Summary of ECD Global Alliance Internet Chat
31 Oct 2015

8 Attendees

- A member had left a message that he wouldn’t be at the Chat, and that "All systems are go" for his kidney stone surgery this week. He will probably miss next week as well.

- A member was on her way home to Lexington after dropping her son off at the Cincinnati airport. He will work at a golf course in New Zealand until March 31, 2016!
  She is going to go to Memorial Sloan Kettering (MSK) sometime before November is out. She feels that she needs more support than is offered in Lexington. The neurosurgeon and orthopedic surgeon “here are clueless...never heard of ECD”. She is going to see Dr. Diamond in the next 2-3 weeks, before Thanksgiving, but she has no exact date yet. Her medical info from Lexington was faxed to him, and he can get the NIH information. He actually talked with her when she was in Houston!

- The member from Canada, whose husband is going through the early stages of getting a diagnosis, has had some news. The test on his biopsy came back as possible for ECD, but further testing was needed. This cannot be done local to them, so they are in the process of having it tested at the National Institutes of Health (NIH). They will do a basic test for ECD to confirm it, and check for the BRAF mutation. These tests will be free. The first positive test was on the urine test by Trovagene, so now they need to see if the tissue biopsy is positive as well.
  She is finding it “darn frustrating” having to explain things from the start to each doctor, but at the end it is worth it “if they get it”, and help you to get to the next stage. They will be seeing a rheumatologist soon who was recommended by another ECD Canadian patient.

- A member who is taking vemurafenib (V) has been on a dose of 3 in the morning and 3 in the evening since May 2015. Her insurance is paying for this. She is having major problems with her right shoulder and her arm is beginning to tremble. She has bad pains which radiate down the arm and are getting worse. The shoulder pain started after she began the V. The pains that she was having in her legs, due to areas of sclerosis, went away. Her PET scan doesn't show any hypermobility, but MRI shows a moderate tear in the rotator cuff. One orthopedic doctor had the radiologist shoot it up, under fluoroscopic guidance (a technique of seeing where you are putting a needle or injection by using X-rays) with cortisone. But it was no help at all. She doesn’t know whether the shoulder damage was caused by ECD. It was suggested that she could try either heat or cold compress to help the pain, and maybe elevating her shoulders at night, on a pillow or something similar might help. She is seeing Dr. Diamond in a week or so – if she cannot wait that long, perhaps her family doctor may prescribe a pain killer. She is going to start rehab (BTW her son-in-law is a Physical Therapy (PT)), but she feels that she may need surgery first. She gets in a warm water pool several times a week, just to do her own PT.

- A member who has had V treatment said that because she was on 3+3, there is also a chance that joint issues could be side effects from V. He knows that a number of ECD patients have had to stop treatment because they couldn't tolerate the joint pain.

- Kineret was discussed as a further treatment. One member is on it, and Dr. Janku has suggested that it might help the member with the bad shoulder. Someone had read that it is no good for a patient with CNS disease.

The possibility of trials of Kineret in ECD was raised. It was said that such trials would be unlikely to get sponsored as there just aren't enough ECD patients to run many stand-alone clinical trials. Even the V trial is done in a basket study with 10+ different other cancers. It is to be hoped that the efficacy for V in BRAF+ subjects in the Phase 2 study, is so good that a Phase 3 study isn't needed. After all, there is no existing drug to compare with.

- Another member said that before he had ECD diagnosed he had a problem with his right shoulder. He couldn't even lift his arm up. Vemurafenib pretty much took care of his eyes, brain, spine, bones, etc. and now he has full motion in his right shoulder, without pain. The pain was never fully diagnosed as being from the brachial plexus (a complex nerve "interchange"!!), rotator cuff (muscles around the shoulder) or bone. His orthopedic surgeon just sent him for PT, and wasn't curious to look into it further. "He would have no knowledge about it" and "would mess everything up".

  He is not aware of anyone in the V study that is also on Kineret because of pain. He didn't think that you are supposed to use other treatments during the trial, unless it is for another condition, not for the ECD.

- In a new member's case, he has both joint and muscle pains. The shoulder pain actually came from his spine. When that was operated on, his shoulder pain went away. His wife said that sometimes it is the act of doing something positive that will help to make things feel better.

- Access to drugs in different countries was discussed. At the moment there is no access to V in Canada, as V is not yet approved by the FDA in the US. There are greater restrictions on drugs in Canada. Health Canada usually follows the FDA after a few years. They set their own guidelines and "in some cases they are really strict" about what is allowed.

- It was pointed out that since the V trial isn't happening in Canada, they can only follow the FDA, and approve V, after the US does, and even then, it may take another 5 or 6 years. Perhaps the best approach is to promote knowledge and education about ECD. Have an ECD treatment center, eventually. In Canada, if someone has one of the more common medical problems, there are special centers that help them and they can get excellent medical treatments. Some places, like Toronto Western Hospital, are better than others, but with ECD being not so well known (understatement of the year!!), all the doctors are still learning. They are still with their local family doctor, who has been excellent in going beyond what they have asked him to do, and has been in contact with NIH. One of the reasons that this family doctor is good, is he is a good listener. They are also still with the same neurosurgeon and, hopefully, will soon see a knowledgeable doctor. They still have to deal with "educating" the heart doctor, but will gather the necessary information; information that we all share in the trials will make life for others better.

- A US member said that he was particularly concerned about the situation in Canada, because his parents, siblings, and many relatives are Canadians. In the US, most regular doctors can't even afford the time to get up to speed. He doesn't have local doctors anymore, he can't find one who knows enough about ECD. When he goes to Memorial Sloan Kettering (MSK), Mayo, or University of California San Diego (UCSD), he asks them to do any other tests or checkup for him. "Since we are 'big clients', most of the time, they'd do it".

- The member who has macular edema said that her eyes had improved this past week. She is "doing OK". Still tired, and still taking sorafenib (she is ARAF+, not BRAF+!). She had asked Dr.

Diamond if Sorafenib and Kineret would be a possibility. Since she is not on a trial she will have a lot of flexibility to use different combinations, as long as she can afford it.

Her biopsies have never been conclusive for ECD. She was first diagnosed with Rosai-Dorfman Disease (RDD). After her second biopsy, her doctors said that it mostly favored ECD. Also, her disease presented itself in the long bones, kidney and skull/orbits like ECD.

When they thought it was RDD, she was treated with Cladiribine, and Clofarabine (both chemotherapies). She has also been on Kineret, and takes prednisone too. The Cladiribine and Clofarabine kept her stable. The drug that she is now on (Sorafenib) has shown little bits of improvement. Her biggest issue is her eye problem. She keeps getting macular edema, and this has been hard to control. She sees a retina specialist where she lives, and also at MSK. They are hoping that the Sorafenib will take care of the disease, and therefore the eye issues.