

Summary of ECD Global; Alliance Internet Chat

11 Oct 2014

10 Attendees

- A message was left by a member saying that she was doing OK. Her shoulder is doing pretty well after surgery. She still cannot move it on her own, but the pain is now tolerable.
- A member who has been on vemurafenib (V) for quite some time is due for a check-up in the next week. Before this she was on interferon for 1.5 years, but there was no major improvement. She has had no major side effects from the V, only joint pain when she was on a higher dose. Her weight is stable. She thinks that she has lost a couple pounds since being able to be more active.
- A member told us that he is about to start V. He has "a bunch of tests" scheduled to establish the baseline, to see whether he is responding. Before this he has been on interferon, which has kept him stable since 2005, but leaves him totally exhausted.
- A member has been on Anakinra for a little over a month. She comes from Florida. She has disease in her orbits, skull, long bones and left kidney. The Anakinra has helped in that she does not get fevers or night sweats anymore, but her vision is not doing well. Her right eye used to be her "good" eye, but not anymore. She gets macular edema (swelling of the tissues in the eye in front of the piece of retina that sees in most detail) and has had several shots of Avastin. The only side effect of the Anakinra has been injection site reactions. It gets red and itchy and is very annoying.
- A member came on who has not been with us for a while. He has had a particularly difficult time. He is a single parent with 2 sons, the younger of which is a special needs child. The elder is not being helpful. He has decided to "put on his big boy pants!" He lives in Nevada.
--He is not doing so well, and has major shoulder problems. When he was first put on V, it put him in the hospital with pancreatitis. He was in the hospital for a week, then on medicines for about 3 weeks. He started back on the V at a smaller dose, and then built back up to 4 in the am and 4 in the pm. A PET scan that was done last week has shown some growths around his pancreas and he has put on about 40lbs in the last couple of months. He has not had a doctor's visit since this scan, and still has 4 MRIs due before his next visit to the oncologist. He thinks that the growths are due to the V, but he will have to wait and see.
--His first biopsy was in the pelvis, to get some bone marrow, although it was supposed to be a tibial biopsy. This extended the delay until diagnosis by 3 months. Interferon did not help him. The doctors wanted to start V. They had to do a third biopsy before they proved him to be BRAF positive. So far, the disease is just in his long bones, but now he thinks that it has worked into his shoulders. He also has a thickening around his pituitary gland. This is connected with the ECD but he has not had it biopsied. He thinks that the disease is not under control, and finds this extremely disheartening.

--He does not have a support system of friends or family, and has 2 sons to care for. He is taking V, DDAVP for DI, and 2 pain killers, acetaminophen and oxycodone. All they do is take away the extreme sharpness of the pain, nothing more.

--In March, he went to NIH, and he was already having shoulder problems which have been getting worse since. This is thought to be related to the ECD. He remembers reading in the summaries, that others have also been having shoulder problems. At first they thought that he had a frozen shoulder, which is somewhat treatable. He had a manipulation done, but it seems to be getting worse again already. The NIH also found that he had pneumonia. He thinks that this has left some scarring. His pain hasn't gone away or even lessened, and his doctor is now talking about doing actual chemotherapy.

- A member who has ECD in her orbits has been being treated with Anakinra. The ECD had greatly improved although this did take a few months. She had some site reaction from the shots, but it had "gotten better". She was recently diagnosed with lymphoma, as well as ECD. This was diagnosed from a biopsy of swelling on her left leg. She has had to stop the Anakinra while she is having chemotherapy for the lymphoma. The ECD was first found in her right leg, but quickly showed up in the left. She had a biopsy of a knot of tissue on the left leg and this showed lymphoma. She was asked whether Anakinra had affected her blood counts. She said that she has had trouble with low white blood count for years, because of the medicines for her Rheumatoid Arthritis.

--She is in the middle of the treatments, and has just finished her 3rd chemo cycle (Rituximab + CHOP). She had very little side effects on Anakinra, and she may be able to start it again, even while she is on chemo. The doctors are not sure!!

--Her vision comes and goes-sometimes good, others not so good, but it did improve while she was on the Anakinra. She did not have macular edema, and never needed any of the treatments for that. She told us that her vision is getting bad again, since she is not on any ECD treatment now.

- A member came on whose father has ECD. This is affecting the meninges/base of the brain, long bones, orbit, a ring around the aorta, and abdomen. He also has Diabetes Insipidus (DI). He has been on interferon for 18 months and stable, but his doctors are concerned about side-effects. Recently his hemoglobin has fallen and he has had two blood transfusions in the last 3 weeks. He is taking a break from the interferon, and if the blood values stabilize, the doctor is thinking of using Anakinra.

--He had quite an improvement in the brain, but everywhere else is mostly the same. He is BRAF negative, but he has the NRAS mutation (the member thinks that this was the first documented case). The doctor is exploring a MEK inhibitor, which is supposed to work like V, but has not been used in ECD. The patient is "itching to try it" because he's been so ill on the interferon.

- A member told us about her 22 year old son who has ECD. He has now been diagnosed with Hodgkin's disease (a type of lymphoma) for which he was treated with the ABVD chemotherapy regime for 6 months. For the last month and a half he has been in hospital. A couple of months after his last treatment he developed pneumonia. This was thought to be due to the bleomycin he had been taking (one of the chemotherapy drugs). He has lung fibrosis and they are too

damaged for further treatment for the Hodgkin's. So no treatment until they get better. He is now in a rehab center because his oxygen needs are too great for him to be at home.

- A member told us her complicated path to an ECD diagnosis. She was diagnosed with Rosai Dorfmann Disease in 2012, and then in 2013, they said it was probably ECD. In July of this year, she had another biopsy of her tibia and this "favored" ECD. Her first biopsy was of her frontal skull, and the second one was of the tibia.