# **Summary of ECD Global Alliance Internet Chat**
# **26th April 2014**

10 Attendees

- There were at least 3 members on the Chat who were on V, and all three said that it was doing very well for them. One member, who had bad reaction to V is now on a reduced dose.

- A new member came on who is from Turkey. He has been ill for 2 years. He has had pains in his leg, breathing problems, and has a heart problem. He asked a member how much the disease affected her walking ability. She told us that her sense of balance was really bad, and that her legs were stiff. A "bad combination for walking"!

- A few of the other members, on the Chat, had also had heart problems, and this had helped the doctors to come to a diagnosis. The new member lives in Turkey and has some relatives in England. It seems that he isn't on any treatment at the moment. He also has not been tested for the BRAF mutation. He asked how he might get this test done, and where. It was suggested that he should get in touch with the ECD knowledgeable docs in the states, and that they can then help his doc to send any samples to where they will be properly tested. It was also suggested that his docs could get in touch with some of the European experts, for instance Dr Haroche in Paris. Another route would be to go on the ECD website and get in touch with Dr. Estrada-Veras. He can then give some information about European contacts.

- A member told us that he had not been on the Chat recently because he has been without an internet connection, but hopefully he will be able to keep connected for a while now. He goes to the NIH next week and this will be his first visit. He has been trying to organize this since before the San Diego conference. He has had a lot of problems affecting his legs. He has fractures at both ends of one of his tibias (the bone in the upper leg). This has been caused through lack of blood supply; which, in turn, is caused by his irregular bone marrow. He is wearing a special boot at the moment. It was this or a wheelchair! Now his other leg is causing problems. He needs to use a walking cane, and always uses a cart when at the mall. He has had a hard time getting a MRI of this leg; but at least it should get scanned at the NIH. It took 2 years, and 2 biopsies to get him a diagnosis of ECD. He also has Diabetes Insipidus. His leg problems prevent him from exercising, and his insurance company has been giving him a hard time about his pain meds. He started the big V but had to go without it, for almost a week, because they were playing games. He developed acute pancreatitis (a very rare side-effect) when he started V, and is now taking 3 and 2.

- A member who is now on V told us that the results of his 3 month scans, and lung tests were "amazing". His pleural effusion has reduced in size from 26 mm to 20 mm, his lung capacity has risen from 51 to 82. Lung diffusion up from 53 to 73 (this is a measure of how well the lungs absorb oxygen). "In short .... I can breathe." His cardiac MRI took 2 hours, as his was such an interesting case! The doc who did it said that it was the worst heart he had ever seen - covered in growths and ECD involvement. Professor Peter Hillmen is "the Hero who has saved me." He is applying for funding to become the specialist Cancer care centre for ECD in the UK. The good news was spoilt a bit because he had some Transient Ischaemic Attacks (TIAs) last week. A TIA is a stroke that only has effects lasting a few minutes. He knew that he has had a high blood pressure for years. When the ambulance came it was 212/174!! He thinks that the raised blood pressure is probably due to the desmopressin treatment that he takes for Central Diabetes Insipidus (CDI). CDI is caused when the hypothalamus gland in the brain, doesn't cause enough ADH (antidiuretic hormone) to be released by the pituitary gland (also in the brain). Nephrogenic DI is when the kidneys don't respond to the ADH that the pituitary releases.

- Another member has CDI caused by a thickening around his pituitary gland, and he doesn't have any ADH production. He uses a desmopressin nasal spray and, so far, after two years use, his blood pressure is still good. The nephrologist says that the spray is the best treatment. Just occasionally he gets a burning sensation in his nostrils.

- A member who is about to go the NIH asked about whether taking someone else was necessary. A member, who has already been to the NIH, said that her mom and sister came with her. They stayed in a nearby hotel, and she was, pretty much, by herself all day at the hospital. She was busy with lots of appointments. They had wheelchairs, and helpers, that get you from A to B.

- A member has just had acupuncture to see whether this might help reduce the pain. She will let us know how this has done next week. She has also bought some hot packs and cooling mats to try.