Summary of ECD Global Alliance Internet Chat on 30 Nov 2013

12 Attendees

- A European member, who has been taking thrice weekly interferon, for a short while, is having bone pain, and is off his food. ECD tissue has just been found at the back of his eyes. It was suggested that his doctors might like to contact Dr. Haroche in Paris (julien.haroche@psl.aphp.fr), Dr. Dagna in Italy (lorenzo.dagna@unisr.it), or Dr. Vaglio in Italy (augusto.vaglio@virgilio.it).

- A new member came on who has ECD affecting his internal organs (around the aorta, kidneys, liver, pancreas, and in the bones). He has Diabetes Insipidus, and a low testosterone level. His lungs are the worst affected, with test results indicating only 50% function/capacity. He has two heart valves that are affected. His treatment is supposed to have stopped the ECD in his bones, but he still get pains in the wrists, knees and ankles. He is BRAF positive. He gets the results of a pet/ct scan soon. If that shows it has spread further, then, if the hospital can get the funding, the docs are thinking of trying vemurafenib. He has just been asked whether the docs can use his scans in a medical journal article, in order to inform other doctors.

- A member told us that she is starting vemurafenib in the next 2 days. She is starting on "½ dose". She is feeling well, but her legs "don't work". They are stiff, and her balance is poor. She is anxious, but hopeful. She went to San Diego and heard the presentations given there. But, she won't be having any sunny vacations this year! The length of treatment isn't known, nor how long the photosensitivity lasts after the treatment ends.

- Another member will be starting vemurafenib in January. He will be switching insurance companies at the first of the year. He won't be part of the trial, because that would entail monthly trips to NY from LA! He will be taking it as an individual, through his doctors here in LA. However, they will stay in touch with the Sloan-Kettering researchers.

- A member who has recently had a further biopsy taken for the BRAF mutation test is upset, because no one seems to know where everything was sent, and it sounds like the proper tests were not done. The member is thinking about getting a different insurance company. It seems that none of the docs are talking to each other! It was suggested that he might like to contact the people at http://www.patientadvocate.org/ who may well be able to help.

- A member who had started Gleevec about 3 weeks ago, had to stop it. He is "swelling all over". This drug has worked well for another patient. It was suggested that his doctors might contact the doctors at Sloan-Kettering in NYC. They have an entire team researching

ECD and may be able to help. Contact Omar Abdel-Wahab, MD (abdelwao@mskcc.org). Another contact would be Filip Janku, MD at MD Anderson in Houston (fjanku@mdanderson.org).

- There were 2 UK patients on the Chat. One had an upcoming appointment in Leeds, which is quite close to where the other UK patient lives. They arranged to meet after the appointment. [This didn't work out, but they did meet up at one member's house a couple of days later, and had coffee, cakes, and lunch. This was the first time that either of them had met another patient.] There was another UK patient at that same clinic! She was also going to "come for tea". But she had a 4 hour wait, in the clinic, to see the doc, and then another hour wait at the pharmacy, for her medicines! Tea was not on her schedule after this! (This means that the Prof in Leeds has 3 patients at the moment, at least!)

- We were reminded that the conference doctors urged patients, who tested negative for BRAF, to get retested using ultra sensitive testing. There have been a number of false negative results reported.

- Last week, Kathy talked to a patient in Detroit who is joining the vemurafenib trial, through her local Detroit doctor.

- The trials based at the NIH, and Sloan Kettering, may help with travel costs. Members are concerned about having to travel from the West Coast, to the East every month.

- Sloan-Kettering and MD Anderson are currently involved in the US trial of vemurafenib. GSK is has additional BRAF-inhibitor drugs, dabrafenib and trametinib. Dr. Estrada-Veras, at the NIH, is going to be trialing these two drugs, hopefully beginning in early 2014. This will be the first trial of these drugs to ECD patients.

- It was discussed whether all the ECD patients on the West Coast might not be able to influence an institution there, to join the trial. A member said that his doctors at Cedars-Sinai, in LA, were interested in being a West Coast center of excellence for ECD. It was suggested that those doctors might contact Dr. David Hyman. He could give them the information about the trial and discuss how they might go about joining the trial.

- After the Chat had finished, a member came on. She had missed the chat because she has become very forgetful. She has been "doing alright", just feeling a little sick and dizzy, "not too much to complain about".