Summary Of ECD Global Alliance Internet Chat
14 Sep 2013

9 Attendees

- A message had been left by a member, whose son has ECD, telling us that he has had a 6 month check up with Dr. Janku, at MD Anderson, in Houston, TX. He was found to be positive for the BRAF mutation. But, because he has been stable for a while, they have decided to keep him on Gleevec.

- A new member came on, and introduced her husband, who has recently been diagnosed with ECD. He is not being treated yet, and his BRAF status is being checked before deciding what to use. If he doesn't have the BRAF mutation, it will be interferon; if he does have it, then the treatment will be different.

  He has, like the rest of us, had a "long and winding road" to the diagnosis of ECD. His journey began with lymphoma, moved to Sjogrens and now on to ECD. This had made treatment a little tricky! He asked whether any others had gone through this sort of story. One member has had similar problems, but now she is in "remission" from her ECD and also from her, original, lymphoma. She is not on any treatment now, except a small dose of DDAVP for Diabetes Insipidus. She was treated with 2-CDA (cladiribine).

  The new member has pain in the bones of his, legs, arms and even skull. There is bone loss in his jaw and interstitial lung disease. Fortunately, his balance is still good. He, and his wife, were asked whether they were interested in him participating in the NIH study. That would give a clear indication of what systems have been affected by ECD (all without any cost to him, or to his insurance company!). He said that he would be interested, but that, at the moment, he was recovering from a bone biopsy.

  He was told that he could call Dr. Estrada-Veras at NIH Bethesda (or email - estradaverasji@mail.nih.gov) - and express his interest in participating. Dr Estrada-Veras is leading the study, at the NIH, to try to learn more about ECD. There are also other research/treating doctors in the US and Europe that are very knowledgeable as well. Members should feel free to contact Kathy (support@erdheim-chester.org) if they have questions. Her phone number is at the bottom of all the emails. Another useful "tool"

- A useful "tool" to make connections with people who know about ECD is www.rareconnect.org.

- Another “newby” to ECD was on the Chat and agreed that there is a LOT to process. His diagnosis is only a couple of months old, and he has been started on interferon. He is yet to know anything about his BRAF status.

- It was said that “there are many good people here, and everyone is good about helping to answer questions, and share their experiences”. Oftentimes, folks exchange emails, and then email back and forth to share information. If Chatterers want to share email addresses, they should send an e-mail to Kathy giving permission for this to happen.

- Treatment of ECD depends, to an extent, on whether you are positive for the BRAF mutation, or not. Doctor and patient preferences also have a role.

- The new member asked whether interferon was usually the first line of treatment. This is the case. One member on the Chat has been on interferon since Nov 2005, and stable since 2006. This member has been recently tested, and is BRAF positive.

- Members gave details of their current treatments.

  One uses Anakrina injections twice a day. He has tried Gleevec, but didn't tolerate it, and Chemo, was also difficult. He has been on Anakrina for 3 years, and blood tests are stable. But it is hard to know how much Anakrina is slowing the disease down. Also, this patient ruptured one of his intervertebral discs last weekend. His right leg gives way, and he can't walk without a walking frame. He was due for another CT scan of his kidneys this week, but this has been postponed. His doctor is not sure whether there is any histiocytic involvement in his disc degeneration. An MRI with contrast is needed to determine this, but contrast is not good for his kidneys.

  Another told us that he takes 40 milligrams of methotrexate once a week, and has been stable for the last 3 years. Methotrexate is a chemotherapy drug from way back, and is now being successfully used, at low doses, in rheumatoid arthritis. This means that many doctors have experience of using it! It is used because it "calms down" the immune system in rheumatoid arthritis, and it seems to do the same in ECD.

  A member summarized his current symptoms; extreme fatigue, swallowing problems, loss of motor control in the right hand, and balance issues, but no bone pain.

- The topic of “remission” was discussed. For this to be the case there should be no uptake/metabolic activity by histiocytic deposits highlighted on PET-CT scan.

- One member has been told he is in remission, because a scan didn't show any active areas. He won’t be having more annual scans unless he gets worse! Many members mentioned that they had had a considerable radiation exposure due to all of the tests. Some said that they glowed in the dark, but could save money on Christmas trees!

- A member is going back in to see Dr. Janku next week to see what the "next step" will be. Her husband, who is being treated for cancer at the moment, has 2 more chemotherapy sessions, and will then have more surgery.

- A member is due to see Dr. Daniels in San Diego. The doctor is talking about having to do another bone biopsy, for gene sequencing for the BRAF mutation. The member's paperwork says that he is BRAF positive, but more tissue is needed because the samples are "too decalcified". He has had 2 biopsies so far, and Dr Daniels was talking about a needle biopsy this time.

- It is more difficult to determine the BRAF status from a bone biopsy, and, sometimes, that is the only place that can be biopsied. It is looking like more than half the ECD patients have the BRAF mutation. There are a couple of treatment options for such patients that are showing very good results. The new BRAF inhibitor treatments are looking very good, at least in the short term. If a patient hasn't yet been tested for the BRAF mutation, an easy way to do this, without cost, is to enroll in one of the studies. If a member has already been to the NIH, then they will have been tested, and should be getting the results sometime. Dr. Estrada is writing a paper asking for a grant to do a study on ECD patients and one of the BRAF-inhibitor drugs.

- A member, who is a single parent bringing up 2 children asked about support. He has not been able to find any in his area. Some members mentioned their spouses as chief support, others mentioned friends. It was suggested that he reach out to some of the support services in his community to help with his children. It was said that, it must be very hard to deal with ECD given his situation.

- It is looking more and more like ECD may be like a cancer...many of the new treatments are cancer treatments. It was suggested that he might try approaching a local cancer center.

- A member had just got home from walking her dogs. She was congratulated on still being able to do this. Good exercise activity for her, and good exercise for the dogs. She had to quit her job because of pain, and is only 20 years old. She now walks dogs, around her town, because the walking helps. She told us that she has been having some very bad dizzy spells lately, and has been given some medication to start soon. Also, she needs to increase her salt intake, and will need a spinal tap soon.

  She was asked about her BRAF status. She has not heard the result yet, but, since she has been to the NIH already, she will have had the test done. She was told that she could call Dr. Estrada-Veras if she wanted to know the result.

- A member had just arrived home, from Chattanooga, having been for a CT of his back. The member is, virtually, wheel-chair bound. He is experiencing a lot of pain in his lower back, and down the left leg. Both feet are numb. He told us that the doctor was saying that he was "just too old"! His wonderful spirit was remarked upon, but he said that his "spirit is about worn out". He was encouraged to carry on by other Chatters.

- Pain was also mentioned by another member. It is mostly in his legs, and they do "scream" at times, and also around his wrists.

- It was noted that, in last week's Chat, a comment was made that there are only two people, in the group, that are still able to drive. Of the registered members, there are more that still drive. Reasons for not driving may be dizzy spells, double vision, a history of having had seizures, and loss of the necessary coordination it takes to drive safely. Like everything else with ECD, it seems to be different in different patients.

- The upcoming San Diego conference was mentioned. So far 40+ people have signed up to attend. About a dozen or more doctors are expected to be there, from the US and from Europe. Some members, on the Chat, said that they would be going. One said that he wished that he could go, but just "can't make it happen".