

# Diagnostic spectrum of female pubertal delay

## Introduction

Delayed onset of puberty is quite a common presentation in adolescent endocrine clinics, and the most common cause, particularly in boys is considered to be constitutional delay of growth and maturation. In girls, however, it is more likely that there is a significant underlying problem.

## Aim

To review the aetiology of pubertal delay in female patients referred to a single tertiary centre.

## Methodology

All female patients referred to the endocrinology clinic with delayed puberty, arrested puberty and primary amenorrhoea between January 2007 and December 2012 were identified using our clinic patient database.

A review of medical case notes was carried out to identify the aetiology of pubertal delay, and information was also obtained on investigations, treatment and outcome. Patients with known conditions associated with pubertal delay, pituitary/gonadotoxic therapy or secondary amenorrhoea were excluded.

## Results

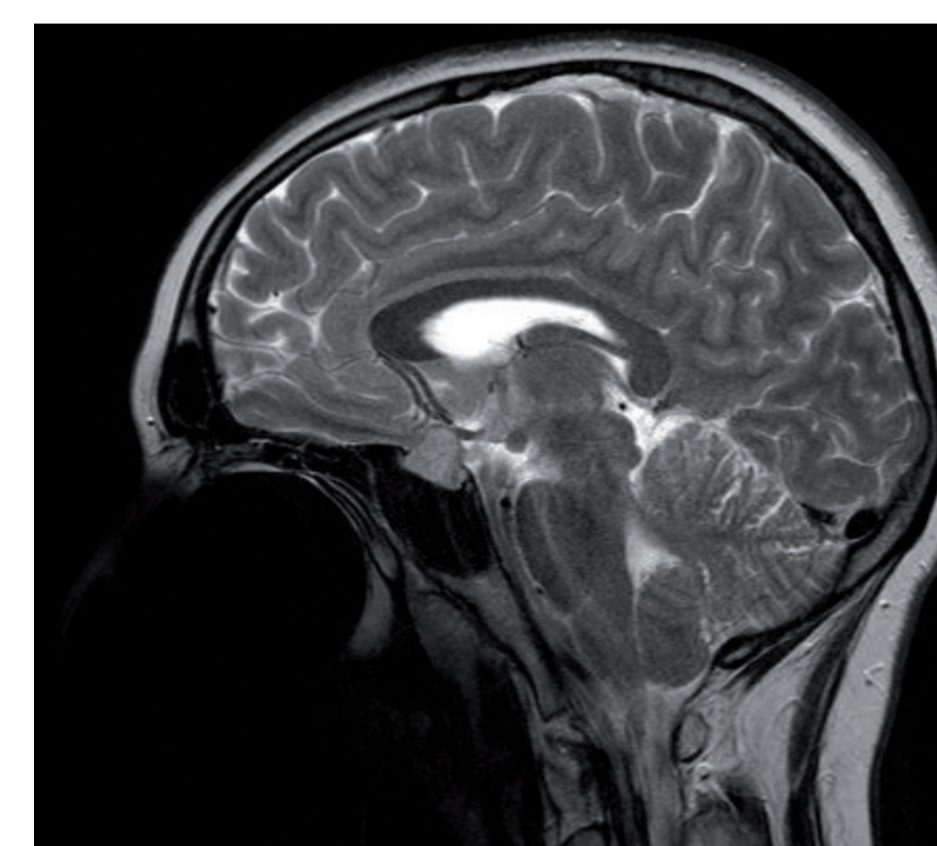
Thirty three patients were identified with a median age of presentation of 15.5 years. A total of fourteen different reasons for pubertal delay were found in our population.

The three most common causes were low BMI, constitutional delay (no abnormality found and no therapeutic intervention required for onset of menses) and idiopathic primary ovarian failure, but intracranial lesions (craniopharyngioma, prolactinoma), structural abnormalities of the genital tract (Mayer-Rokitansky syndrome) and genetic/chromosomal anomalies (Androgen Insensitivity Syndrome, Turner Mosaic) were all identified.

