

## **Summary of ECD Global Alliance Internet Chat**

### **05 Dec 2015**

#### **5 Attendees**

- A member who has recently had surgery to remove kidney stones said that he had now had all the bits of rubber (stents) that were left behind 2 weeks ago (deliberately!) removed. The highlight of his week had been that a lady doctor had drawn an arrow on his inner thigh, to point in the direction of his "bits", just in case they tried to get to his kidney via his ear-hole by mistake! She then initialed the drawing! He has never been drawn on by a woman before, and isn't planning to wash for weeks.
- A newly diagnosed member has been having back pains. He hasn't yet had any scans because he hasn't yet seen a doctor who knows about ECD. Another member said that he had started with a lesion in his spine. Another said that, a little while after being diagnosed, it was noted on CT scans that he had some crush fractures in his lower spine. He didn't have anything done to fix the fractures, and didn't need to wear any kind of support. He would not have known about them if the doctors hadn't told him!
- It seems that quite a few ECD sufferers have lesions in the spine. Ordinary X-Rays don't show these up as well as PET scans, which show active areas of disease.
- A member who has been on vemurafenib (V) had tried to go without to help reduce his side effect of painful limbs. This hasn't really helped and he is back on a dose of 2 in the morning and 2 in the evening.
- A member said that one of the "scary parts" of ECD is that it seems to progress quietly (under the radar), often without symptoms until one of the affected areas starts to show itself.
- The mother-in-law of a member had complications with her spine. Much of the stuff she had to use, such as the walker and the wheelchair, are now being used by her son-in-law.
- The speed (or lack of) with which the services move for people with ECD, or other rare diseases, was commented on. The member in Canada will be seeing a doctor soon and will try to get some answers regarding his spine pain. Hopefully, there will be some recommendation for V. It may well take a while to get that in action, but they have to wait till then. In their province of Canada, V is one of those extra special drugs which have to be reviewed by a jury/judge to decide on the feasibility of the insurance company to cover it.
- A member asked whether it would be possible for him to enter any of the ECD studies. It was pointed out that none of the studies are "free", except for the ones under the National Institutes of Health (NIH). Only the actual drugs are free for most clinical trials. Going to the US is not possible at the moment. Travel insurance will be problematic and the travel may be too hard on the member with ECD. However, it may be easier to organize a trip to the United States than to battle with insurance in Canada. They hope that they will have a doctor that can help with the insurance part. One of the recommended trials, BRAF and MEK inhibitor, will not be possible as the period for joining the trial will be out. If V is approved in the US this will help in trying to get it in Canada. They will be relying on the articles in New England Journal to help to make the approval of the extra special insurance happen.

- A member who has been through all of this said that he would recommend just going for V. It has a good track record for BRAF+, and people are able to get it off label in the US. He thinks that others in the V trial are trying to do the same. It's just taking up too much of the patients' time to be traveling monthly, indefinitely. Much easier to get V off label!
- The member hoping to be started on V has had 8 or 9 MRI and a few CT scans, but no PET scans. He is waiting for a doctor to order a PET, but they are waiting for further confirmation of the ECD diagnosis. They were not in agreement with the two assessments that had been made. One had looked at the X-rays; the other was the urine test.  
The PET scanner in their city area is earmarked for cancer only. They will have to push for a PET after they have seen the doctor later this month.
- It was said that PET scanning is often the key thing that the doctors look at during treatment. However, it may not be viewed the same way for making the initial diagnosis.