

Summary of ECD Global Alliance Internet Chat

14 Dec 2013

10 Attendees

- Before we started, Susan, a new Chatter came on and said hallo, and that she found the Chats to be helpful in her efforts to understand ECD. She asked about signing out from the Chat room. The answer is that you just close the Chatzy page and you are "signed out".
- markmcelwee also came on early to let us know that he would probably not make the Chat due to other commitments. His daughter, Kit, has ECD and has recently had 2 seizures, and been restarted on Keppra. She will be attending the NIH ECD study in the last week of January for a return follow-up.
- A member has recently started vemurafenib. She has not been feeling good, and has a lot of joint pain.
- We were told by one member that he had been at Sloan Kettering last week. This had been a "pretty good experience" and he is waiting for the biopsy results.
- A member told us that he had read the report of a recent MRI. It said that the ECD has now progressed up though and beyond his knees, to involve his upper legs. He is not on any treatment at the moment. His doc stopped his treatments because they weren't working. His doc is waiting to hear back from Dr Estrada-Veras to know where to go next. He is also waiting for the results of his bone biopsy. Dr E-V has the tissue block now, to run the tests. His Dr. is trying to get preauthorization to start the vemurafenib. He had been on Pegasys since August, but the recent bone scan has showed progression. He is going to try to go to the NIH in March, unless Dr E-V says that he should go there sooner.
- A member asked about problems of lightheadedness and spinning around. Spinning around is usually because the inner ear is not working properly. Lightheadedness is due to the blood pressure falling (like a faint), or due to lack of sugar in diabetics. Her Doc has told her of the need to keep hydrated, but even with drinking more she is still having problems.
- Members discussed the difficulties of finding pain-relieving medicines that actually worked. Members mentioned the various medications that they are using.
- A member is now on week 5 of Pegasys. The attacks of pain lessened, at first, but then returned to their usual intensity.
- A member was asked when symptoms first began. She was only 14, and is now 20. Joint pain was her first symptom. She had it for about 2 years before she realized

that something was wrong. Now, she only has pain on rainy days, or when she doesn't move much. Nobody had expected ECD to be the cause of all her symptoms, but a biopsy of her rash gave the answer. She has given up her job in a fast food outlet, and is now a dog walker. She is pretty active but not like she used to be.

- The fake sign-language interpreter at the memorial service for Nelson Mandela was mentioned. It was agreed that this sort of thing should not be allowed to happen!
- Dr. Diamond at Memorial Sloan Kettering (MSKCC) was mentioned as being a "Really caring and sharp guy". He is lead neuro-oncologist for the ECD trial at MSKCC. He saw a member because there were tumours on the brain. This member has only been to Sloan once so far, and has not been admitted into the trial yet. If he has the BRAF mutation then he will probably be entered.
- A member has been in the trial since August. She has lost over 70lbs of fluid, and got her mobility back. She is "living an almost normal life again". She needed a scooter back in August, but can now walk unaided, and walks on the treadmill at 1.2 mph for five min at physiotherapy.

She started seeing results after just three days of vemurafenib. She is taking 4 + 4. The member who has started vemurafenib, and is having trouble with joint pain, is only taking 2 + 2.

- Another member was interested in this as he expects to be one of the next recipients of vemurafenib. Hydrocodone is not working for him now. He wonders what he will do if his joint pain worsens, once he starts vemurafenib. He is having a baseline ECG, and he is going to see a dermatologist for a skin baseline examination.
- A member who has already entered the vemurafenib trial told us that she had MRI, PET, ECG, dermatology review, a 6 min walking test, blood work and a heart ultrasound before starting.
- Members discussed their inflammation markers. One had found that his CRP decreased after interferon treatment. But for another member the inflammatory marker levels did stabilize a bit on interferon, but they never went down. Then the markers started to steadily climb. Coupled with a recent bone scan showing progression of the ECD, the "team" determined that the interferon wasn't working.