

Summary of ECD Global Alliance Internet Chat

25 Oct 2014

8 Attendees

- Others came on after the Chat had finished.
- The change in timing was discussed. We were reminded that the change to winter time occurs on 26 October in Europe and a week later in the US. No one liked the longer winter evenings that we will soon be having.
- A member has his reports from his recent tests. They have shown that the disease is still spreading. It is now in his shoulders. A tear has been seen on the right side, and it looks like he will have to have surgery on the shoulder. The tear is in the superior labrum in the right shoulder. There is more disease in his legs. They found thickening and edema of the middle glenohumeral ligament, and it seems that he has developed multiple micro fractures in his ankle. He is on vemurafenib (V), and started at a dose of 4 pills the am and 4 in the pm. This caused acute pancreatitis and put him in the hospital. He is only the second person on record to have had chemically induced pancreatitis due to V. The drug was stopped and then gradually built back up. He has been on it for almost a year now, but it has not slowed the ECD. He has "had no improvement at all, it has actually gotten worse". He also has QT prolongation (the QT interval is part of the heart's electrical cycle) which is already borderline abnormal.

He goes to see the doctor this week. He doesn't know where they will go now. He has a feeling that actual chemo is next on the list; he had interferon before V. He had a further bout of pancreatitis 2 weeks ago. The disease appears to be only in his bones, although they think that it is the cause of the thickening around his pituitary gland.

He said that he has asked several times about anakinra. He even asked Dr. Estrada-Veras about it when he was at the NIH. He was told that they "wanted to see how another patient was doing on it first" since his symptoms are so unusual. There is only one ECD patient with similar symptoms, and he is in Ireland. He said that he has trouble getting his doctor to consult with any of the other doctors that have some experience with ECD.
- Another member mentioned that he has had chemo with cladribine (2CDA). This had not been too difficult to take. It just didn't work for him. A number of other patients are on 2CDA, and, for a member whose daughter has ECD, it was the treatment that pushed her into getting better.
- Then the members discussed Kineret (anakinra). A number had found it to be good for bone pains. A member, who has the ECD mostly in her bones, had great improvement with it, but it had to be stopped when she needed treatment for lymphoma (which the doctors found her to have as well as ECD!!). She is going in for round 4 of her chemo next week.

- Another is using anakinra which is helping her bones, and another was doing well on it before he started V. This member was stable on anakinra. On V, he had a slight improvement at first, but has been stable since. His dose has just been reduced by Dr. Janku. He had started at 4 and 4, but had lots of side effects. He then went to 3 and 3, and was a bit better. Now he is on 2 and 2. Dr. Janku says that "2 and 2 is plenty for ECD patients, and the goal is an improvement in quality of life."
- One member taking V is on 1 and 2 which she told us was plenty for her. "I wouldn't be able to move on 4 and 4", she told us. She is doing well. She had a check-up at the hospital the previous week. Her tests are very stable, and she is feeling good and exercising every day.
- A member who has also been having shoulder problems has been doing better since having surgery. She is not wearing her sling now. It just hurts if she moves too fast without thinking. At surgery, they saw many things in the ligaments, and some extreme inflammation. She had the surgery because she had a slight rotator cuff tear, and bone was riding on bone.
- A member had seen his oncologist this week, and all was stable. The doctor is retiring soon, and said that he was sad to be leaving. He said that he is going off to join a rock band! The new doctor is particularly interested in rare diseases, which is fortunate!
- A member, whose wife works as a nurse, bemoaned the fact that the politicians are "playing games", and that the nurses are given all the bother.