Activity in the virtual clinic – taken from:

Unit 6: Epilepsy and Paroxysmal Disorders

Section 9: Epileptic Encephalopathies of Childhood, Adolescence and Adult Life

Activity 11:

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Read Chapter 9E in Epilepsy in Children (pages 157-160).

Now look at the following referral letter for a child you are going to see in outpatients.

Dear Doctor
Thank you for seeing this 19-month-old boy whose parents have asked for a second opinion. He has rather frequent febrile seizures, which are not responding to treatment.
Clinical examination is normal. He has had a normal CT scan and his EEG normal.
Yours sincerely

You obtain the following history:

He is the second child, with a normal 5-year-old sister. There is a family history of febrile seizures in an aunt and two distant cousins of the mothers, but no history of epilepsy. Perinatal and early developmental history is normal. At the age of 7 months he had his first febrile seizure. This was a 15-minute right-sided clonic seizure for which he was admitted to hospital. One month later he was admitted again with a febrile "generalised" tonic clonic seizure lasting 40 minutes, however his father is adamant that this seizure started on his left side. Following that he was started on sodium valproate. Over the next 4 months he had 4 further admissions with long "febrile" seizures sometimes right sided, sometimes left and sometimes generalised. His mother thinks that he was not febrile during all of these seizures. At the age of 14 months lamotrigine was added to his treatment. In the past 5 months he has had very frequent clonic seizures which are usually 10-15 minutes long and more often afebrile than not. He has started having subtle jerks of limbs trunk or face and his parents feel his balance is worse than before.
Clinical examination reveals slight truncal ataxia and occasional fragmentary myoclonic jerks of one or other arm neck or eyelid. There are no other abnormalities.

His parents ask you the following questions. List the key points you would wish to convey in your answers to these questions. You will of course spend much more time discussing this with his parents!

1. What is wrong with him?
2. Does he have epilepsy?
3. Why is he not responding to treatment?
4. Why does he seem to be getting worse?
5. What is going to happen?
6. Is it genetic?

Commentary 11:

1. It is likely that he has Dravet syndrome (Severe Myoclonic Epilepsy of Infancy (SMEI). This can be quite a difficult syndrome to explain to parents.
2. Yes this is a type of epilepsy and not febrile convulsions. It would be important to determine what the parents understand by the term *epilepsy* and explain how you are using the term.
3. You explain that Dravet syndrome is one of the more difficult epilepsies to treat and that the usual treatments do not work very well. You would not want to be totally nihilistic about this, but introduce the concepts of difficult to treat epilepsies.
4. He may be worsening because of the natural history of the condition ie in the second year of life other seizure types tend to develop and become more frequent, which is often associated with slowing in development, clumsiness and ataxia. It is also possible that seizures have been exacerbated by the introduction of lamotrigine. A recent study suggested that lamotrigine should be avoided in this syndrome because of the possibility of seizure aggravation.
5. The long-term prognosis is poor, both for seizure control and cognitive development. You need to introduce this fact to the parents. There is also an increased risk of early death in this syndrome including an increased risk of sudden unexpected death (SUDEP).
6. This will be quite a difficult question to answer. Firstly it is exceptionally unlikely that any future children will have this epilepsy. On the other hand genetic factors are important and the family history of febrile seizures is likely to be relevant. You might also explain that many children with this syndrome have a specific mutation in a sodium channel - SCN1A (you will, of course, have to explain what a sodium channel is) and that 95% of these mutations are *de novo* ie. not inherited from their parents. It may not be very useful at this stage to go into such detail. However you should either provide it as written information or direct the parents to a source of such information.

