

Summary of ECD Global Alliance Internet Chat **20 June 2015**

3 Attendees

- A member told us that she was doing “pretty well”. Her pain has been more constant than it was before, but not as sharp or strong. She is, hopefully, in remission. She has found it hard, going to the hospital all the time, while trying to take classes. Now that she does not have so many appointments, she hopes to graduate this year!
She still has an irregular bone marrow, and the lesions and cysts are still there. But some are smaller, and none seem to be active. She has no eye problems.
She was on interferon for well over a year and then, most recently, on Kineret. She is BRAF positive. She was on Kineret for about a year, and the doctors believe that the Kineret is what brought her into remission. She sees Dr. McNeer of the University of Chicago, and was diagnosed 5 years ago (but has had trouble for 8 years). Originally her doctors said that his lesions were juvenile xanthogranulomata.
- A member is now in week 8 of her course of vemurafenib (V) at a dose of 2 in the morning and 2 in the evening. She has always been very active, and has noticed her lack of energy, since being on the treatment. “I’m getting tired of not much energy.” She is seeing the oncologist during the next week, after an ECG and blood work. She wonders when she should have more scans. She had a number of scans (brain, heart, lung, etc.) at NIH in late April. She is seeing a private doctor in Lexington. This doctor asked the member, in late May, when she should order more scans. The member thinks that, maybe, she should have a brain MRI soon. She is the first ECD patient that this doctor has seen, so they are “feeling their way”. Another member said that she would suggest scans after 2 - 3 months of being on the drug (but she didn’t pretend to be an expert!).
- A member said that she was interested to hear about the length of time for which the member, who is now thought to be in remission, was on Kineret. She had been on Kineret for 4 months but her doctor took her off it as she was ARAF+, and another drug was recommended for her. The Kineret had really helped with her symptoms and her fevers went away, but after stopping the Kineret they returned. She is now on Sorafenib. Her doctor has said that usually, when you feel pain while on the drug, this means it's doing something! She also has an eye problem, macular edema, which is only responding to steroids, and that this is only her right eye. Her left eye “does not want to cooperate”. Another member has dry eyes in the morning and uses OTC eye drops, and this helps.
- The member who is on V said that she hadn't had fevers since she started the V, and her bones don't hurt much; but her shoulder is hurting, as well as the balls of her feet. She can't walk barefooted. She was diagnosed with ECD about 6 months ago, but has had skin lesions for about 12 years. The doctors had always said that they were reticulohistiocytes. When she was found to have lesions in her brain and odd looking thigh bones on X-ray, she was diagnosed with ECD.
- One member said that she had initially been diagnosed with another very rare condition, Rosai-Dorfman syndrome.

- One had been asked by her dermatologist to be part of a conference. All the doctors came in and looked at her. Then, when they talked afterwards, one of the doctors said that he had seen a case like this before, with the brain lesions, cysts on the lungs and the irregular bone marrow throughout the body.
- And the other member had been part of a "show and tell" twice, at the University of Louisville because the "smart" dermatologist thought that she had multicentric reticulohistiocytosis (ANOTHER very rare disease). She had kept telling the doctors who saw her that her joints and legs really hurt.
To summarize these journeys towards a diagnosis of ECD "we could write a book about wrong and unusual diagnoses".
- All three of the chatters will be going to the ECD convention in October in Houston. One said that she grew up in Houston, so is excited to go back. The Houston Zoo is just up the street, and she plans to go there on Sunday, because she grew up going over there, and hasn't been to that zoo for at least 45 years!