Summary of ECD Global Alliance Internet Chat
28 Feb 2015

9 Attendees

- A member, whose daughter has ECD, told us that she had been to see her neuro-ophthalmologist on the previous day. She only has sight in her left eye and it has been getting worse. He ordered an MRI, and is concerned that her period of remission is over. This is a worrying thought. She doesn't have the BRAF gene. Her husband is well, but he is also legally blind, and so he can't drive. It's difficult for them to get out anywhere.

- The mother of a member with ECD told us that he went on vemurafenib (V) medication in January, and is doing very well, taking 4 pills in the am and 4 in the evening. He has a lot more energy, and his body "doesn't seem as tight". He is participating in the V trial. He had been on Gleevec for a couple of years, and was doing fine. He wasn't sure whether he wanted to "rock the boat" by going on V. But he was persuaded to try and is doing well. He has had 2 episodes when the bottom of his feet itched badly, but soaking them in warm salt water took care of the problem. He leaves on Tuesday to stay a week at MD Anderson for treatment. They think that he has a basal cell cancer (BCC) on the top of his head, and will be happy when Dr. Janku checks it out.

- A member who is going to Sloan remembers talking to Dr. Estrada-Veras before he decided to go there. Dr. Estrada-Veras had said that he was starting a new trial (which would have been of the combination therapy); but, because of the severity of this member's eye conditions, he couldn't wait any longer. He is sure that the combination trial drugs at NIH would have worked for him, but at the time, there was no alternative. The problem with the clinical trials is that, say one got in a V trial, whether it worked or not, the same person would not be eligible to get into the combination trial at NIH.

- A member came on who was diagnosed 10 years ago. He had been on Interferon Alpha for 6 years, and had masses behind both his eyes, but no effects on vision. He has never been on V. He is BRAF positive and is in the NIH trial for dabrafanib and tremetinib. He has been on the drugs for 28 days now. The only side-effect so far has been fevers. He is having 4 of his tumors tracked- 2 in the kidney, 1 in the heart, and 1 in the brain. No changes have been seen in the heart or kidneys, but the one in the brain has decreased approximately 30%. MRI shows dramatic reduction in infiltration of the left cerebral peduncle, and almost complete resolution of infiltrates around his pituitary.

  Since he has been on the trial drugs, the time between needing DDAVP doses has gone from 18 hours to 36 hours. He hasn't read any literature yet where the diabetes insipidus (DI) has actually improved, so he is quite excited. He can't imagine being able to leave the house without worrying about DDAVP!

  He has to go to the NIH on months 1,2,4,6,8,10, and 12. Amongst the other tests, they do a cardiac MRI, with drugs that slow down the heart. This is done on the last day at Suburban

Hospital across the street. It is all a big time commitment, but it has "been worth every second". In the 10 years since he has been diagnosed, he has never seen any type of regression until now.

He had heard about the V trial but he didn't want to have to pay for his travel etc. He decided to wait for the combination therapy trial. He saw Dr. Scheel at John Hopkins who wanted to treat him with V, but he strongly preferred the combination treatment so decided on the NIH. He preferred the combination because studies in melanoma patients show less incidence of skin cancer with combo. He would rather attack the "dysfunctional" pathway at 2 points rather than 1 and, theoretically, there is less probability of drug resistance by adding Tremetinib. Another member said that he had heard that this combination therapy is better than V, and with less side effects.

- We also have a member who has been found to have a genetic mutation, but not the BRAF mutation, and is on Sorafenib. She is the only one on this drug, and so this is not part of a study. She is being troubled by side-effects. She has been on the new drug for about a month, but not the full dose. Her vision has been getting worse. They have not been able to establish a correlation between the eye tumors and her vision. The MRI is not showing lesions in the orbits, but the doctor said that this could be because they're small and not showing up. She is still getting macular edema, and they are assuming that it is being caused by the lesions. But, recently, when they gave her a steroid shot in the right eye, the edema went away but her vision got worse! She is seeing an ophthalmologist at Sloan, called Dr. Brian Marr.

- A member, who had had to stop Kineret while she had chemotherapy for lymphoma, is back on the Kineret. All her check-ups have had "good reports". She has another PETCT booked for April.

- The member who has started to reduce her dose of V (now at 1+1), has only been doing this for 9 days. She said that she didn't feel any different yet.

- The various costs that appear when you are involved in a trial were discussed. No health insurance is needed if you are in a trial, everything is covered. The member on the trial lives in Maryland, within driving distance of the NIH. Most visits are at least 3 days, if not 5. He tried to price the cost of the trial per patient with meds, clinical care, and travel, he guesses that the cost is at least $250K per patient! There had been another patient there, at his 1 month visit, who had come from Ireland!

- A member is going to the NIH for the ECD study in mid-March. He has just stopped taking interferon to be ready for the study, and is getting a lot of leg pain. He asked what other people took for pain, other than narcotics.

- Towards the end of the chat, a member got on who had been trying to join us for 30 minutes. He is having great difficulties with his computer. "We need to get a new one!"