

Summary of ECD Global Alliance Internet Chat

10 Jan 2015

7 Attendees

- A new member came on before the Chat. She had also come on last week, and asked whether anyone had balance issues due to disease in the "cerebral cortex". Many of us do have problems with our balance. This is due to disease in our cerebellums. This is the part of the brain that controls muscle coordination and balance.
- Another "newbie" came on before the Chat. She has a number of medical problems, and the doctors have now mentioned ECD. She has been diagnosed with hypercalcemia, Paget's disease (a high bone density), and now maybe ECD. She has seen the videos of ECD and it was frightening. She hopes that someone can tell her something about this disease.
- Two European members discussed the stormy weather that they are having. LOTS of wind and rain, but no serious damage.
- The UK member had a check-up appointment in the next few days. His main doctor had been to the Bethesda gathering last year, and the Norwegian member had been briefly introduced to him!
- Another UK patient has recently seen this doctor. She spends most of her time on a Greek island, and flies back to the UK for doctor visits. When the doctors tried to get her seen by one of the junior sidekicks, she said that she would "prefer to see the Prof himself, because she had come quite a long way"!!! And it worked because she did see him!
- The Norwegian patient has been on vemurafenib (V) for more than a year, and is feeling well. She was diagnosed in 2011, after 5-6 years of searching for an answer. She is due a check-up next month. She has an MRI scan every 3 or 4 months, and she has had a couple of PET scans. Everything is stable, and the disease is not active. Her walking is a bit unsteady, and she has bad balance, and really stiff legs, and Diabetes Insipidus. She told us that she was slowly getting better. Before she started V she was just getting worse. She has made great progress this year. She had been thinking that she was on her way to needing to use a wheelchair, but "now the tables have turned!"
- A member came on who is being treated with V by Dr. Diamond at Sloan Kettering. He has just been to Sloan for scans, and these show that the tumors in the left orbit have shrunk a bit. His vision was very bad, and getting worse before he started with V. He was diagnosed last May (although he was originally misdiagnosed as a case of Lymphoma). He has very extensive involvements: in the orbits, brain, other parts of the head, spine, and all the long bones, joints, etc. V is taking care of all these areas. He had dramatic improvements in the first week with the highest dose. Then he needed to cut down, but he continued to improve over time.

- A member, who also goes to Sloan Kettering, is due some tests in a few weeks. She also has lesions in her orbits and these affect her vision. She went in July of 2014, and is now going back to see how the Anakinra has worked. She has been on it for a little over 4 months. She is BRAF negative and could not go on V. Anakinra has done nothing for her sight. "We'll see if it's improved anything else." The drug did improve her symptoms, though.
- Members discussed how the doctors tell that the disease is inactive. This is done using the PET-CT scans.
- A member who really does have lymphoma AND ECD has had a report on her lymphoma treatment. It was a good report and there will be no more chemo for now. She had been on R-CHOP chemo for the Lymphoma. She is seeing Dr. Janku in the next week to see what is going on with ECD. She is BRAF+, and was diagnosed with ECD about 3 years ago. She was doing well on Anakinra before the lymphoma was discovered.
She was asked whether she would be considered for V. She doesn't know but sees Dr. Janku in the next week.
- The urine test for BRAF was mentioned. We learned that the test is very accurate, and has a direct positive correlation with one member's clinical results. For example, when his sight was improving rapidly, the BRAF counts were dropping very fast. He had to do 15 samples, in the first 5 days on V. They were trying to see how the levels linked to scans.