Another activity…the virtual ward round – taken from:

Unit 9: Metabolic, Nutritional & Systemic Disease

Section 7: Neurological complications of systemic disease

Activity 1:

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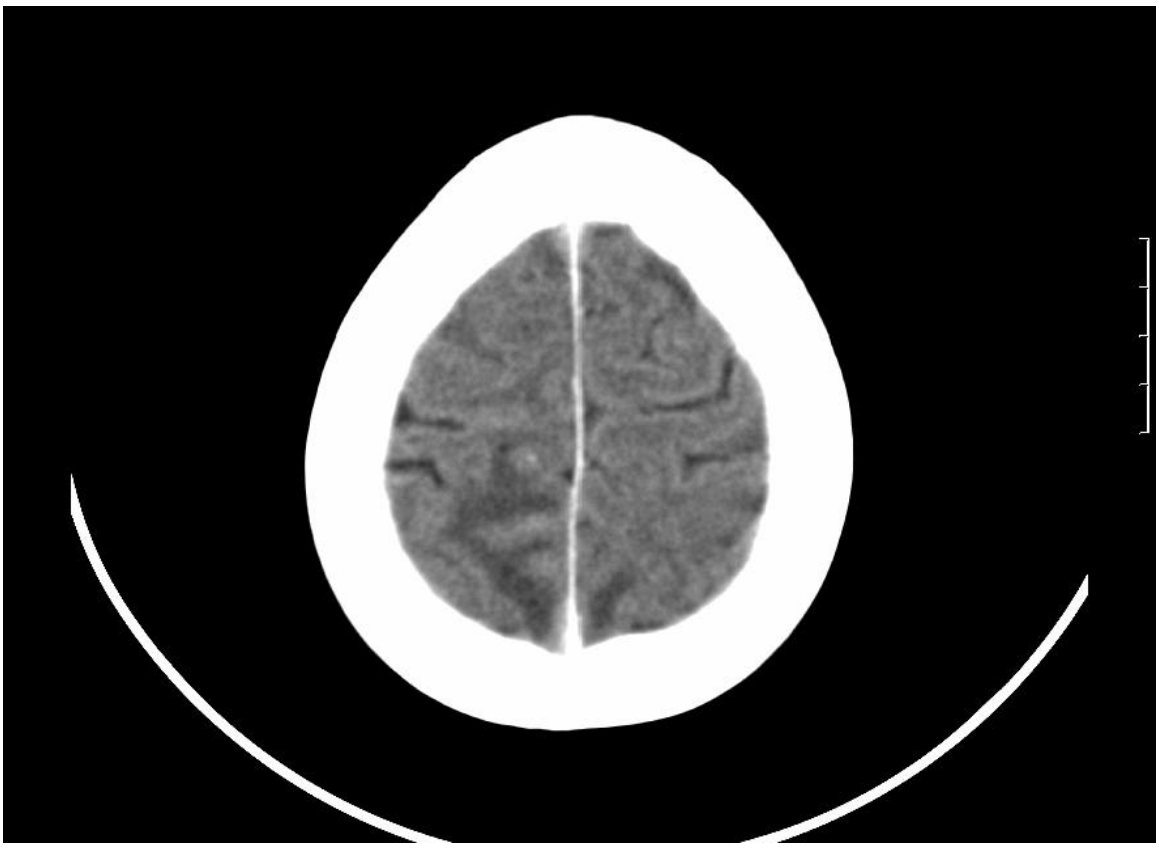
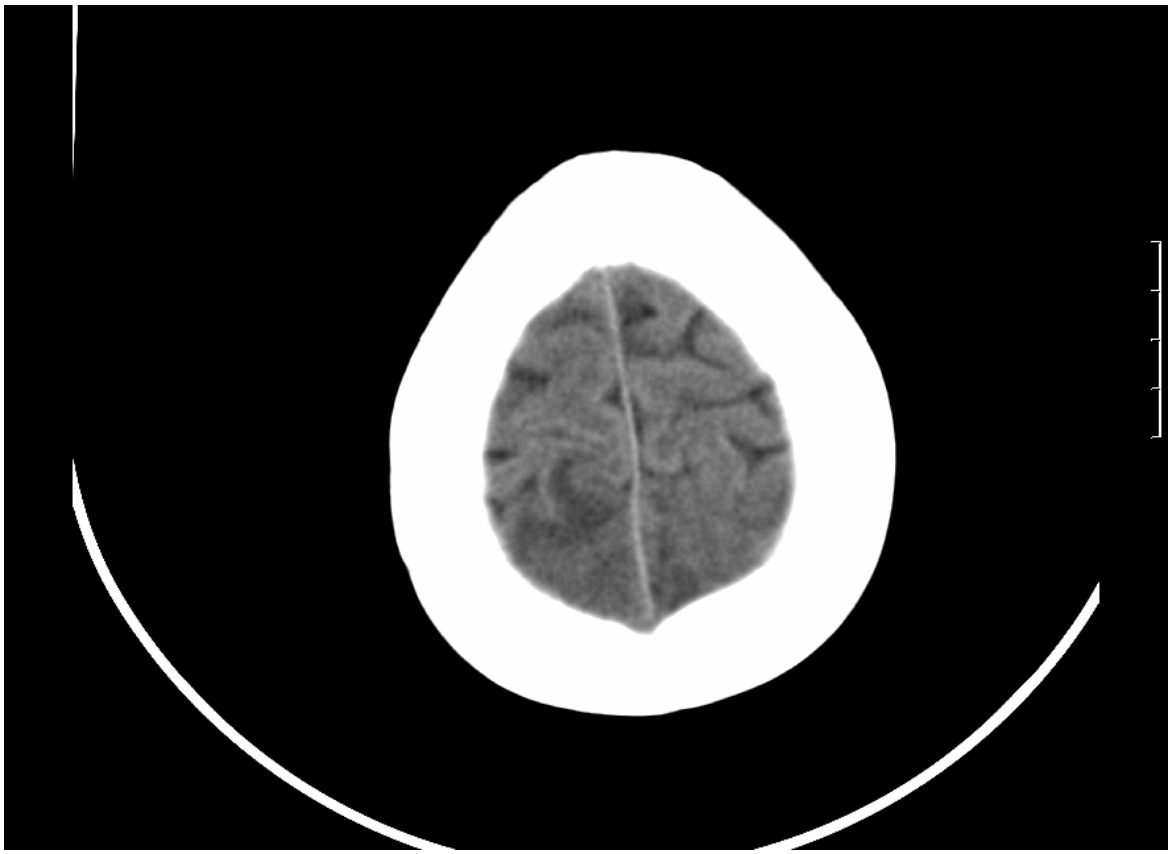
The ward round... you have been asked to see two children who have had some seizures overnight and who have been treated with phenytoin acutely.

