

## **Summary of ECD Global Alliance Internet Chat** **04 April 2015**

9 Attendees

- A woman came on who has a sister diagnosed with ECD, who is very ill and is now in a community hospital. She was diagnosed in January, and has been getting worse. The doctors have said that they won't give her the recommended medication in this hospital.  
The sister with ECD is based in Florida and not on medication at present. She is BRAF negative. A member said that vemurafenib (V) would not be the drug of choice for her, and it should be interferon. This is often considered a first line treatment, and many patients have used it. We did not know whether it would work in advanced cases.
- The medication situation is difficult, especially in the US, because there is no standard treatment for ECD, and it is such a rare disease that the drug companies aren't going to spend much time and effort on us. Also, the drugs that have been tried, are often frighteningly expensive. Interferon is often used although expensive (one member was on \$360 a week's worth), and the newer experimental drugs such as Zelboraf (vemurafenib) are more like \$300 a day!! That is one reason why the members who are on these drugs are almost always involved in trials. [Editor's note: Drug trials are needed to get treatments approved for ECD. For a trial to be successful, patients must enroll. At the conclusion of a successful trial, a treatment can become approved for ECD which will make it easier to have insurance companies and others payers pick up the cost of the treatment for other ECD patients.]
- A member told us that he had been on interferon for 10 years before starting vemurafenib. Another member was lucky enough to respond to the interferon, and it has kept him stable. Another member had interferon treatment, but "it didn't work for me".
- Those on Vemurafenib (V) then compared their doses. One member, who was not on the chat has reduced her dose to just 2 a day. Another had started on 8 pills a day, but he wasn't able to stand taking that many for more than 5 days, and dropped to 6 after a week's break. He is now on 6 pills and considered to be "in remission". He believes that one can go onto 2-3 pills as maintenance. His tumors were in his eyes and in his brain, but V has reversed damage from all his tumors and bone lesions. He was asked whether V had raised his blood sugars, and the answer was that it had not. We were reminded that some of us are treated with Kineret (anakinra).
- One of us said that his doctor at Mayo is a pioneer of interferon, and that he had led many of the clinical trials of its use. And another said that the doctor that she saw at Duke was trying to put her onto interferon, but she had "nicely" declined, and begun her correspondence with Dr. Estrada-Veras at NIH. She is starting the phase 2 trial in two weeks, and is BRAFv600 positive. Since she is BRAF+ it was said that the Dabrafenib and Trametinib (D&T) trial should work for her. She asked whether anyone knew whether her hair would fall out. She is supposed to get it cut and colored, before she goes to NIH, but she doesn't need to spend a lot of money on hair that can't be kept! A

member already on that trial hasn't heard much about people losing their hair, although he has lost his eyebrows. He has heard of someone getting curly hair. The member about to start the trial said that since her hair is very straight, "curl would be nice".

She has disease in her cerebellum and bone. She has never had a PET scan, just a brain MRI and X-rays of her legs. At the NIH, she will have brain MRI, CT, and whole body PET scans. "You should have a pretty good idea where everything is." A member was sure that NIH will "scrutinize your whole body before giving you the most expensive drugs for ECD". A member, who used to be a family physician himself, said "You can get locked up for scrutinizing whole bodies in the UK!" He had never dared to do this when he was still working!

- It was said that Sloan-Kettering is experimenting with a variety of drugs for BRAF negative patients. Dr. Diamond would probably be a good contact to get an opinion.
- It doesn't take too long to get a slot at the NIH, and it is free (paid for by tax dollars!). The member who soon starts the trial, was phoned last week, and was told that she is being scheduled for 26 appointments in 5 days!
- Member's difficulty with insurance was mentioned. It was said that, in general, people don't have problems getting Kineret or interferon approved. The woman, whose sister has ECD, said that she has been asking for authorization of interferon for 2 months, and finally has been in phone contact with a hematologist who has said that he would apply.
- A new member came on from California who has 2 teenaged children. She was diagnosed in November 2014. The doctors couldn't figure out what she had. They thought that she had MS, and she went on chemo treatment. It was very strong and intense, but it did nothing. They wanted to go into her brain for a biopsy, because she was dizzy and had lost her balance. Before the surgery, they asked if she had any weird lesions on her body. She had an "unusual thing" on her breast. They biopsied it and that is how they found out, "so no surgery". She had been getting no answers at Woodland Hills Kaiser, so she went to the top Kaiser in Sunset, in LA. She was diagnosed by Dr. Gaulwalla, a man who doesn't take no for an answer!

She is taking vemurafenib, 3 pills in the morning and 3 in the evening. She is not in a clinical trial so, she will be getting it "off-label". Her insurance company is paying for the V. She only has to pay \$30 a month!!! It was remarked upon how rare it is to get approval for ECD. It was said that the publications by Haroche, Hyman, Diamond, Janku, must have been cited.

She has been on V since November, initially at 4+4, but she broke out in an awful rash. She sees her doctor every month, and gets her blood tested. She has tumors in her cerebral cortex, but her doctor did a MRI and said they are fading. She still has the lost balance. She has also had PET scans. Her doctor is new to ECD (surprise, surprise!), so he "doesn't know much", but he has been looking on the web about the disease. She has another PET scheduled in two weeks to see if the disease is still active.

Health care providers and plans were discussed. Her care is with Kaiser, and it is an HMO plan provided through her husband's work.