Family Central Early Puberty about Three Sisters

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Introduction

Precocious puberty is defined in girls by the appearance of secondary sexual characteristics before the age of 8. Unlike the boys, the central origin is most often idiopathic. The familial nature encourages the search for a genetic mutation that can explain this early maturation of the gonadotropic axis.

Results

	Observation n°1	Observation n°2	Observation n°3
Age	5 years	6 years	5 years 3 months
Circumstances of discovery	Isolated mammal development		
Tanner Stage	S3P1A1	S3A1P1	S3A1P1
Size upon diagnosis	112.5cm (+1DS)	123cm (+2DS)	120cm (+3DS)
Hormonal balance	Oestradiol = 25pg/ml FSH = 3.53mU/ml LH =0.53 mU/ml Cortisol = 79.9 ng/ml ACTH=17pg/ml	Oestradiol = 15pg/ml FSH = 2.37mU/ml LH = 1.1 mU/ml Cortisol = 55 ng/ml ACTH = 6.7pg/ml Inhibine B = 29pg/ml	Oestradiol = 8pg/ml FSH=4.4mU/ml LH=0.28 mU/ml Cortisol = 186 ng/ml ACTH = 17pg/ml Inhibine B = 23pg/ml
Bone age	5 years	6 years	5 years
Pelvic Echography	an enlarged uterus (40mm * 18mm * 14mm) with multi follicular ovaries	the uterine height was 33.7mm with a development of multi-cystic ovaries.	l'hauteur utérine était à 33,7mm avec un développement des ovaires multi-kystiques.
Hypothalamic-pituitary MRI	Normal		
Treatment	Decapeptyl at a dose of 50µg / kg / month		
Tracking time / Size	10 months /129cm	2 years/136cm	2 years/129.5cm

Discussion

Central precocious puberty (PCP) is defined as the development of sexual characteristics before the age of 8 years in girls and before the age of 9 in boys, related to premature activation of the hypothalamic pituitary gonadic axis. PCP may be secondary to congenital or acquired brain damage. The majority of PCP in girls is idiopathic.

In the idiopathic forms are especially girls with a high BMI, 2 mutations of the GPR54 gene would be at the origin, with incomplete penetrance.

A study of familial forms of CPP suggests an autosomal dominant mode of transmission but with variable penetrance, lower in boys than in girls. Family forms deserve investigation because it is difficult to distinguish between environmental and genetic factors, both of which are involved in pubertal induction.

If the therapeutic management is currently well framed with regard to pubertal follow-up, it is necessary to pay full attention to the possible consequences of precocious puberty, in particular weight and body composition monitoring with incentive to physical activity and control of nutritional intake, monitoring of bone mineralization with the initiation of early calcium supplementation if necessary.

Long-term follow up should be proposed so as not to neglect the risk of ovarian functional hyperandrogenism related to the development of a polycystic ovary syndrome

CONCLUSION

Except isolated observations showing an activating mutation of the GPR54 gene described in patients with PCP, no gene is involved to date in idiopathic PCP. Indeed, despite the identification of several genes (IAP, TTF1, Nell12, FGF1Rc) regulating the synthesis or secretion of GnRH, the physiopathology of this premature reactivation remains hypothetical

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