

Summary of ECD Global Alliance Internet Chat **August 11, 2018**

6 Attendees

- A new chatter came on today. She has been treated for ECD since being diagnosed in 2012. Six-months ago an ECD specialist at University of California, San Diego (UCSD) ordered her first PET scan. She only moved to and began treatment at UCSD last year. She was told at that time that both her femurs and tibias were affected, as well as her heart, kidneys, ureters, and abdominal wall.

Since the first PET scan, she developed increasing bone pain. She had a follow-up scan a month ago with some discouraging results. Her bone involvement had nearly doubled, and the disease has also infiltrated the bone marrow in her pelvis and throughout her spine. There is also some hypermetabolic activity in the tissues of her left thigh.

Her oncologist at UCSD is coordinating care with a local oncologist because of transportation issues. She has been placed on Mekinist and stopped her previous treatment of Kineret. She had been in Kineret for six years. She was on 100mg injections daily and improving until she hit a plateau after three years. She remained stable for three more years. The disease then progressed over the last six months. She will be having weekly blood test while on Mekinist. She also has diabetes insipidus (DI), for which she uses desmopressin spray. She had symptoms of DI since she was three years old but was only diagnosed when she was 27 (in the middle of her second pregnancy). She is now 35.

After years of hospitalizations and blood test, she has next to no usable veins, and is keen to have a venous port inserted for test. She asked if anyone else on the chat had port access, if so, what does it feel like when it is accessed for blood. She has never had a port and not sure of what to expect. She asked whether members had any measure of success in the use of Mekinist and being BRAF negative. She has found that ECD can be very isolating. She has had signs of ECD since childhood, but no one knew enough about the disease back then to find a connection. Just a year after she was diagnosed she lost her husband (to hospital negligence). Her daughters were two and five at that point. Then last year, she lost her father to liver failure.

- Another member new to the chat came on. She has been a member for two years, since her diagnosis. She lives in Holland and sees a doctor there, but gets advice from Prof. Haroche in Paris. She visited Prof. Haroche once, just after diagnosis. She found out about him through the ECDGA website and asked her local doctor to contact him. She is BRAF positive and taking dabrafenib (D) and Mekinist. Her bone pain stopped when she started taking the medication. Her main complaint now is tiredness. She has also suffered from Diabetes Insipidus (DI) since 2007, for which she takes desmopressin tablets.
- A French member said that Dr. Haroche had confirmed his diagnosis in July 2016. He had previously been seen in Strasbourg. He goes to Paris every six months. He has had ECD in the legs, lungs, and has DI which is treated with desmopressin. He is BRAF positive and had been treated with Kineret for two years. He still has a lot of pain and is very tired. His Kineret dose has been doubled over the last three months. He is on Kineret rather than the other medications used for ECD, to avoid the recurrence of a depressive illness that he had about 15 years ago. His BRAF status was only discovered three months ago. It has taken several biopsies to get the result.

He said that though the disease did not show on his face, does not mean that he isn't sick. The pains are real! He thinks that some of his pains are starting up again in his fingers and feet. He has also been feeling sick. He goes out into the garden and finds that the plants make him feel better. He gets a lot of cranial pain and he remembers that he saw many doctors that could not find much relief. He doesn't have much on the way of skull lesions outside the pituitary stem.

- One member mentioned the use of medicinal cannabis for symptom relief. Another member said he "wanted to keep a freedom to think and drive as long as I can without turning [to] people on the street."