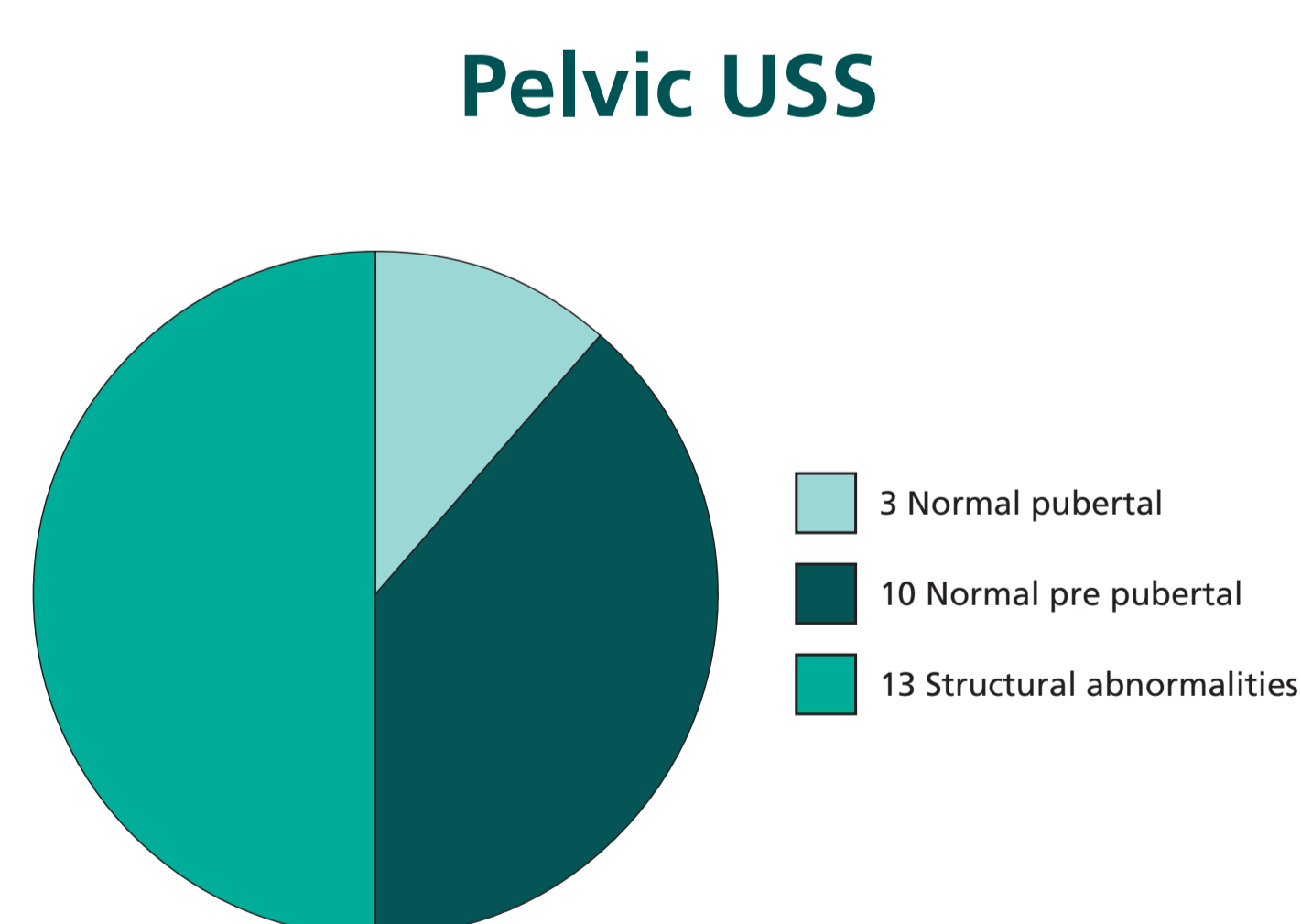
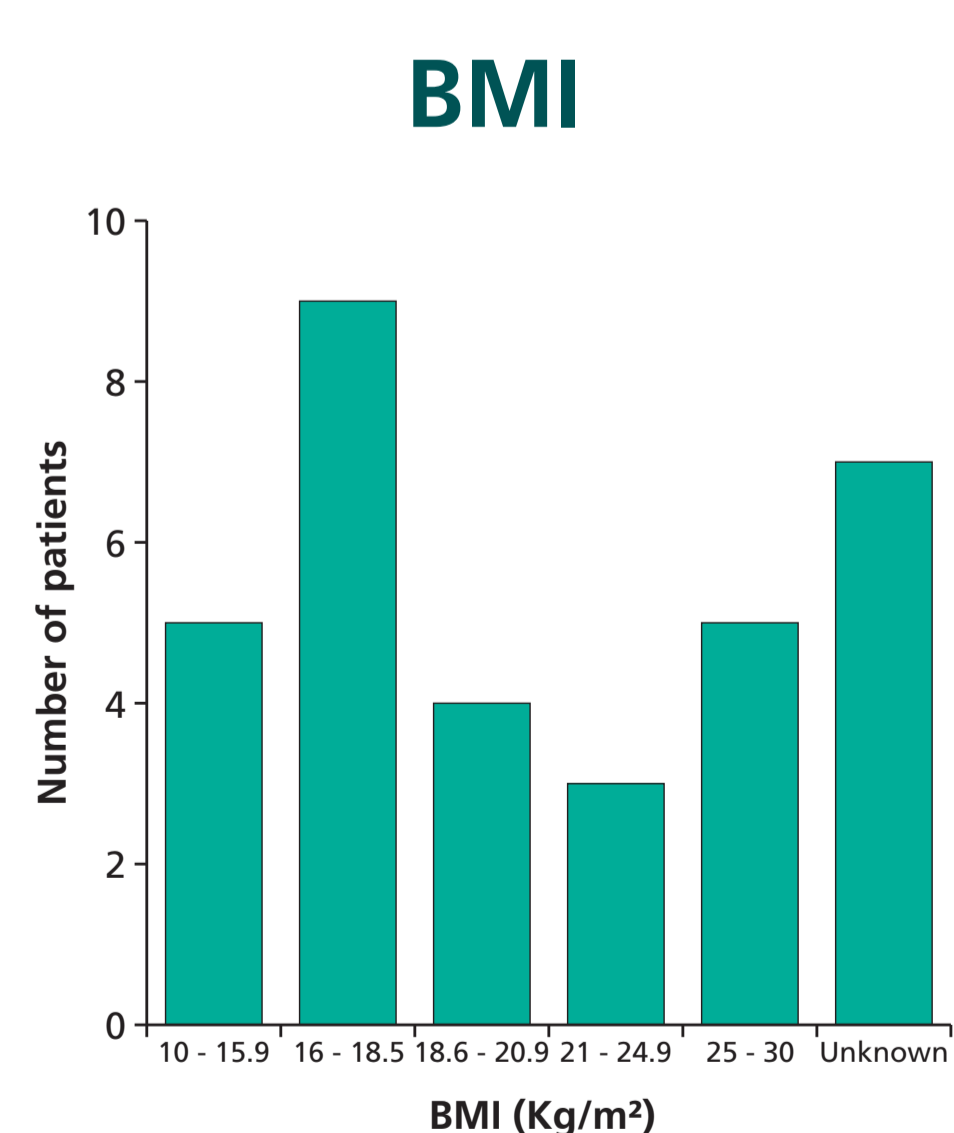
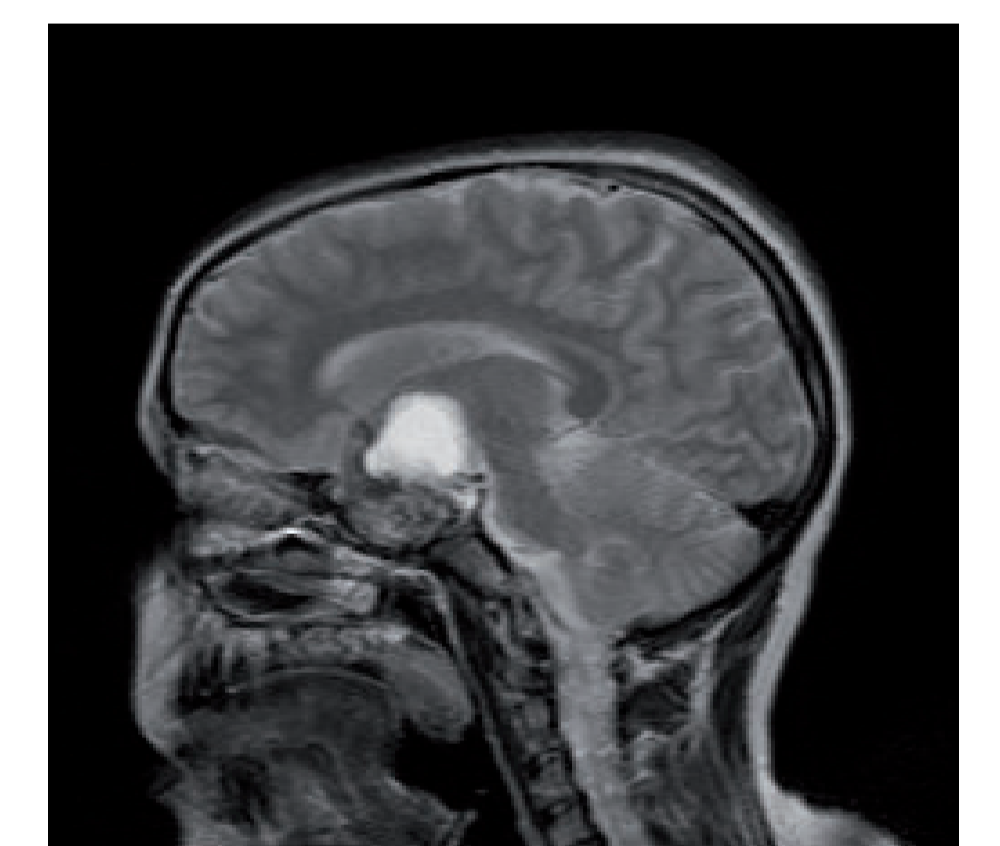
Karyotype	Number of patients
Normal	22 patients
46XY : Androgen insensitivity syndrome	1 patient
46XY : SF1 mutation- see poster No: 51	1 patient
46xt (x4) (q13q31.3) :Ovarian dysgenesis	1 patient
45X in 13/30 cells and 46XY SRY + ve in 17/30 cells : Turner mosaic	1 patient
45X in 36/38 and 46XiX (q10) in 2/38 : Turner mosaic	1 patient
Karyotype not done in	6 patients

