Summary of ECD Global Alliance Internet Chat
June 21, 2014

7 Attendees

- A member has posted a pic on Facebook. This has been seen by another member and he congratulated her. She has been doing well on V (vemurafenib), although she has had a lot of joint pains in the last couple of weeks. She is due for her 6 month check-up and an evaluation of the effect of V very soon. She has had quite a busy week with gatherings before summer vacation, work, PT group, and visits to other cities. So she is tired and home alone tonight. Yesterday was a pain free day, but she woke up with pain in her elbow. She said that "at least my legs are working". She walked 3 km on the treadmill today and yesterday. Her improvement is slowly progressing, "just have to be patient!" She is able to walk around at the hospital without an arm for support, or her walking sticks.

- Our woodturning member has, at last, managed to get out into his shop and do some work. He told us that, for about the last month, his shoes have got tight and he needs a shoe-horn to get them on. He also has constant pain in his lower legs and feet, although this is not too bad until they are touched. He was asked whether he was maybe retaining fluid. It was suggested that he might try to go up a size in shoes. This might be more comfortable.

- The wife of a member has had an injured foot, and then a bad cold as well. He said that she ALWAYS has things happen to her, and if it's a cold or flu, she'll get it worse than any of the rest of the family! (She is not an ECD patient.)

- A member is having a lot of pain in the bone of her left leg. Her Doc has checked for a blood clot but she is "ok there". The ECD is in all her leg bones and in her arms, too. Dr. Janku has been out of the country, but she has an appointment with him in two weeks. She is going to go to the NIH on Aug 18.

- Some members told us that they were "getting excited about September in Bethesda!" Some train tickets have been bought already, and hotel reservations have been made.

- In the previous week, Kathy told us that she had been in San Diego at a Drug Information Association conference. It was a conference that was devoted to the process of getting drug approvals. There were many opportunities to tell people about ECD. The DIA organization "paid her way" because they wanted the voice of patient advocacy groups at the conference. She also got to meet with Dr. Kurzrock while she was in San Diego, and also with a representative from Trovagene (the company that is developing the urine/blood test for BRAF). They hope to use this test to monitor BRAF mutation status, to see how well the V is working. Research is beginning to suggest that the urine might be used to monitor BRAF mutation status. The teams from MD Anderson and Sloan-Kettering have both published early papers about what they are seeing and it looks promising.

- A member told us that some researchers believe the incidence of the BRAF mutation in ECD patients may be higher than originally thought. No one really understands why some ECD patients are BRAF positive and others are not. It is the BRAF mutation that causes cells to increase on their own, which is the essence of ECD. These researchers think that if a test doesn't reveal the BRAF mutation, then there is something wrong with the test, and believe a more refined test will show the mutation in more patients. Some researchers wonder if there is perhaps another mutation occurring, that can also lead to ECD.

- A member asked us whether anyone has grown taller or had their feet grow! She has always worn around a size 10, and all of a sudden, 2 weeks ago, none of her shoes fit, and she is now a size 11 or bigger!! She has also grown half an inch taller!! Her doctors have no answers for her. She has been joking that she is going to have to special order her shoes now, or just become a clown.

- A member who has been having back pain asked for advice. She is using a TENS machine and takes a number of medications. She has already been to the NIH, and is due to go again next month. There will be a pain management specialist talking in Bethesda and it is hoped more will be learned about pain management techniques.

- We were also asked by a member, whether we had any nerve damage in our feet, legs, and hands. Some ECD patients do have nerve issues. It was suggested that she might benefit from talking about this to her doctor. Checking vitamin levels may be helpful. If some of these are low, then they can cause neurological problems, and some patients take vitamin supplements for this reason.

- A member who started V in mid-January was asked whether he was seeing any improvements. After about two months, he noticed some definite improvement in that he was urinating more, suggesting that the blockage of his left ureter was being reduced. However, he has not noticed any further improvement.

- The aims of the Alliance were discussed. They were "boiled down" to: support of patients and families (e.g. this chat line), research into the disease, and raising awareness of the disease. It was said that the community is making great progress, but it is never enough, and there is always more to do. We do seem to be moving forward thanks to patient involvement, generous donors, and caring medical professionals.