CASE FIVE

Short case number: 3_24_5

Category: Endocrine and Reproductive Systems
Discipline: Obstetrics & Gynaecology
Setting: General Practice
Topic: Primary Amenorrhoea [SDL]

Case

Ellie Brown, presents to your GP surgery with her mother stating that she has not had her period. On history, you find that Ellie is 14 years of age and has never had a period. She had breast bud development at the age of 11 years of age, followed by pubic hair growth, axillary hair and a growth spurt. Her mother is concerned that things seemed to have stopped since then. She is otherwise healthy (playing quite a bit of sport), has no significant medical history and is not on any medication.

Questions

1. Discuss the usefulness or otherwise of using an age cut-off for primary amenorrhoea and determine (and explain why or why not) if you need to investigate Ellie's complaint.
2. Summarise the 5 important changes that females undergo at puberty.
3. Discuss the aetiology of primary amenorrhoea using the 3 broad classifications of secondary sexual characteristics (present or absent) and height (normal or short).
4. In a flow chart, describe the investigations required to determine if this is, or is not, constitutional delay.
5. Summarise the management of women who have normal secondary sexual characteristics with primary amenorrhoea.
6. Summarise the management of women who have absent secondary sexual characteristics with primary amenorrhoea

Suggested Reading

- Abbott, J., Bowyer, L., & Finn, M. (2014). *Obstetrics and Gynaecology: an evidence-based guide (2nd ed).* Australia, Elsevier
- Dewhurst's Textbook of Obstetrics & Gynaecology, Edmonds K [editor]. Blackwell Publishing. 2007. Chapter 38

Discuss the usefulness or otherwise of using an age cut-off for primary amenorrhoea and determine (and explain why or why not) if you need to investigate Ellie's complaint.

Failure of development of any secondary sexual characteristics by age 14 = Investigate

Menstruation should occur within 2 years of the development of secondary sexual charachteristics (i.e. no later than 16)

If 16 and no menses yet - can investigate.

Most often just a consititutional delay and completely normal.

Summarise the 5 important changes that females undergo at puberty.

1. Breast Development
2. Pubic Hair
3. Axillary (Underarm) Hair
4. Growth Spurt
5. Menstruation Begins

+

Hips Widen

Fat distributed to hips & Thighs

Development of Reproductive organs (Uterus, Ovaries, labia Minora grow)

Discuss the aetiology/cause of primary amenorrhoea using the 3 broad classifications of secondary sexual characteristics (present or absent) and height (normal or short).

Secondary Sexual Characteristics Present (Normal Height)

- Imperforate Hymen (Outflow Tract Obstruction)
- Transverse Vaginal Septum (Outflow Tract Obstruction)
- Absent Vagina and functioning uterus
- Constitutional Delay
- Resistant ovary Syndrome

Secondary Sexual Characteristic Absent (Normal Height)

- Hypogonadotrophic Hypogonadism
- Anorexia or Excessive Excercise
- Hyperprolactinoma
- Turner Mosaic

Secondary Sexual Characteristics Absent (Short Stature)

- Hypogonadotropic Hypogonadism
- Congeital Hydrocephalus
- Trauma
- Malignancy
- Turner Syndrome

In a flow chart, describe the investigations required to determine if this is, or is not, constitutional delay.

See Tutor HB answer

Summarise the management of women who have normal secondary sexual characteristics with primary amenorrhoea.

#1 DDx should be some form out outflow tract obstruction

- PVE
- TVUS

Summarise the management of women who have absent secondary sexual characteristics with primary amenorrhoea

#1 - Measure patients height

Normal Height
Gonadotrophin levels
(if low = Hypogonadotrophin hypogonadism)
(if High -> do Karyotype testing)

Short Stature - Do other tests, it didn't say. But probs still do gonadotrophin levels.

ANSWERS

Question 1

For the majority of pubertal girls menstruation is the final result of a series of events which result in sexual maturity. Maturation of the hypothalamus through several years of late childhood begins a cascade of events which finally result in the establishment of the normal menstrual cycle and menstruation. Amenorrhoea will result when there is a failure of function in any of the organs involved in this cascade.
It is useful to look upon secondary sexual development as the criteria for investigation and management of primary amenorrhoea. As a general rule, failure of the development of any secondary sexual characteristics by age 14 should be investigated. In the presence of secondary sexual characteristics, menstruation ought to occur within 2 years of the establishment of this development (i.e. by 16 years of age). Therefore, a girl who has not had periods by 16 years of age should be investigated. Commonest cause in practice is constitutional delay rather than an anatomical or genetic abnormality. Often one just has to allay mother and daughters fears and reassure them of "normality"

Question 2

There are five changes that occur at puberty, known as the secondary sexual characteristics. In girls these are breast, pubic hair, axillary hair development, the growth spurt and the onset of menstruation. Ovulation may not occur for up to 5 years after menarche, therefore girls may have irregular periods during this time.

Question 3

Aetiology of Primary Amenorrhoea.

