Case Report

DOI: 10.5455/2349-3291.ijcp20140516

Development of secondary sexual characters in a case of idiopathic hypogonadotropic hypogonadism

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Received: 26 April 2014 Accepted: 9 May 2014

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ABSTRACT

We report a rare case of idiopathic hypogonadotropic hypogonadism in a 15 year old adolescent girl, who presented in gynecology outpatient clinic with complaint of primary amenorrhea and poor development of secondary sexual characters. Her clinical examination revealed small, underdeveloped breasts (Tanner stage II), absent axillary hair and sparse pubic hair (Tanner stage II). Ultrasound pelvis showed uterus of 4.76 x 2.29 x 1.25 centimeter, small and hypoplastic with very thin endometrial lining, bilateral ovaries were small with 1-2 small follicles. Magnetic Resonance Imaging (MRI) of the hypothalamic-pituitary unit showed no abnormality. Further workup was done to detect other causes of hypogonadism, which was normal, hence we concluded the patient to have idiopathic hypogonadotropic hypogonadism.

Keywords: Primary amenorrhea, Idiopathic hypogonadotrophic hypogonadism

INTRODUCTION

Amenorrhea is the absence or abnormal cessation of the menstruation. Primary and secondary amenorrhea describe the occurrence of amenorrhea before and after menarche, respectively. The prevalence of amenorrhea not due to pregnancy, lactation or menopause is approximately 3% to 4%. 2,3

Evaluation of primary amenorrhea is indicated when there is failure to menstruate by age 15 years in the presence of normal secondary sexual characteristics or failure to initiate breast development by age 13.⁴ The prevalence of IHH has been estimated at between 1 in 4000 and 1 in 10000 males. It is reported to be between two and five times less frequent in females.⁵ Idiopathic hypo gonadotropic hypogonadism is caused by an isolated defect in Gonadotropin-Releasing Hormone (GnRH) release, action, or both.⁶ Other associated non

reproductive phenotypes, such as anosmia, cleft palate, and sensorineural hearing loss, occur with variable frequency. In the presence of anosmia, idiopathic hypogonadotropic hypogonadism is classified as the Kallmann syndrome, whereas in the presence of a normal sense of smell, it is termed normosmic idiopathic hypogonadotropic hypogonadism.⁷

We would like to report this case as; this is an uncommon cause for hypogonadism and infertility especially in females and represents one of the few treatable forms of hypogonadism.

CASE REPORT

A 15 year old adolescent girl, presented in gynecology outpatient clinic with complaint of not attaining menarche and poor development of secondary sexual characteristics. She also complained of generalized

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weakness & malaise for past 5 months. A detailed history was elicited regarding any history of drug intake, history of cyclical pain abdomen, headache, difficulty in vision, or difficulty to smell. There was no history suggestive of eating disorders, excessive physical activity, delay in milestones and chronic underlying conditions. Her family history was also not significant. Her mother attained menarche at 12 years of age and her younger sister attained menarche at 11 years with normal secondary sexual characters.

A detailed clinical examination was done. Her vitals were within normal limits (pulse rate: 70 beats/minute, blood pressure: 110/70 mmHg and respiratory rate: 16 cycles/minute), height: 158 centimeters, weight: 44 kilograms and Body Mass Index: 17.6 kg/ m². Upper body segment: 68 cm, lower body segment: 90 cm and her arm span was 168 cm. There was no pallor, no hirsutism or acne, no classic features of Turner syndrome. Her secondary sexual characters were noted. Breast development (grading as per the Tanner' scale of sexual maturation) was Tanner stage II, no axillary hair and pubic hair (grading as per Tanners scale of sexual maturation) was Tanner stage II. Per abdomen was soft, non-tender, no mass and, no organomegaly. External genitalia were female type, no clitoromegaly and hymenal opening was present. The length of vagina was approximately 8-9 centimeter as measured with red rubber catheter. Per Rectal examination showed a small and hypoplastic uterus.

Routine investigations such as hemoglobin, total leucocyte count, differential leukocyte count, random blood sugar, erythrocyte sedimentation rate, liver function test, renal function test, peripheral smear and coagulation profile were done which were found within normal limit. Follicle stimulating hormone (9.9 mIU/ml), luteinizing hormone (4.5 mIU/ml), thyroid stimulating hormone (2.9), serum prolactin (14.1 ng/dl) and cortisol (21.0 µgm/dl) were normal. Ultrasound pelvis showed uterus measuring 4.76 x 2.29 x 1.25 centimeter, small and hypoplastic, endometrial lining was very thin and both ovaries normal with few small follicles. MRI of brain & pituitary did not reveal any abnormality. It was concluded that primary amenorrhea and poor development of secondary sexual characters was most likely due to hypogonadotrophic hypogonadism which is commonly due to systemic illness or idiopathic.

Conjugated estrogen (0.625 mg/day) was started and continued for six months till breakthrough bleeding occurred. Then cyclical estrogen (0.625 mg x 21 days) and progesterone (5 mg x last 10 days) was given .The treatment was continued for one year till adult maturity was attained. At the end of one and a half year of treatment clinical examination revealed improvement in breast development (Tanner stage 4) and pubic hair (Tanner stage 4) with few axillary hairs. Ultrasonography on 9/2/2013 showed uterus: 5.2 x 2.1 x 2.9 centimeter, endometrial lining: 5 millimeter, left ovary: 2.3 x 1.5x 1.5

mm, right ovary: 2.5 x 1.2 x 1.2 mm. Since patient had reached adult maturity with optimal secondary sexual characters she was shifted to cyclical oral contraceptive pills.

DISCUSSION

Hypogonadism is defined as "inadequate gonadal function, as manifested by deficiencies in gametogenesis and/or the secretion of gonadal hormones". The major objectives of the initial assessment of a patient with possible hypogonadism are to distinguish primary gonadal failure (hypergonadotropic hypogonadism with low estradiol and increased FSH and LH levels) from hypothalamic-pituitary disorders (hypogonadotropic hypogonadism with low estradiol and low to normal FSH and LH levels) and to make a specific diagnosis.

In hypogonadotropic amenorrhea there is poor follicular development as the pulsatile secretion of gonadotropins is suppressed. This type of amenorrhea is often associated with, constitutional delay, stress, eating disorders, excessive physical activity etc. The preliminary step in the diagnosis is a detailed history and physical examination with the assessment of the development of secondary sexual characters. Adult females have little or no development of secondary sexual characters, although in some patients it may be almost normal. Plasma LH, FSH and estradiol concentrations are often low in such women. Kumar A and Mittal S showed that out of 48 cases of primary amenorrhea, 54% had Mullerian anomalies, 23% had hypogonadotropic hypogonadism, 17% had hyper gonadotropic hypogonadism.

CONCLUSION

The diagnosis of IHH is established when there is presence of clinical features and laboratory findings consistent with hypogonadotropic hypogonadism, along with the absence of secondary causes of hypothalamic hypogonadism. The choice of therapy is determined by the goal of treatment. There is paucity of long-term research studies on the identification of women at risk for complications related to a decreased estradiol levels. In order to better describe the pathogenesis of these rare cases, further more research is needed.

Funding: No funding sources Conflict of interest: None declared Ethical approval: Not required

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DOI: 10.5455/2349-3291.ijcp20140516 **Cite this article as:** Singh N, Mathew A, Chauhan RC, Parvathavarthini. Development of secondary sexual characters in a case of idiopathic hypogonadotropic hypogonadism. Int J Contemp Pediatr 2014;1:55-7.