

Summary of ECD Global Alliance Internet Chat **25 July 2015**

10 Attendees

- A member left a message to say that he would be missing the Chat because he had been having fevers and feeling awful most of the week. This was probably due to a urine infection. "Aunty Biotics" seemed to be getting things better.
- A new chatter came on seeking information. She lives near Toronto in Canada. Her husband may have ECD, but it is taking a while to get it confirmed. The "general" symptoms that come with ECD seem to match his – but she would like to know more about how others deal with these.
- A member, who had previously been diagnosed, got this confirmed by biopsy (3 samples of bone from his right leg). X-rays were taken before of his legs and arms, since there were lesions there, and it would be a fairly easy way to get a sample.

He had had bulgy eyes and he had started to get lightheaded at times. CT scans of his abdomen showed masses consistent with ECD. He had slight white masses around his kidney (hairy kidney). He said that there was no telling how long he might have had the disease. It could have been for several years. He is only 35. His speech, gait, and swallowing were all affected. His balance was "God-awful". It all changed once he started Vemurafenib (V). On June 1st, he was walking like he was 98 years old. By July 1st, he was able to jog!

He emphasized the importance of being tested for the BRAF gene, if ECD is confirmed. He tested positive and the medication that he was started on worked within days. He said that there was research into a way of testing BRAF levels in the urine. The only side effects he has had are extreme sensitivity to sun and hair thinning. He now feels "absolutely 100% normal"!!
- The woman whose husband may have ECD said that he was 48. They think that there are other things that have been happening in the past 6 years that may be connected to this. She had heard that the test for the BRAF gene is not always positive (only positive in 50% of ECD cases). She was told that if she goes to the ECD Global Alliance website, she will see a "laundry list" of drugs, a lot of which are hit-or-miss in their effects on ECD. Only vemurafenib has been shown to have very high response rate, for patients with ECD and the BRAF mutation.
- The best type of doctor for diagnosis and treatment was discussed. Dr. Diamond, at Memorial Sloan Kettering (MSK), is a neurologist. One member was diagnosed by an oncologist/hematologist and another had a team of doctors diagnose him. A radiologist had suggested it, and the other doctors had no clue what ECD was! The chief hematologist at Stony Brook had never had an ECD patient, but the pathologist had diagnosed ECD 3 times.
- It was advised that if the wife of the possible new ECD patient goes to MSK in NY there is "an amazing doctor" whose name is Dr. Diamond. He is treating over 30 patients with ECD. She was recommended to get a diagnosis, and then find a doctor who is knowledgeable about ECD and has treated patients before.

She asked about the appearance of lesions on X-Ray. One of the members who has been having treatment said that the lesions are symmetrical (on both sides of the body). He has them

everywhere, but nothing caused him pain. It was about a month before treatment that he started to notice symptoms-all related to lesions in the brain stem. This is what causes ataxia (poor coordination in arms and legs, and slurred speech).

- Effects of V with having children were discussed. It was said that there was no information about this at present, and that "precautions" were advised.
- A member who has waited a long time to find out his BRAF status has heard that Dr. Estrada-Veras at the NIH found that he was not BRAF+. However, Dr. Estrada-Veras has encouraged him that other treatments can be used. There are trials starting soon for BRAF- patients with new drugs that have come onto the market. He was diagnosed in 2007 and put on interferon right away. He has been stable for a long time but he would like his balance back. He was first troubled by a "wicked thirst" and depression. Then his legs started hurting.
- A member has discovered that there is a guy, who only lives 10 minutes away from him, who has ECD. He is on interferon, and was diagnosed in 1998.
- Someone noticed that alcohol makes balance worse, and that using a cane when getting up can help.
- The member who has relocated to San Diego is going to have his PET scans done at University of California, San Diego (UCSD) for the first time, and will meet with Dr. Kurzrock again next Friday. He was on the V trial at MSK and is now "in remission". It is time to find out if a lower dose of V will do the trick for maintenance.
- The member, who has macular edema as well as ECD, has been having a hard time. She now has edema in both eyes and prednisone is not helping.
- A new chatter came on whose fiancé has ECD, is BRAF + and has been on V. This was the first Chat that she had joined, although she has followed the summaries, since finding the Alliance a couple weeks ago. He is not part of the trial at MSK, but is monitored by Dr. Scheel at Johns Hopkins. She asked whether anyone on V was also taking tamsulosin/Flomax. They think that he is having a reaction to the drug combo. He has been taking V for 18 months, and was on Flomax for years before. They are wondering whether there may be an interaction between the two drugs that was unknown before. Dr. Scheel says none of his patients are on the two together.
He has had the V side effects (skin sensitivity, burning; shoulder pain; hair loss), but he also has incredible fatigue and pain, and issues with his kidneys that can't be explained by V alone. He has had elevated creatinine levels, obstruction of the kidneys, and severe flank pain. He has been trying to wean himself off the Flomax. Now the doctors are thinking of trying to switch his meds.
He hasn't had any tests that show improvement in ECD, but he was able to have ureteric stents removed as a result of what the V did for his fibrosis.
- A member taking V joined. She was at the beach (but protected from the sun!!!) for a week or so. Her PET scan scheduled for the previous week was cancelled because her insurance company denied it! A brain MRI is scheduled for Aug 2, after she is home from vacation (she hopes that that doesn't get denied too!). She would like to hear that the V is shrinking her cerebellar/brain stem lesions.