

Primary amenorrhoea

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Abstract

Primary amenorrhoea is defined as the absence of menstruation by age 14 years in the absence of secondary sexual characteristics, or by age 16 years if secondary sexual characteristics are present. Regular menstruation is regarded as a sign of reproductive health thus absent menstruation causes a significant degree of stress and anxiety to the individuals involved. Puberty is governed by a complex neuro-hormonal system with input from multiple endogenous and exogenous factors, and pathology at various levels in this system can result in amenorrhoea. The causes range from genetic, anatomical, and endocrine to constitutional delay. A stepwise diagnostic approach is key to guiding appropriate investigations whilst minimising the physical and psychological impact on young adolescent women.

Keywords adolescent; adrenarche; amenorrhoea; gonadotrophins-releasing hormone; menarche; menstruation; menstruation disturbances; puberty; reproductive health; sexual maturation

Introduction

Amenorrhoea, the absence of menses, can be transient or permanent and is a symptom that reflects potential dysfunction in the hypothalamic-pituitary-ovarian axis or anatomical anomalies in the uterus or outflow tract. Amenorrhoea can be classified as either primary or secondary depending on the presence (secondary) or absence (primary) of previous menses. Primary amenorrhoea, the focus of this article, is defined as:

- the absence of menses by age 14 years in the absence of secondary sexual characteristics or
- the absence of menses by age 16 years in the presence of normal secondary sexual characteristics.

Primary amenorrhoea is rare with <5% of adolescent girls presenting to gynaecological services. Whilst the average gynaecologist will see only a few cases, the importance of sensitively managing the initial presentation and diagnostic work up during the vulnerable adolescent years requires all gynaecologists to have a good understanding of the subject. (This article should be read in conjunction with the previous review published in this journal: Basak S, Prakash A. Investigation and treatment of primary amenorrhoea. *Obstet Gynaecol Reprod Med* 2013; **23**: 364–369.)

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Puberty

Puberty is the transition to sexual maturity, with the most visible changes being the growth spurt and development of secondary sexual characteristics. It proceeds in a predictable fashion and is staged using the Tanner system (Table 1). The average age at onset of puberty is 11 years but this is influenced by many factors including genetics, general health and BMI. In most girls the earliest sign of puberty is breast development, although 15% may have pubic hair as the initial sign. Menarche occurs around 2 years after the onset of puberty.

The primary driver for puberty is the onset of pulsatile gonadotrophins-releasing hormone (GnRH) secretion from the hypothalamus (see Figure 1). The increasing frequency and amplitude of these pulses stimulate the release of luteinizing hormone (LH) and follicle-stimulating hormone (FSH) from the anterior pituitary gland. These hormones then stimulate the production of estradiol by the ovaries. In the beginning stages of puberty, estradiol stimulates breast development (thelarche) and the skeletal growth spurt. In the later stages, the specific combination of FSH, LH and estradiol leads to ovulation and the menstrual cycle begins (menarche). Around the time of the onset of this increase in GnRH pulsatile secretion, Adrenarche, a physiologically distinct event, begins. Dehydroepiandrosterone (DHEA) and androstenedione are released from the adrenal gland, leading to growth of pubic and axillary hair, maturation of apocrine sweat glands and acne. Thus, an individual with a defect in the hypothalamic-pituitary-ovarian axis can still develop pubic and axillary hair.

Causes of primary amenorrhoea

Disruption at any level of the pathway will result in primary amenorrhoea. The causes can be classified by the presence or absence of secondary sexual characteristics and the level at which the pathology exists. Constitutional delay of puberty accounted for 14% of attendances in one large case series; it is more common in boys than girls. A thorough history and examination will help to guide the most appropriate investigations. Almost all causes of secondary amenorrhoea can also cause primary amenorrhoea but they are not discussed in this article. A suggested diagnostic flow chart (Figure 2) and table of differential diagnoses (Table 2) provides guidance for day to day management of such cases.

Tanner stages of breast and pubic hair development

Tanner stage	Breast development	Pubic hair development
Stage 1	Prepubertal	Prepubertal
Stage 2	Breast bud	Sparse growth along labia
Stage 3	Further enlargement of breast and areola	Darker, coarser hair.
Stage 4	Areola and papilla form a secondary mound	Spreads to mons. Hair adult type
Stage 5	Mature stage	Hair spreads to thighs

