

Summary of ECD Global Alliance Internet Chat

7th Sept 2013

7 Attendees

- The Chat started with some comments about the weather. In the US, one member had had a lot of flooding, and one was having good weather. In the UK, the fine weather was coming to a close. Today, one town in the South of England had a storm leaving a layer of hailstones one inch deep!
- The son of a member is cleaning out his house to rent it out. This house is in Florida.
- Members discussed their "wobbliness". One is fairly unsteady, as usual, but hasn't had a "proper" fall for quite a while (it'll happen one of these days!). Another member said that he was wobbly too.
- A member told us of some "exciting" events this morning. He had started to get out of bed, and was sitting up, when his head swam so badly that he just fell back in bed. He is on at least 7 drugs, and he doesn't know whether one of these might be to blame.

Members agreed that "At least the bed is soft!" and that "That's the place to fall!" The floor is not a good falling down place. They also agreed that a "nice soft lady" would have been nice, but the member's wife was in the den. He was on his own.

- A member has made contact with a knowledgeable doctor in San Diego, a Dr Daniels. The member has been lead to believe that this Dr works with Dr.Kurzrock. The member will need to have another biopsy to do gene sequencing, to find out whether he has the BRAF mutation. This member has noticed that, every once in a while, some stability problems occur. Vision seems to be OK, but it does get blurry every so often.
- Another member had developed double vision, and needs to have strong prisms in his glasses (or close one eye!). He couldn't carry on working as a family physician. His voice was too slurry, and the patients used to think that he was drunk! He might fall off his office chair if he leant over too much to look at something. He had to stop driving because he had some epileptic fits, just a couple of major seizures. Then he started to have partial seizures. During these, the patient can feel a bit peculiar, and others think that he has gone "vacant". Other members on the Chat had not had fits, but one told us that he had a couple of "fits", at night, where he has suddenly woken up and his legs hurt badly. He thought that he was having cramps. Then he wondered whether maybe he had had seizure of some sort.

- Members discussed their need and use of medications for pain relief. One has been lucky, having not had much pain (so far). This member doesn't take painkillers, but he does take 10 tablets a day (and 25 on a Tuesday) so he hasn't got room for painkillers too!! One member was on 34 pills a day, and more if needed for pain!!! But, this member is taking 8 vemurafenib pills a day.

Another member has needed a lot of pain relief. One of his first symptoms was extreme pain in his legs. He is on two pain killers, but his legs "scream" all the time. He has also noticed that his legs get so hot that he can hardly touch them. He has had a full body bone scan, and his legs were "lit up like a Christmas tree". So were his arms, and even the tips of his rib cage show some activity.

A member told us that he is able to go for 96 hrs on a 72 hr pain patch. But his legs "scream" at times. He has also started synthroid (a thyroxine replacement drug).

- Grinding bones were discussed. A member has a right knee that wants to "grind bone away". His doctor doesn't want him to have an operation, as he has too many other things wrong. A member has had no cartilage in his left knee for years, and this grinds. Another has a knee that goes "Crunch" when he sits down.
- The kids are now back at school and most vacations are over. One member said that he didn't know what a vacation was. Another has been retired for 6 years, and he is on a sort of "vacation" ALL the time! He was only 45, when "they" offered to "let him go".
- Members discussed disability benefits. Many had been on benefit for some years, but one said that it "Doesn't help much, except for the insurance." One member can't get disability, because he owns "too much equipment". He will have to sell everything before he can get any financial help. This is very tough and has been something that he has had to deal with for a long time. He has a 12 year old son who has cerebral palsy, and several other neurological disorders, and can't get help for him, either.
- The San Diego conference was mentioned. Members are looking forward to meeting each other. One had been diagnosed, with ECD, in late 2001, and only met a fellow patient in person, for the first time, last February, when he was at the NIH.

One member said that although she couldn't be in San Diego, herself, her doctor was going! She has to go New York every month for her supply of vemurafenib (the BRAF inhibitor drug).

- Vemurafenib, the drug that is being trialed, in patients with the BRAF mutation, was discussed. It is an inhibitor for the gene that is mutated in some ECD patients. It was developed for use in people with advanced malignant melanoma (the nastiest form of skin cancer, which is becoming more common in young people). Some

melanoma patient also have the BRAF mutation. Hopefully, vemurafenib might help ECD patients too.

One member said that he was "a candidate" for vemurafenib, but his doctor doesn't want to change medicines because he is stable on interferon. One member has been told that he needs another biopsy, for gene sequencing. Another also needs a biopsy for testing, but his doctor says that a bone marrow biopsy won't help because he is in "Remission". He needs to get worse!! We were told that this drug has had dramatically positive results, on patients in France.

Patients who are on this drug are advised to stay out of the sun, and to use sunscreen. The drug can cause changes in squamous cells, which need surgical removal, in up to 30% of patients. The doctors check for this each month. A member, who has recently started this drug, has had no rash, or other side effects.

- A member is seeing an endocrinologist, in the coming week, to discuss whether the use of testosterone implants is indicated.