

Student Instructions

Jen Ettix is a 3 year old girl who has presented with her mother, Maria. Maria is concerned that Jen is becoming increasingly lethargic and tired.

Please take a history from Jen and Maria, including a social history, and identify the cause of her presentation. You will then be asked to discuss the case with the examiner.

Patient Instructions

You are Maria, Jen's mother. Jen, your 3 year old girl means the world to you. You have been very busy with work over the past year or so, so do not feel that you have got to spend nearly as much time with Jen as you would have liked. Jen has been left with au pairs, a number of whom you feel have not been up to scratch at all. You have been travelling to and from the UK for some time, and finally you and your husband decided to make the move together with Jen, as you heard the schools are great. You previously lived in Italy, and while you will miss the weather, you will not miss your old job: you're taking 6 months off now to 'de-stress'.

HPC:

It was only since arriving in the UK that you noticed the problem. Jen has been so exhausted all the time, she barely manages to play with her friends. You feel that she is suffering at nursery as a result – the manager there says that she is falling asleep during the day. You remember your au pairs mentioning that she was very sleepy and therefore 'no problem'. You're beginning to think that this is actually a problem.

Your daughter looks pale and you are worried that she is not gaining weight as much as she should. She has been passing normal bowel motions and her waterworks seem normal. She doesn't complain of discomfort after eating, but does have pain in her limbs, as you have noticed she doesn't join in as much as the other children when playing games. You originally put this down to the tiredness associated with the big move, but a few months later, it's still there.

PMH:

Jen hasn't really been to the doctors about anything in particular in the past, apart from all her regular vaccinations. She was a good weight at birth and it was a normal delivery. She is eating just as much at home, but doesn't particularly enjoy the English food. You suppose that she'll get used to it!

ICE:

You are really worried about Jen, especially since you have been told she is not gaining weight. You would like to know what the problem is and what you can do to help. You have been on a diet yourself for the past year and this has influenced what you are feeding your daughter as you are trying to eat less carbs and more fruit and vegetables. You worry this may have had something to do with it. Moreover you are concerned that the tiredness and sore limbs may just be some kind of way for Jen to put across that she is not happy after moving to England. You're really worried this is your fault.

FH:

You think there actually might be a problem with tiredness in the family, as your brother and your mother both seemed to be tired and have painful joints in the past. However, your two sisters, who you are in more regular contact with, have not been affected.

SH:

Jen lives with you and your husband and the family dog Romeo. You are stills settling into living in the UK so have not yet developed a strong social network and are worried about your daughter. Your husband works in as a delivery man and you previously worked in business, before taking time out due to feeling overworked. You had put your tiredness down to that!

Examiner Instructions

This is not a particularly common case, and is rather challenging due to there almost being two patients to assess, instead of one. However, students should be expected to focus on Jen, as per the instructions, whilst heavily involving Maria in the history. An exemplary candidate will focus on the social history and Maria's ideas in order to identify this inherited condition.

An example differential diagnosis may sound like:

'My primary differential diagnosis would be a haematological condition: anaemia. This is due to the patient being tired all the time. However it would be appropriate to consider an inherited cause of anaemia, due to multiple family members being effected. Given the family originate from Italy, and the history of joint pain, I would suspect thalassaemia. I would also like to rule out a haematological malignancy due to the fatigue, weight loss and vague pain history.'

Example questions may include:

- What is thalassaemia?
- What other causes of anaemia are you aware of?
- Are there any red flags to identify in this case?
- How do you investigate patients for thalassaemia and what would you expect to find?
- What is the treatment for patients affected by thalassaemia?