

Summary Of ECD Global Alliance Internet Chat

01 Feb 2014

10 Attendees

- Before the Chat had started a new Chatter had left a message. He lives in Israel, and has recently been told that he may have ECD. A biopsy has been sent to the US for an opinion. He has been fatigued for three years, and last year Diabetes Insipidus was diagnosed. He has been on Minirin(=desmopressin) for the last 6 months. He wondered whether there was anything that he could take to help with the fatigue.
- A member, who has just been appointed to a new parish (he was ordained a deacon fairly recently), came on to leave us a message. He had a Mass to perform, and so could not be at the Chat.
- 2 members, who have both been on vemurafenib compared notes. One said that he felt a bit bloated, but was otherwise OK, after 11 days of treatment. His knees and hip hurt, and his skin is reddish. These might, or might not, be related to the treatment. He is taking a dose of 2 tablets in the morning & 2 tablets in the evening. The other member on vemurafenib has restarted it, also at 2&2 without problems, so far. When he first started vemurafenib, he developed acute pancreatitis and had to stop it until he was better.
- A member came on whose husband has ECD. They were at Sloan a couple weeks ago. Her husband had worsening kidney function and a UTI. The doctors at his local hospital hadn't done anything, but, he was due to go to Sloan for a checkup. Dr. Diamond gave him IV fluids and antibiotics, and now he is doing better. He is taking interferon and Anakinra, and is doing pretty well on them. Having a shunt through his liver also made a huge difference. He has fibrosis in the vessels of his liver. He is BRAF negative so can't have vemurafenib. He has had 3 negative biopsies for BRAF, all taken from the rinds around his kidneys. His main problem is severe retroperitoneal fibrosis. He has also had pericardial and pleural effusions. He has had many bone scans done and has changes in the distal ends of his bones. He is "exhausted all the time" and has to take a long nap every day.
- One of the members already on vemurafenib told us that he had had about nine biopsies. None of the needle biopsies gave a result. In the end he had a laparotomy (an operation to enter the abdomen) and then had samples taken from the growths around his kidneys. He has a lot of lung and heart involvement, and the marrow in his pelvis is all fibrotic, and bones have hardened in the pelvis and lower spine.

- The other member on vemurafenib had had a first biopsy that was inconclusive. He had a second one done, and gene sequencing, which showed that he was BRAF positive.
- A member on vemurafenib was told, by the professor who is treating him, that if the fault isn't in the BRAF part of the signaling chain, then it will be in another part. The member thinks that the professor means the MEK part of the chain. One of the major drug companies, GSK, is developing a new drug for that.
- A new Chatter came on. He was diagnosed a couple of years ago, and has been on interferon. They say that it has been effective. He went to the NIH in December, and his biggest issue, at the moment, is being tired. He has also been prescribed some testosterone by Dr. Estrada-Veras. A member told us that his testosterone injection (a depot form which he has every 10 weeks) was due in the next few days, and he was feeling "at a low ebb". Another Chatter uses gel packs every day for testosterone replacement, but his new insurer won't cover this. He doesn't know what his treatment will be changed to.
- A member came on who started vemurafenib 2 weeks ago. He is very excited, especially after reading the reports of success from other members on vemurafenib. He has had some swellings around his body, and is seeing his dermatologist very soon to confirm, hopefully, that there is no skin cancer. He said that the possibility of significant improvement would make him put up with almost anything in the way of side effects. He lived with all the side effects of interferon for 9 years, when that was the only known treatment. "I can handle skin bumps."
- A member said that "They are writing a paper about my heart". He doesn't expect it to be a bestseller!
- A UK member is planning to meet another UK member in the next few days. A different UK ECD patient came to visit a week ago. ECD patients are like buses and trains. None in 8 years, and now 2 in a fortnight! A UK chapter of the Global Alliance is beginning to form!