

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

### **14 May 2016**

14 Attendees

- Some messages were left in the days before the Chat. A new member from Miami wanted to hear about people's experiences on dabrafenib (D) and trametenib (T). She has been on them for about a month and has experienced chills and fever (among other side effects). She hopes these effects are temporary.
- A member came on and said that he was on D now after being on vemurafenib (V) for 8 months. He has no side effects yet, but has only been taking it for a little over 2 weeks. He had had to change from V because his liver enzymes were rising on V.
- The new member said that she had broken out in "itchy hives" an hour after taking D (150 mg) and T (2 mg). She had been on the maintenance dose for about 2 weeks when this happened. She hasn't taken the medicines since, and is taking medication to relieve the hives. When the Chat started she told us that the hives have cleared. She is still on steroids for the rash, but is hopefully restarting treatment at a lower dose in the next week. She sees her neurologist this week and is having bloodwork the day before. The doctor wants to see her liver function and keep tabs on her cholesterol. She hopes that she can stay on D and T, they seem to help with neurological issues. Her doctor managed to get insurance to cover the medications.
- Another member asked whether anyone had tried any essential oil therapy for relief from symptoms. A new member said that he tries to relieve pain with plants. He is waiting for confirmation of the ECD diagnosis.
- A new member asked how the Chats went along as she was new. She was told that we just chat away, ask questions, tell stories, and talk "about everything". She is from a "very small town" in northern Minnesota.

She is thinking about doing an interview with her local paper about ECD. She wants to talk about ECD, because so few people know of it. She has a friend who is a writer on the paper and she wants "to reach out to others that know nothing about it". She thinks that maybe there is someone who is having similar symptoms & is unable to find anyone to help them. We were reminded that there are lots of other rare diseases, and other patient groups will be trying to do the same things.

She is currently under the care of Mayo Clinic in MN. The doctor there was the one to finally diagnose her, after she had seen 5 different doctors. She has gone back to all 5, told them about it, and they all asked what the heck it was. Dr. Garibaldi did a biopsy of her abnormal eye tissue. He was the one to diagnose the ECD and referred her on to Dr. Habbermann. She is not BRAF+.

She has not yet started any treatment. She had a PET scan to see if she had ECD anywhere other than the eye tissues, and it revealed a thyroid cancer. This had to be dealt with first. The cancer was stage 1. She says that she "was lucky to be unlucky", because if she hadn't had ECD she would not have had the PET scan and the cancer would not have been found early. Her next appointment regarding a start of treatment for the ECD is June 1. She doesn't know the doctor's plan yet. The doctors say that the thyroid cancer & ECD were not related in any way.

- The member who is in the first phase of his V treatment is continuing to get better. His wife told us that he is standing up longer (or leaning longer).
- A member who is on V and has been having lots of hand problems had cortisone injections in both hands this week, for carpal tunnel in one and trigger finger in the other. She thinks that Dr. Diamond wants to change her treatment from V to a MEK inhibitor such as D. Her scans are stable, but not improving. She said that she is having a stress echocardiogram to be sure that her heart can handle it.
- The member who thinks that Dr. D may want to change her treatment, said that the target for the medicines was "everchanging". Vemurafenib was right initially, but after a year her bones, joints, and brain may be asking for something else! Her liver enzymes are ok, but her total bilirubin is slowly rising. She doesn't know if she has brain progression, but it is not "sleeping" as Dr. D said. The tumor inside the brain is protected by the blood-brain barrier. V is not supposed to be able to readily cross the blood-brain barrier and this is probably the issue. It usually takes longer for V to work in the brain anyway. Dabrafenib may be a better option for her.
- A new member asked one of the "old-timers" where he had had ECD. He said that he "had it all over", but V took away about 95% of it. He was diagnosed very quickly, in 5 days!! He had had to deal with "eye issues" for a year though, and had no idea that it was everywhere else too. It got to the point where he couldn't even walk, but after just a few weeks on treatment he was able to jog again. He has now changed to D because the V was affecting his liver, and his liver enzymes were rising. He is taking a dose of 3 in morning and 3 in the evening, 50mg a pill. He is due a PET in July. Dr. D probably wants to see how his body is responding to the drug.  
His liver enzymes are almost normal now (one is normal). He hopes he doesn't start to get any fevers or chills. He had bad night sweats before treatment.
- A member told us that Dr. D is trying to get her onto T, but her insurance doesn't want to approve this. She is appealing, but they say the appeal will take up to 90 days!! She is ARAF+ and thinks that the change to a MEK inhibitor would be because it works further down the cascade of mutation. The Sorafenib (she didn't take V because she is ARAF+ and not BRAF+) that she has been taking reduced the lesions in her bones, but has done little for her eyes and it has lots of side effects. She has choroidal (a layer of cells in the retina) involvement and she continues to get macular edema. This (edema) goes away with steroids, but as soon as she begins to wean off the steroids, the edema returns. The steroids are taken orally. She has had steroid injections into the eye, but the edema came back stronger than ever after the effect of the steroid wore off.  
She used to get some night sweats in the beginning. All of a sudden, she would get chilled (goosebumps everywhere) and a low-grade fever, this would be every night. If she stopped the steroids she is sure that this would happen again.
- Another member said that he still has night sweats once in a while. He is VERY keen on exercise, and thinks that it is related to his running, exercise, and a warm shower (so his body is still pretty warm when he goes to bed).
- The topic of health insurance came next. A member, who has just changed from V to D, said that there had been no difficulty getting the change approved. He just called them after the doctor had given him the prescription ( \$0 co-pay too!!). He said that he has Empire NYship which is only available in NY (although he assumes that they have a "regular empire"). They said they'd approve everything with no questions. He also has GHI which "I don't recommend, even for dogs!"

- It's easier for a patient to get approved, if somebody else has already been approved. One of the problems with insurance is that they don't have a joint standard. Each insurance company seems to have their own specific methods. Doctors, like Dr. Diamond, are willing try new things and are producing a lot of data. But even if the treatment works, this is not enough for it to be approved by FDA. However, it does open doors for a future clinical trial.
- A patient who is not on the Chat regularly (because she has been "doing good") told us that she had got a bump on her head that was biopsied. They said that it wasn't a cyst, and is not cancer. It was 'histiocytic', but not necessarily ECD. It was as big as a dime but seemed to be shrinking. It is being sent off for further studies. She is on interferon alpha. She has had a lesion grow a few millimeters in her brain so she has had her dose increased and she finds that it makes her a bit sleepy. She doesn't know her BRAF status. She goes to SAMMC in San Antonio where she has MRI, PETS, CATS, and gets seen by cardiology, ophthalmology, and dermatology.  
Her doctor says that as long as she does well on her current treatment, "we'll stick with it". However, if he gets worried about her brain changes, he may switch her.  
She was diagnosed 7.5 years ago, although probably has had ECD for 16+ years, because she was diagnosed with DI in 1999. From 2000, she was having severe pains in her kidney region, but she went gluten free 4 years ago & has never experienced it again. She is now symptom free.