Summary of ECD Global Alliance Internet Chat
10 May 2014

7 Attendees

- A number of members looked in during or after the Chat.

- A member left us all a message. She has been trying acupuncture as a treatment for her back pains. The acupuncture seems to help a little, but not all that much. She now has had some metal studs inserted in her ear "to send currents into my nerves to circulate everything". She is away this week, but, hopefully, she will be back for next week.

- A new member came on the Chat. His wife was typing for him because he cannot see well. He was diagnosed in March, and has just turned 54. He was originally diagnosed with another rare disease, IgG4 sclerosis. Two months ago they spent an entire month at UCSF in San Francisco. The docs did "every test imaginable" and finally diagnosed ECD. He tested positive for the BRAF gene so he is now on week 8 of Vemurafenib (Zelboraf). He has tumors in both orbits that tested positive for ECD. Before diagnosis they got very large and caused his eyeballs to protrude. He had gelatinous fluid in his eye that looked like Jell-O. The optic nerves were being compressed to the point of almost losing vision. Another biopsy was taken and this allowed ECD to be diagnosed. His vision has improved dramatically on Vemurafenib. The disease is very present in the bone marrow of his legs, so they are very weak and sometimes shake uncontrollably. He is on a medication called Tegretol (carbamazepine) for that. This is in the class of anti-seizure drugs. A friend gave him the nickname "Wheels" since he gets around on a walker. He doesn't like it to be called a walker...so the family calls it a rollator. (Another UK member said that he calls his walking frame his "go-faster Zimmer" because it is a 3 wheeler!). He has other tumors in his body as well. The disease is affecting his balance. The Vemurafenib makes him very, very tired. His wife asked whether any of the other Chatters were on Vemurafenib. None of the members on the Chat were on Vemurafenib. Many of the members who have been on Vemurafenib have been so much better, that they go out partying on Saturdays! One didn't chat a fortnight ago because she was out go-cart racing with her workmates!

- We have a member who plays trumpet in the Houston Symphony Orchestra. Kathy has been to one of their concerts, and said that it was wonderful.

- The mother-in-law of one member has recently died after a long illness. The member's wife is going to stay with her father until after the funeral, and the younger sister of the member is coming to stay and help (="police") him while she is away.

- Another member also uses a rollator. She can walk without it, but it makes things easier. She lost most of her vision due to ECD. She had a tumor on her heart which caused her to collapse. Her blood pressure dropped to 70/40 with a resting heart rate of 148. Her optic nerves were suffocated and she lost most of her sight. Her balance is not good, but her ECD is in remission now. All of her tumors are gone, or are scar tissue now. She only takes a small dose of DDAVP for diabetes insipidus, and takes Keppra for seizures. She has speech issues, especially when she's tired. She was

BRAF tested, but doesn't have it. Her treatments were: Prednisone, then chemo with 2cda (cladiribine) and then interferon. Her doctor is very happy with her situation now.

- A member said that he takes methotrexate once a week. This is an "old" chemotherapy drug. He has been stable for some time (but still wobbly and slurry), and he can't work. He used to be a family physician, but had to retire when he was 45. He has just turned 52.

- One member is on Gleevec, a pill, and seems to be pretty stable. His balance is not the best and at times his speech is not so good. This week his hands have not wanted to work very well. This member went to the NIH in March for his 3rd trip. In April he saw Dr. Janku, in Houston, and on the plane home he got Chicken Pox. A light case, but he missed a week of work.

- A member told us of their family’s links with an NFL team.

- A chatter asked about skin cancer and its association with Vemurafenib. She has a 30 year old female friend who is unwell. The friend has been told by her doctors that she has ECD, and he wants to get her tested for the BRAF mutation. There was a discussion about the clinical studies enrolling ECD patients to evaluate a urine-based test to diagnose for the BRAF mutation.

- A member came on who has just started her first course of treatment with interferon. She said that she had had no bad effects. It has not helped so far but “it’s only the beginning”. She spent a few days in hospital last week, to confirm the ECD and left with the treatment of interferon scheduled for 6 months. She had a tumor in her nose which was CD68+ and Cd1-, but no symptoms. Now, she has pains almost everywhere, and especially in the maxillary bone. She had the tumor removed in September but it has grown back. She is supposed to go back to hospital for more exams (they want to check the hormones). She wondered whether ECD is something one always has to live with once diagnosed, or is it something you can get rid of.