

Summary of ECD Global Alliance Internet Chat **15 Aug 2015**

12 Attendees

- A message had been left after the previous week by a member who had to miss the Chat. She had been due a PET scan, but it was denied by BC/BS (Blue Cross, Blue Shield). She did have a brain MRI, which showed that the lesions in her cerebellum and brainstem have not changed with the vemurafenib (V), after 3 months. Her doctor is going to appeal for her to have the PET.
- The member who had a poor excuse for missing last week came on before the Chat and had ANOTHER poor excuse for this week too! He said that he had spent 4 and a half hours at his local Ataxia Self Help Group, and that he had "had enough for today".
- Baseball games and team allegiances were discussed. One of the baseball watchers tore herself away to tell us about her husband's situation. He has a "working diagnosis" of ECD, but is waiting to get it confirmed. He possibly has "hairy kidneys" and changes in his brain stem. While they are waiting for information, they find that it helps to read the stories of other people, as there are a lot of similarities to her husband's. The abnormalities in his legs (seen on X-Ray) are the only thing that is confirmed to date. Another member commented that "if he's like most of us, it's elsewhere as well"! Things have been adding up towards ECD. There is now the presence of the hairy kidneys, as well as the brainstem changes. These have been around for 7 years causing ataxia. It was said that a CAT, MRI, or a PET scan should give them the information. Also, a biopsy can be done to see whether or not he has the BRAF mutation. He hasn't worked for the last 7 years, and they had to fight to get the Canada Pension Plan Disability for him. This finally came through last year.
- A US member has a similar story of his fight for Social Security Disability in January 2005. He didn't get it until August 2008! Members wondered whether this delay was because ECD was not considered to be a disability, or that it was thought that patients might improve. He thinks that it was due to the lack of information on ECD; the peculiarity of the symptoms, and its rarity. So, he had to jump through all the hoops to get it approved. He was diagnosed in 2005, and has the disease "everywhere". It is in the long bones, above and below the diaphragm, in the brain, and it affects his pituitary gland. He said that with ECD, all of us have some of the same symptoms, AND then we have some symptoms that the doctors have never seen before. This member has been on V since February 2015. He has noticed some improvement, but not as much as some of the others on V. He takes 4+4, and it was remarked that this is a high dose to have been taking for such a long time.
- A woman, whose son has ECD, said that he also has hairy kidneys. Ten years before he was diagnosed with ECD, his details were sent to San Francisco, and they said that he was a healthy male with hairy kidneys. Ten years later they said that he had ECD. The family

was "thrilled" to finally have a name for his illness. By then his speech was getting worse and his balance was horrible. He is now on V. He is still working and is a social worker for Child Protection.

- The member whose daughter has ECD came on. He reminded us that she is now in "remission", and only takes a small dose of DDAVP for her DI. Her main difficulties are her balance, and the loss of sight that she suffered. They are away from home for the wedding of a friend of their son, and then a road trip ending in Jacksonville. The son is a police dog handler and there is a convention there. The dog is there too, and the motel is "loaded" with other dogs since there is a big dog show across the street.
- Our member who has moved to San Diego and started to see Dr. Kurzrock is now taking a dose of 3 in the morning and 3 in the evening of V a day. He had been on 2+2 pills but Dr. K thought a lesion in his spine may be relapsing so, to be safe, he is "back on 3 pills". In himself, he feels pretty good!
- Sun sensitivity was mentioned. It was said that when you're on V on a sunny day, nothing can block the radiation onto your skin when you're outdoors. Sunglasses are very important and one member often uses a pair of amber protective glasses. Another member agreed about feeling the sun through long sleeves, but he usually doesn't get burned, unless his skin is directly exposed to the sun's rays.
- There were a lot of chatters who had had experience of school life. Some were, or had been teachers, and others had been aids. A fair few also had relatives who were teachers. They all said how much they had (NOT) loved marking essays!!
- A member came on after we had finished. She was sorry to have forgotten the Chat. She has been doing well, other than a car accident this week where she was rear-ended. Her leg is slightly sore, but the doctors said it is just a muscle strain.