Secondary sexual characteristics normal

- Imperforate hymen: (often present with mass rising out of pelvis-blood filled uterus and blood distended vagina: haematometra and haematocolpos. Bulging blue hymen visible on parting labia. May have had cyclical pain.)
- Transverse vaginal septum
- Absent vagina and functioning uterus
- Constitutional delay
- Resistant ovary syndrome

Secondary Sexual characteristics *absent and normal stature*

- Hypogonadotrophic hypogonadism
- Weight loss/ anorexia
- Excessive exercise
- Hyperprolactinemia
- Hypergonadotrophic hypogonadism
- XX agenesis/ Gonadal dysgenesis
- Galcatosaemia
- Turner Mosiac

Secondary sexual characteristics *absent but short stature*

- Hypogonadotrophic hypogonadism
- Congenital hydrocephalus
- Trauma
- Empty sella syndrome
- Tumours
- Turners syndrome
- Other X deletions or mosaics

Question 4

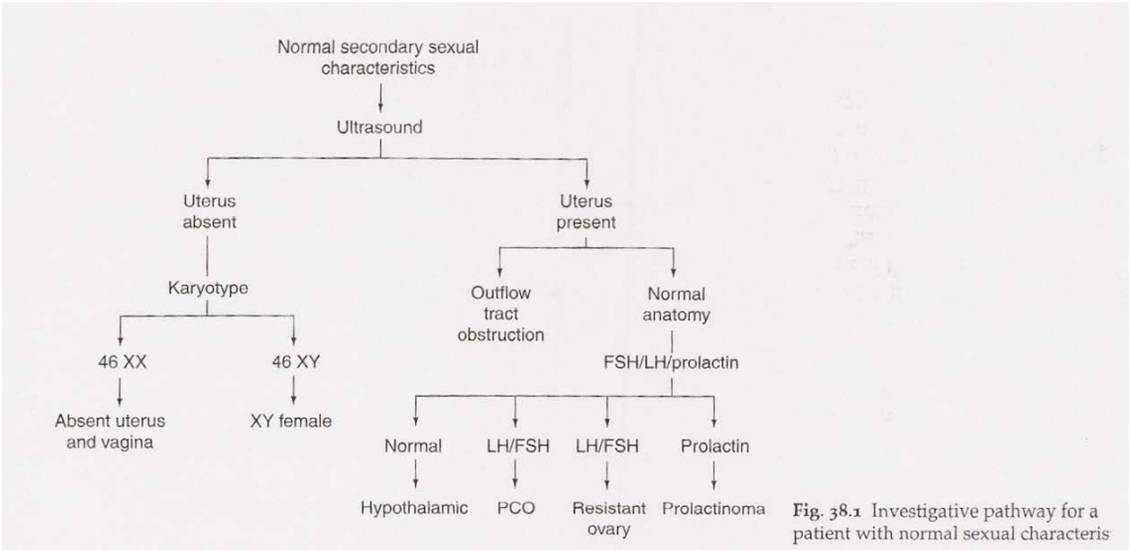

Fig. 38.1 Investigative pathway for a patient with normal sexual characteris

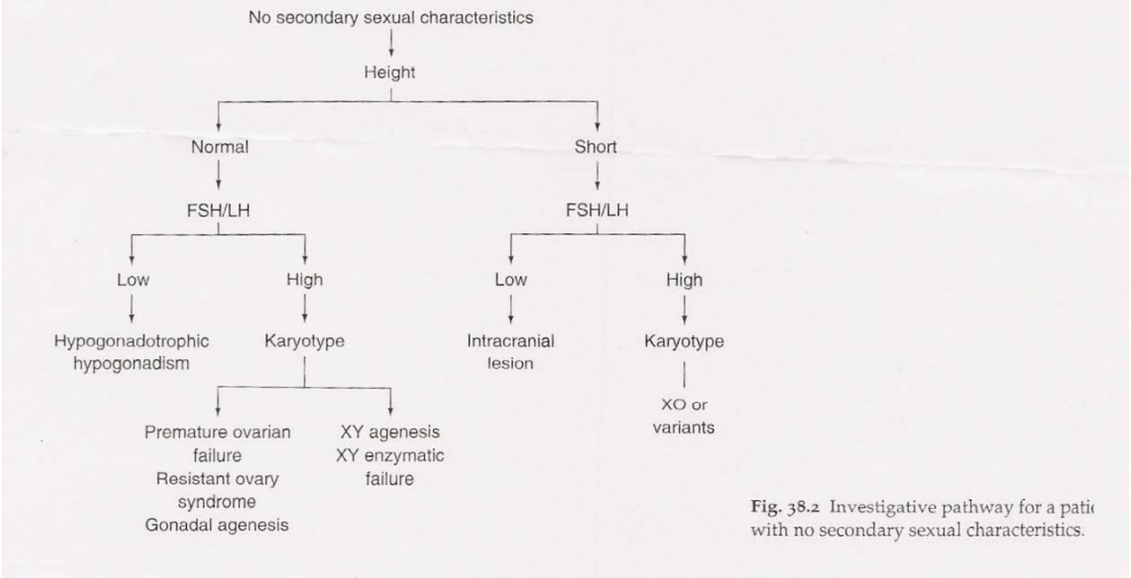

Fig. 38.2 Investigative pathway for a pati with no secondary sexual characteristics.

Question 5

The presence of normal secondary sexual characteristics should alert the clinician to the concept that outflow tract obstruction may be occurring. This is the most common cause of primary amenorrhoea in the presence of normal secondary sexual characteristics. It is thus appropriate to carry out investigations to make this diagnosis. It is inappropriate to perform any physical pelvic examination on these young adolescents. It is simply to arrange for a pelvic ultrasound to assess pelvic anatomy.

Question 6

When there is an absence of secondary sexual characteristics it is extremely important to make an assessment of the patient's height. If the patient is of normal height for age, measurement of gonadotrophin will reveal levels that are either low or high. Low levels confirm a diagnosis of hypogonadotrophin hypogonadism, and elevated levels should provoke the clinician to perform a karyotype.