Table 1

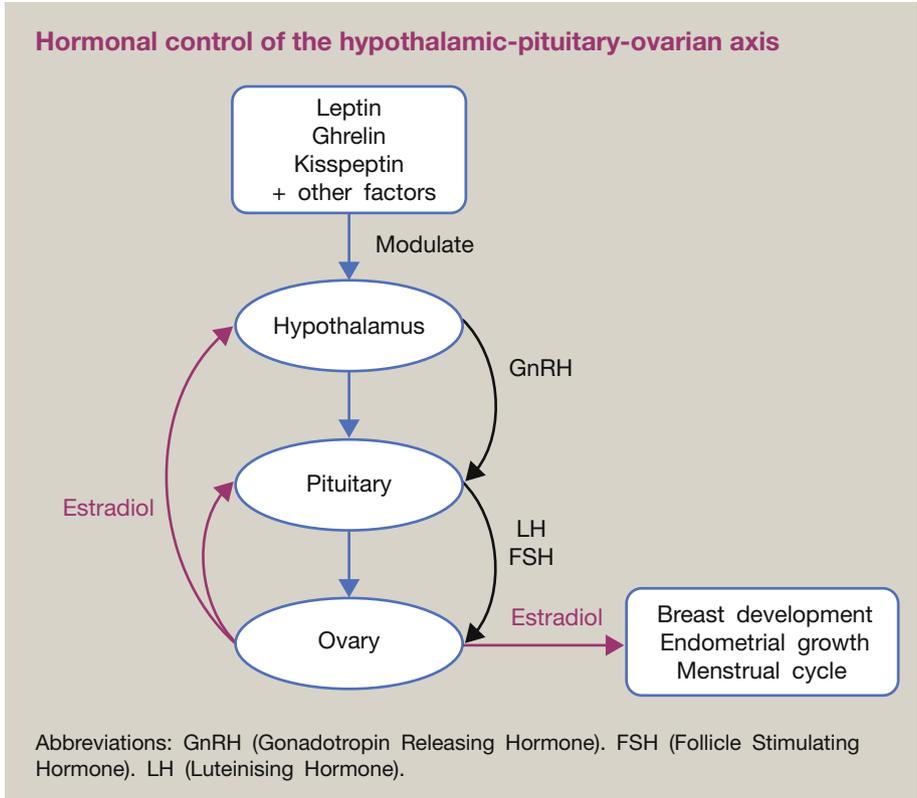


Figure 1

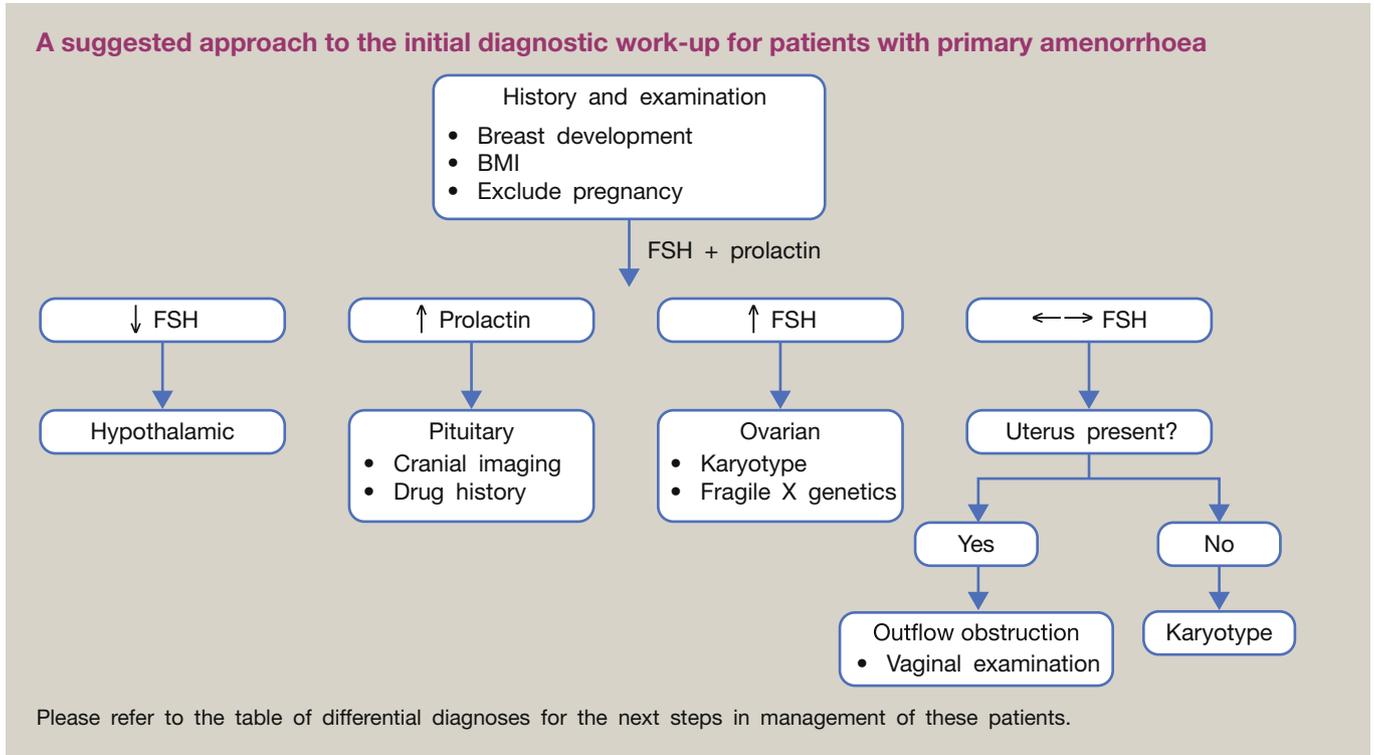


Figure 2

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