Summary of ECD Global Alliance Internet Chat
May 30 2015

7 Attendees

- A new member came on and asked about eye symptoms. A reply came from a member who had had only eye symptoms for nearly a year. They would get irritated, watery, and bulged. He took prednisone to try to relieve those symptoms.

- A member said that he has been told that he is "in remission" with no active disease. He has been treated with methotrexate, an old chemotherapy drug. This was started before Vemurafenib (V) had been developed, and so has never had it. BRAF testing has never been done on him!!

- A new member is starting V this week. He is joining the trial at Sloan Kettering. His insurance company has been giving him a "hard time", but his wife's insurance will cover 100% of the drug costs. He only lives an hour away, and is under the care of Dr. Diamond. Leading up to his diagnosis, he had a biopsy of tissue around the eye, and then had a bone biopsy to confirm that it was ECD. He has masses on his adrenal glands, a rind of soft tissue on his kidneys, lesions in his bones, masses behind each eye, and infiltration of his brain. He is BRAF positive.

- Another member had also had a biopsy of tissue near his eye, but his doctor at Mayo already knew it was ECD from the MRI, before he did the surgery.

- A member who has been on the Sloan-Kettering trial has now come off it. He is moving to San Diego, and is going to get his care from doctors that are based there. He is now in remission. The member who is taking methotrexate, was asked for details of his treatment, partly because the member coming off the trial at S-K wants to find a drug (or drugs) to keep him in remission. The member on methotrexate takes 40milligrams once a week. This is in pill form and he will probably have to take it for the rest of his life, and also to keep on seeing the doctors! The pills are only 2.5milligrams each, so he has to take 16 of them all together. The drug does come at a 10mg size for use in cancer therapy, but this strength can only be dispensed from a hospital pharmacy. If you want the 10milligram size you can only get them by queuing up at the hospital pharmacy, and this takes forever! 2.5milligrams can be found at any pharmacy. He has a friendly family doctor who will do the prescription!!! He isn't troubled by any side-effects.

- A new chatter came on asking about pancreas problems. Her husband has ECD and has recently been taken off the V trial. He has a cyst on his pancreas that keeps growing, and his lipase and amylase are high.

- Another member got acute pancreatitis soon after starting V. This was stopped and then re-started slowly, after the pancreatitis had gotten better. He was then able to carry on with V.

- V doses were discussed. One member only managed to take 4 in the morning and4 in the evening for 2 1/2 days. She was then told to not take anything for 2 days, and then to restart at a dose of 2+2. It was mentioned that a dose of 4+4 is "tough". A member who has been on a V trial said that he only knew of one patient who was able to tolerate 4+4 for 1 year.

- The member who is just about to start V, asked how long it took to start working. One member replied that it "didn't take too long to feel a difference". Another member told us that within 1 week of using 4+4 V, his eye issues (bulging, double-vision, pink eyes, etc.) were all resolved.

- A member told us that she had fallen that day while taking out the garbage. She expects her butt and hip will be "black and blue tomorrow".

- In order to assess any progress while on V, you just have to record your activities on log sheets. One member had tried making her own flow chart for that day on her computer, but her computer skills are "awful". She is using the fact that she has lesions in her brain (brainstem and cerebellum) as an excuse! Her femori (thighs) and tibiae (shins), and the tops of her humeri (upper arms) are affected, and she has skin lesions. Five weeks ago she spent a week at NIH, and had lots of scans, etc. The largest lesion in her brain was 2mm too small for her to enter the new Phase 2 drug study, so she went home to Lexington, KY, found a private oncologist and began on V.

  Her bone pains have improved since starting V but she is still dizzy. The oncologist freely admits that the member probably knows more about ECD than she does. But she is more than willing to study it...and she knows about "V". She took V 4+4 for 5 doses, stopped for 2 days, and then started back on 2+2. She is now "doing ok". She takes frequent naps, and eats small meals, full of protein, each day.

- We were asked what we did in the way of work. One had been a nurse, but is now working as a secretary. There is "too much sitting"! She works 3 days a week and spends the rest of the time exercising, physical therapy and going to other appointments. She is not in a trial, but has twice been to the NIH and has been on V for more than a year.

- Another one was also a nurse. She had to quit her part-time job because of her dizziness, and the fact that her oncologist told her not to drive.

- Continuing with the medical theme, one member had been a family physician. He retired on medical grounds aged at just 45. If he gets to the age of 65 (the normal retirement age for men in the UK- although the government is increasing this gradually) he will already have been on a pension for 20 years!!

- One chatter is a business consultant. And "all of us are full time/ part time ECD researchers". Another is a second grade teacher, on medical leave until he is well enough to return to work.

- The associations of V with hair loss etc. were discussed. One woman has developed curly hair and looks "like a poodle now", after being on V for one year. A male patient lost hair, eye brows, and eye lashes. Another female had not had any changes in hair (loss or otherwise), but would "like some curls!"

- A member who has been on V for over a year, with good results, said that she had no idea of how long she would continue with V. Her doctor in Norway has no plans for her to stop.

- Nobody knows what the long term holds in the way of side-effects from taking V. Some longer term side effects may well eventually develop. One member, who had some contact with the NIH, was told that kinase inhibitors should be taken indefinitely. But also, most people think that some kind of ongoing maintenance drugs/approaches are necessary for all ECD patients, whether they are in remission or not. It is now possible to be monitored using urine or blood for

BRAF mutation measurement. It seems that V can help you get to remission, but while some symptoms will go away, others may remain even in remission.

- One member is BRAF negative and has been taking a different drug than the others. This had to be stopped recently, but she is now back on a low dose and slowly working herself back up. She assumes that she will probably be on the drug for 2 or 3 months before more tests are done.
- Members were pleased that there is a group like the Global Alliance. With such a rare illness it is great that, through this group, we know that we are not alone on this journey.