An uncommon case of adolescent with POF

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The authors are employees of Civico Hospital. They declare that no other conflict of interest exists.

Introduction

Premature ovarian failure (POF) is uncommon in pediatrics and when occurs during the adolescence is typically iatrogenic or due to chromosomal anomaly. Also many genes have been identified that contribute to the development of POF, and most of these mutations are extremely rare.

Case report

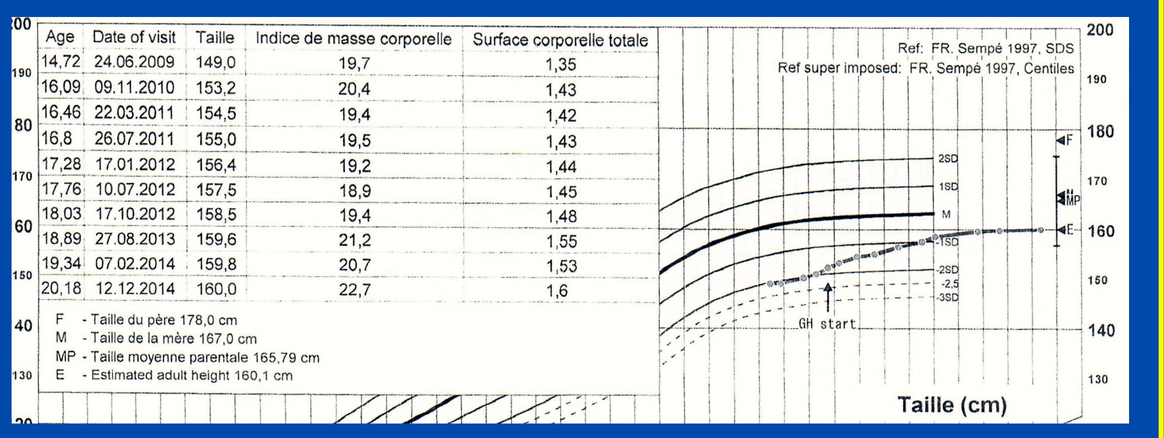
We describe a case of 15 years old female presented short stature and secondary amenorrhea, after a normal puberty but without peak height velocity.
She was well being with no history of illness, drugs or radiation and no family history of ovarian insufficiency.
Clinically besides short stature (-2 sds), we highlighted an involution of secondary sexual characteristics with very small mammary gland (4 cm).

Biochemical investigation revealed eleveted gonadotropins (LH 65 U/l, FSH 185 U/l) confirmed by LHRH test that allowed to diagnose an hypergonadotropic hypogonadism.
Kariotype was normal and adrenal and ovaries antibodies were negative.

	T 0’	T 15’	T 30’	T 60’	T 90’	T 120’
FSH (U/L)	145,3	> 200	> 200	> 200	> 200	> 200
LH (U/L)	52,5	233	> 250	235,5	169	144

Pelvic imaging by tras-abdominal ultrasound and MRI showed a very small ovaries and uterus. Nevertheless the last abdominal MRI failed to identify the ovaries.

We started with estrogen replacement therapy, until the dose of 10 μg/die. She had regular mestrual bleending and an adult development of uterus and mammary gland.
She received a GH replacement therapy to improve the final height and she catch her target height.



Conclusion

Probably a genetic etiology, maybe a gonadotropic receptor dysfunction, is implicated but the certain cause still remain not known.

References:
•Gowri V, Al Shukri M, Al-Farsi FA, Al-Busaidi NA, Dennison D, Al Kindi S, Daar S, Al Farsi K, Pathare AV. Aetiological profile of women presenting with premature ovarian failure to a single tertiary care center in Oman. Post Reprod Health. 2015 Jun;21(2): 63-8.
•Brauner R, Pierrepont S, Bignon-Topalovic J, McElreavey K, Bashamboo A. Etiology of primary ovarian insufficiency in a series young girls presenting at a pediatric endocrinology center. Eur J Pediatr. 2015 Jun;174(6):767-73.
•Rege G, Foidart JM, Nisolle M, Pintiaux A, Béliard A. [Premature ovarian failure, from genetics to clinical]. Rev Med Liege. 2012 Jul-Aug;67(7-8):413-9.
•Ebrahimi M, Akbari Asbagh F. Pathogenesis and causes of premature ovarian failure: an update. Int J Fertil Steril. 2011 Jul;5(2): 54-65.

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