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Case 1

You are a medical student on your General Practice (GP) placement. Lily is a 30-year-old female who presents to the practice with difficulty falling pregnant. Lily and her partner James have been having unprotected sexual intercourse for 2 years and have not yet fallen pregnant.

**Broadly, what are some differential diagnoses? Think about specific areas that might be affected (specific conditions are not as important for this question).**

Being the thorough Griffith medical student that you are, you take a detailed history from Lily.

Lily reports that she had her first period when she was 13 and that she never had normal periods. Due to the irregularity of her periods, she began taking the combined oral contraceptive pill when she was 16. She took this regularly, until 2 years ago, when her and James started trying to fall pregnant. Her last menstrual period was 10 weeks ago. Over the past two years, she reports having a total of 12 menstrual periods (approximately 6 per year). She has no other medical history, takes no regular medications and has no known allergies.

**How long is a normal menstrual cycle? What is the normal menstrual blood loss? In what way is Lily’s menstrual cycle abnormal?**

**Define the following terms.**

|  |  |
| --- | --- |
| Amenorrhoea |  |
| Dysmenorrhoea |  |
| Menorrhagia |  |

**What is the difference between primary amenorrhoea and secondary amenorrhoea? What are common causes of each? Which does Lily have?**

On examination, you note the following findings:

● BMI 31

● Acne

● Androgenic alopecia

● Hirsutism

● Acanthosis nigricans

**What is hirsutism?**

**In light of the above findings, what is the most likely diagnosis?**

**What are the three main overarching features of this condition?**

**Complete the table:**

|  |  |  |
| --- | --- | --- |
| **Type of cell:** | **Cells responds to which gonadotropin?** | **Function of these cells** |
| ***Theca cells*** |  |  |
| ***Granulosa cells*** |  |  |

*It is important to note that the pathophysiology of this disease isn’t completely understood, and the site of the primary defect is unclear.*

**There are some key changes in the levels of circulating gonadotropins. Explain what the change is and why it occurs?**

**What happens to the levels of testosterone and oestrogen in the blood?**

**Explain how the pathophysiology of this disease leads to each of Lily’s symptoms:**

|  |  |
| --- | --- |
| **Obesity** |  |
| **Acanthosis Nigricans** |  |
| **Anovulation** |  |
| **Virilisation** |  |

**What investigations would you like to order? Discuss why you are ordering them and how they help rule in/rule out diagnoses.**

**What might you think if Lily...**

|  |  |
| --- | --- |
| ...was an Olympic athlete? |  |
| ...suffered anorexia nervosa but is currently in remission? |

Given that Lily and James are trying to fall pregnant, the GP prescribe ***clomiphene***. You look up this mediation in the AMH and you find that it “inhibits hypothalamic oestrogen receptors”.

**Explain how clomiphene may provide benefit to Lily in falling pregnant.**

Ten months later, you are on your ED Placement at Logan Hospital. Lily presents with vaginal bleeding and abdominal pain. The pain came on suddenly while exercising and is located in the right lower quadrant. She states that she now has had regular 28 days menstrual cycles with 4–5-day periods. However, her last normal menstrual period was 6 weeks ago.

**At this stage, what are some of your differential diagnoses?**

**What key test do you want to order in this scenario?**

**Are there any other tests that you would like to order? Discuss why you might want to order these investigations.**

The above tests you ordered confirm that Lily has a viable intrauterine pregnancy but has a collection of fluid in the Pouch of Douglas (yes - it’s seen outside of the anatomy lab!).

**What is the most likely diagnosis now?**

Now that she is pregnant, Lily has some questions about what tests she needs to have during pregnancy. Among other things, you mention some screening and diagnostic tests.

**What does the combined first trimester screening (cFTS) test involve? What does it look for?**

**If the cFTS is positive, what is your next step?**

**What are the three viable autosomal trisomies?**

**What is non-invasive prenatal testing (NIPT)?**

**Please provide feedback for this case at:** [https://forms.gle/R64a83Cf7UgRYc168](https://forms.gle/R64a83Cf7UgRYc168?fbclid=IwAR2IJMsL2N2nkuJ3T7Dh-1XAPZthHc-uun2qlesNUR0AaT4lPut1r5BKg6E)