Diagnosis	Number of patients
Low BMI (secondary to eating disorder or athletic training)	7
Constitutional delay	6
Primary ovarian failure (under investigation)	5
Primary ovarian failure secondary to tubo-ovariectomy for haemorrhagic cyst of the ovary	1
Craniopharyngioma	2
Mosaic Turner syndrome	2
Mayer Rokitansky Kuster Hauser syndrome	2
Prolactinoma	1
Hyperprolactinaemia	1
Idiopathic hypogonadotrophic hypogonadism	1
Ovarian dysgenesis secondary to balanced translocation	1
Androgen insensitivity syndrome	1
46XY DSD secondary to SF1 mutation	1
High BMI	1
No diagnosis yet	1

### Craniopharyngioma

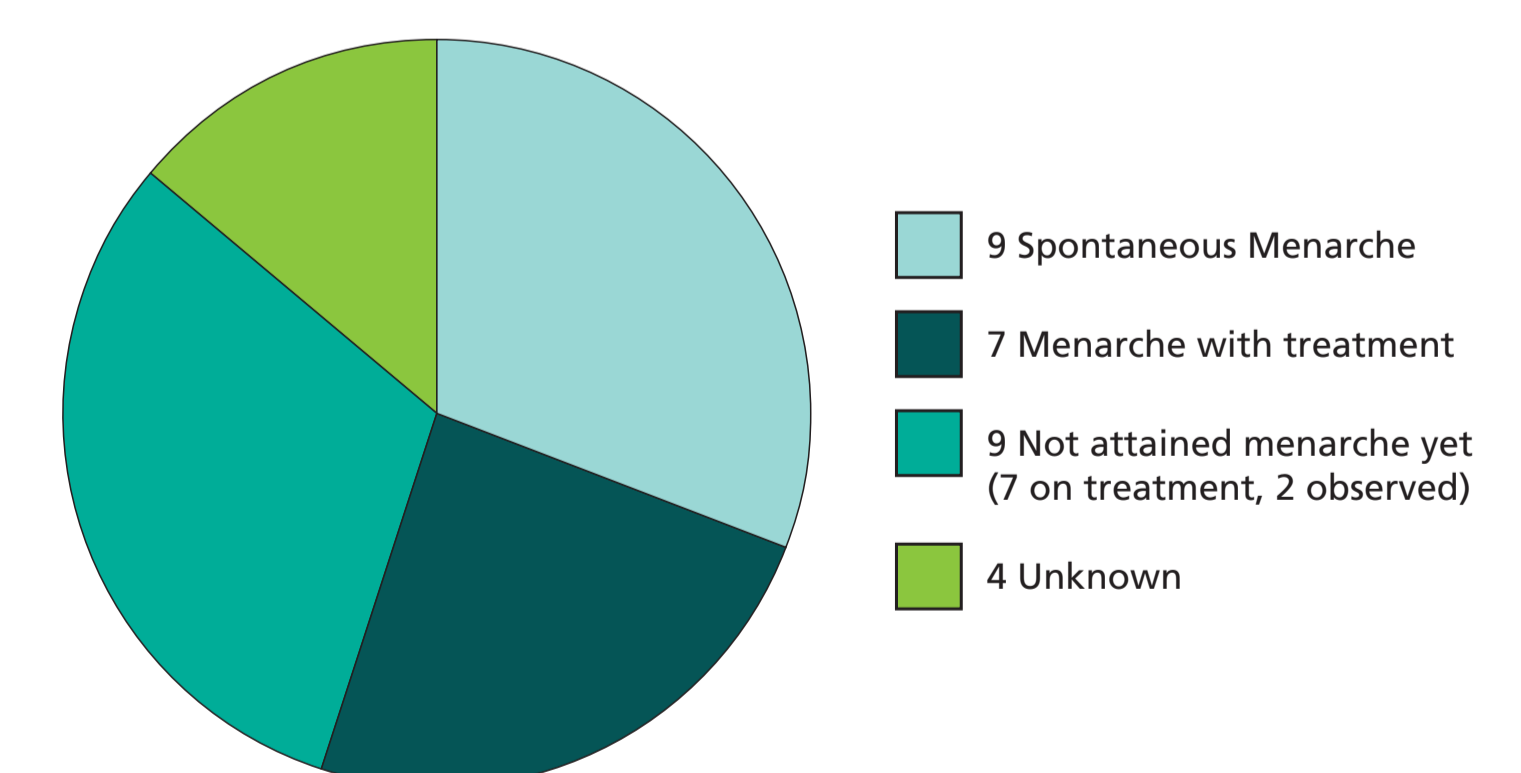


### Prolactinoma



## Outcome

Of the 29 patients who were expected to attain menarche, 30% did so spontaneously and 24% required ethinyloestradiol treatment. A further 24% are on treatment and being monitored.



## Conclusion

Although simple maturational delay can be a common cause of delayed puberty, in our study we found that a large number (88%) of our patients had a significant underlying aetiology. Seven girls (21%) had a marked eating disorder or other reason for a very low BMI. These results confirm the importance of thorough evaluation of all girls presenting with delayed puberty.