The first patient is an 8-year-old girl who had a haematopoietic stem cell transplant three days previously. She is being immunosuppressed with prednisolone and cyclosporine (CyA) and has recently been hypertensive. Documented in the notes are three generalised seizures, an infusion of phenytoin at 10mg/kg with no seizures after this was given, and a reduced level of consciousness with a GCS of 8.

On talking to her parents you find that she was complaining about blurred vision yesterday evening before the seizures started and that, apart from her leukaemia, she has been previously fit and well.

Examination shows her to be conscious but disorientated. She appears to be blind in her visual behaviour, but her pupils react to light with no afferent pupillary defect. She has a left hemiparesis. You think you know what has developed but want to confirm it with imaging. Whilst arranging a CT-scan of her brain, you ask the ward team to treat her hypertension, stop the cyclosporine and ask the laboratory to do the sent CyA concentrations urgently. In the scanning room, you see this:





What are the main scan findings?

Commentary 1:

There is low density in the white matter of the occipital lobes bilaterally, extending up into the right parietal lobe.

Activity 2:

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Returning to the ward, you consider what to say to her parents. You are in a hurry, but they are obviously worried. In the end you decide to say *three things*.

As you struggle to find your swipe card, your resident rescues you with the news that the CyA concentrations were above the therapeutic range.

So what are you going to say to her parents? If you are unsure, read Paper 1, by Hamit Ozyurek et al on “Reversible posterior leukoencephalopathy syndrome”.

Commentary 2:

You should have said something along these lines:

1. The condition is called the reversible posterior leukoencephalopathy syndrome and you have seen it in many other children undergoing similar treatment (to add credibility to your information).

2. The most likely causes were the hypertension and/or cyclosporine.

3. Now that these are being treated, you expect her to start to recover over the next day or so and are hopeful that recovery will be complete.