months old male patients admitted to pediatric endocrinology department. He was born to unrelated healthy parents, on 40th gestational week, 3000 gram. When he was 8 months old, genetic analysis was performed due to his dysmorphic features and he was diagnosed PWS. He had flapped ears, small hands and feet, scrotal hypoplasia, descended testicle and delay of neuro-motor development on physical examination. His anthropometric measurements were shown in table 1. Thyroid function tests, IGF1, IGFBP3 levels were normal. ***When he was 19 months old, a decrement in height percentile was observed***. ***His growth velocity was 3.8 cm/6 months, IGF levels were normal, height percentile was greater than 3 percentile, bone age was appropriate to his age*** **so** before performing growth hormone stimulation tests, neurosecretory dysfunction was evaluated. 4 hours of sleep profile was obtained in order to evaluate HHA. GH peak levels were low (<15 ng/dl), mean density was low (<3,5 ng/dl), and number of peak levels was low (<3 times 5ng/dl). He was diagnosed neurosecratory dysfunction. Radiologic evaluation of neurohypophysis was normal.

Figure 2. Deletion of paternal chromosome 15 is seen in PWS.

Table 1. *Anthropometric evaluation of the case*

Anthropometry	13 months	18 months
Weight (kg)	11.5 (75-90p)	12 (25-50p)
Height (cm)	77.7 (50p	81.5 (25-50p)
Height SDS	1.01	0.1
Body mass index (kg/ m²)	19.16	18.07
Bone Age (ay)	15	18
IGF 1 (ng/ml)	74.4 (0,1 SDS)	255 (2.3 , 3 SDS)
IGFBP3 (ng/ml)	4933 (>3SDS)	2963 (1.28, 2 SDS)

Discussion

90% of etiology of pathological short stature in PWS cases is due to GH deficiency. Cases with NSD, as in our case in younger ages, are very rare. ***NSD may be a prodromal stage before evident GH deficiency develops in PWS cases***. There isn't any consensus about treatment in this stage. In fact, there are a few opinions such as early GH treatment may normalize body fat composition and may help reaching target height. This case is represented to discuss the early GH treatment in PWS cases diagnosed with NSD in younger ages

References

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