Summary of ECD Global Alliance Internet Chat on
12April2014

7 Attendees

- A new member came on. She was only given a diagnosis of ECD on March 14th. She is from New York, and was diagnosed in Rochester. She started with swollen legs, and the hospital thought that she had lymphoma. She has lesions on her kidneys, lungs, heart, her limb bones and her tail bone. Recently her jaw has been affected. She has white patches on her eyelids, and "pockets" under her eyes. She has "tissue" behind her eyes, and this has caused problems with vision. She has also been troubled with drinking lots of water (up to 5 GALLONS a day!!).

  Before the ECD was diagnosed she had been suffering from Post Traumatic Stress Disorder, having been held up at gunpoint in 2001. The excess thirst had been put down as psychogenic polydypsia. She has now been tested for Diabetes Insipidus. These tests show that she does have this, but is not on any treatment at the moment.

  She started Prednisone 3 weeks ago, and the first 2 were "wonderful", but she doesn't "believe that it's working at all now". She would like to stop taking it all together. She does have the BRAF mutation. This was only confirmed after countless tests and biopsies.

  Interferon, 3 times a week, starts on the 14th. It has been difficult to get this all approved by the insurance company. Her Doc has said he won't know whether it is working at all, for 6 months (One member mentioned that interferon had been difficult for him to take, whereas another member said that she had not had any difficulties at all. It just hadn't worked on the ECD!). She had a PET scan this week for a baseline and will have another in 6 months, to see if there has been any change.

  She has just applied to be seen at the Sloan Memorial. Another member has met the doctors there, and thought that they were very good.

  It was suggested that she might contact Dr. Estrada-Veras, at the NIH (estradaverasji@mail.nih.gov). The NIH study is focused on learning more about ECD. ECD patients go to the NIH hospital in Bethesda for 1 week, and lots and lots of tests are performed. At the end of your stay, the results are gone over with you. They will send the results back to your doctors. The NIH staff sometimes work with Sloan-Kettering. There is NO cost to the patient, or to the insurance company. The NIH has historically paid for travel and food from anywhere in the US, and lodging for the ECD sufferer, and a travel companion. The study is open to all ECD patients, no matter where they may live in the world.

- The Sloan-Kettering team (Dr. Eli Diamond, et. al.) was given a $50,000.00 grant in 2013 from the ECD Global Alliance for ECD research.

- A member who has started vemurafenib told us that she had had "enormous results in just 3 months". Prior to this she had been on interferon for 2 years, but that had made her feel tired, and not better.

- The ECD Global Alliance patient/family gathering in Sept was mentioned. Everyone is encouraged to go.

- A member is going to find out, soon, how she is doing. She has tests all lined up for next week. She and her husband, who has also been ill, are both doing better.

- A member came on, and apologized for being late. She had been in a Go-Kart race with other students from her school! There were 36 drivers signed up, although they weren't all there. She had come second!

- A member had recently been out to dinner, and when going to a bar afterwards her boyfriend got stopped by the doorman. She had been holding onto her boyfriend as they walked, and the doorman thought he was too drunk, because her bad walking made him walk badly too.

- A US member has met the widow of an English member. She has recently moved from the UK to the US.

- A member is shortly going to the NIH to have her wisdom teeth removed, and they will take a bone scraping as she has irregular bone marrow in her jaw.

- Members mentioned some problems they were having with muscle cramps. It was pointed out that this may be caused by abnormalities in the blood levels of things like potassium. This can sometimes be treated fairly easily with supplements, and so it is worthwhile to mention these symptoms to the doc.

- Members mentioned their visual difficulties due to the ECD. These were, quite commonly, the reason why members had stopped driving.