Central Precocious Puberty Appeared in Infancy Period in a Patient of Sotos Syndrome

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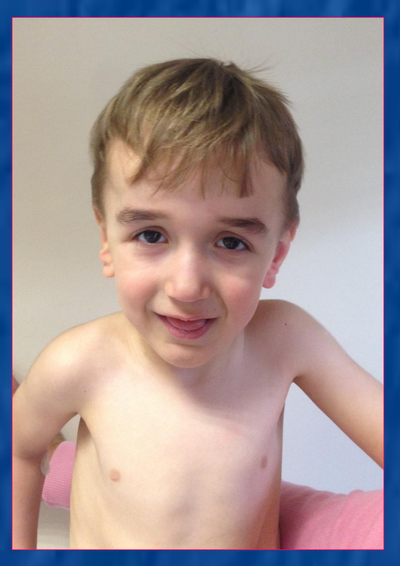
Background: **Sotos syndrome is a rare syndrome; with distinctive clinical findings include typical facial appearance, learning disability; and overgrowth. Advanced bone age can be detected in some cases while precocious puberty reported only in two cases until now.**

Case:

- **A 6,5 months of age male infant admitted to clinic with neuromotor delay and macrogenitalia. He was second child of unrelated healthy parents, and birth-weight was 4200g. In physical examination: height: 81 cm (HSDS: +5,6), weight: 10.5 kg, RBMI was 90%, testicular volume 4 ml bilaterally, penis length was 6.7 cm. Mild facial dysmorphism with global developmental delay were noticed.**
- **In laboratory evaluation, high basal testosterone level, and high basal and stimulated gonadotropins (Table 1) were confirmed central precocious puberty. Cranial imaging studies revealed normal pituitary gland.**



LHRH analogue at dose of 250 mcg/kg/month was started. Because the HHG axis was not controlled efficiently, the dose of LHRHa increased to 500mcg/kg/month. At the age of 2,5 years, increase of testicular volume to 8 ml and penile length to 9 cm were detected. Bone age advanced to 4.5 years. Cyproterone acetate 50 mg/day was added to treatment. With combined treatment, patient's clinical and laboratory progression was controlled.

Hormonal assessment of the patient

Age (years)	Bone age (years)	Basal LH (mIU/ml)	Basal FSH (mIU/ml)	Testosteron (ng/dl)	Peak LH (mIU/ml)	Peak FSH (mIU/ml)	
0,6	1	1,56	1,02	88	43,3*	3,65	Diagnosis
1.28	2	3.52	0.39	67	6**	0.54*	GnRH a (250 mcg/kg/month)
2,5	4,5	1,61	0,13	58	6,53**	0,53*	GnRH a (500 mcg/kg/month)
							GnRH a And cyproterone asetat

* 100 μg of GnRH (gonadorelin acetate, Ferring Standard). **with leuiprolid asetat stimulation

Genetic analysis of *NSD1* → **No deletion** → ***NSD1* heterozygous mutation: NM_022455.4:c.5177C>G (p.Pro1726Arg)**

Conclusion: **Central precocious puberty can be accompanied with Sotos syndrome, and overgrowth can be related either to syndrome itself, and precocious puberty. Treatment can also be very challenging with required high dose and combined treatment. Although we can not explain the reason of central precocious puberty in Sotos syndrome, it can be related to mutation characteristics of *NDS1*, or other underlying reasons that need to be demonstrated.**

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