

## **Summary of ECD Global Alliance Internet Chat** **July 7, 2018**

### **9 Attendees**

- A member started the Chat off by explaining why he was using a new name. The new name is something he was called at home when he lived in Columbia, SC. He now lives in North Carolina.
- The member who had his broken neck plated with titanium came on. He managed to walk up and down stairs twice this week when the "physioterrorist" was at his house. It was the first time that he had been upstairs in 11 months! He said that "you only get to break your neck occasionally" so you have to take the recovery a bit slowly. He was diagnosed with ECD in the early 2000s and had to retire when he was 44, so he has had 12 years "rest" already! The injections into his knees' nerves seem to have worked REALLY well, killing a lot of the pain and allowing more movement.
- A new chatter from the UK came on. She is still able to tackle stairs; however, ECD is "eating" into her leg bone, so she is unsure how long stairs will be her friend. She lives in Norfolk, UK. She has had steroid injections into her knee, but it didn't make a lot of difference. She had these for two years (the doctors thought that they were treating rheumatoid arthritis).
- Our French member had to excuse himself as he needs to use a translator program, and he could not tell whether it was working properly.
- A member said that "steroids destroyed my back." She got osteoporosis and suffered from ten vertebral fractures. She has progressed from wheelchair to walker, and is now walking on her own. She had to use the steroids because it was the only thing that worked on her macular edema, which was destroying her sight. Then Mekinist came along, so she could change medication.
- The "neckbreaker" has grown an "extra kneecap" on the inside of his right knee over the medial collateral ligament. This stops him from bending the knee more than 45 degrees. They are calling it heterotopic bone formation. Its VERY rare, so there is no standard treatment (surprise, surprise). Another member said that she had mentioned the new kneecap to a friend who is an Orthopedist. He said that he would love to see the x-ray, so the neckbreaker will try to get hold of a copy.
- The new UK chatter said that she has something similar. She has just finished a course of radiotherapy this week, to see if this can break it down. The doctors don't want to operate because of the blood vessels involved!  
She sees Dr. Peter Hillmen and Dr. Tal Emir in Leeds, UK. The neckbreaker was Dr. Hillmen's first ECD patient! They "go back years." The neckbreaker said that when other patients have gone to Leeds they have all met up for lunch. Some patients have stayed over at his house rather than staying at the hospital hotel. She might give that a go. His contact details are [simon.dedrick@gmail.com](mailto:simon.dedrick@gmail.com).  
His BRAF status isn't known despite all the hospital care he has had. In the last 12 years, his hospital stays add up to more than one year!! The BRAF mutation has not been tested for because he first got ill in the "bad old days" before BRAF or vemurafenib. When he had any

biopsies taken the doctors didn't know what to look for. He was the Bradford Consultant's very first Google Diagnosis! He went through interferon and cladribine treatment, got more ill and was given a high dose of steroids and methotrexate. The methotrexate is a very old and cheap chemotherapy. His illness then resolved mostly, so the doctors wouldn't want to give him the fancy stuff anyway.

- A patient who is BRAF- is only on Mekinist (trametinib) since March, and her finger nails are breaking down. Another patient has been on trametinib for a little over a year, but she has not had any issues with nails and hasn't seen it mentioned in the literature.
- The French member has been taking Kineret 200mg / day for two months. Before that he was on 100mg / day for two years. He is BRAF+. There have been no major effects with Kineret, but he had a depressive illness 12 years ago and that was the reason for trying the drug.
- A patient said that Kineret had helped her a bit with ECD, but did wonders making her feel better generally. She is no longer on it, though.
- For the new chatter, Kineret did not work. The doctors are struggling to get better drugs. She is BRAF-. Cobimatanib has been mentioned.
- Dr. Diamond's patients have seen much improvement on Cobimatanib. In many cases, the disease has resolved, so other members thought that she should do well. She said that this gives her a positive view of the treatment. She was refused funding for another drug by NHS England which would have been great.
- A member said that she has a growth under the skin on the inside of her wrist. Another had a small lump that came up fast, and it was painful and hard. She was going in for a trigger-release of a finger, so the surgeon took the other lump off too. It was a convenient time to get it taken off and it showed up only two weeks before she was scheduled for finger surgery on the same hand.
- For one member, the ECD showed up at first with skin lesions, then leg pain and slurred speech.
- For the French member, he thought that he had osteoarthritis. He retired at 57 and is now 60. ECD was recognized two years ago.

He also talked about tenosynovitis. This is an irritation of the synovial lining around joints and ligaments. It is often located in the intersections of the fingers and it is this that makes your fingers ache.

- A member is going back to NYC in September. While there, she is also seeing a Dr. Eliza Greer about her pituitary gland. She is on 1mg daily of Mekinist.
- The other chatter on Mekinist takes just 0.5mg every three days (an ultra-low dose) and is doing very well. The low dose has resolved her long bone problems. She goes back to see Dr. Diamond in October.
- A member came on who had had a fall and broken her femur where it enters the pelvis. She said that this had been very painful ("a lot of morphine and screaming"). She broke it on a Thursday evening, but did not have surgery till Monday morning, because no one wanted to do surgery on an ECD cancer patient.
- A new chatter came on asking for advice about hand/finger pain. He is taking six tablets a day of Zelboraf and is BRAF+. One patient said she was lucky not to have pain there. Another does have finger pain and Dupuytren's contracture. Her Ortho friend looked at them, and he suggested shots in the ligaments. "Not if its steroids!" she said.

A member said that the pain in his fingers was less after one year of Kineret and spa

treatments.

The new chatter also has big calluses on his feet. These will get better after the vemurafenib treatment. Other members have tried lotions, but nothing helped! One said that good supportive shoes are needed, even when getting out of bed at night. Rubber footwear made the skin worse.

- Swallowing was the next topic. A member is due for an endoscopy because she has been having trouble swallowing. She didn't pass the "swallow test." The food would get stuck in her throat and she would get some back flow. She wondered whether this was ECD related or a new problem!

It was said that some ECD medications effect swallowing. A member said that he has some difficulties, and that a CT scan shows his throat is oval instead of being round. He chokes a lot. The neckbreaker splutters and chokes, too. He needs to chew everything a 100 times! And he still has lots of "lovely" coughing.

Another member is always the last one to finish eating, because she has to chew everything down to nothing. But she is still left with a feeling that she has something stuck in her throat.

- A member said that, over the last few weeks, she has had "major chest pain" up to her jaw which nitroglycerin took away! She also had loss of clear vision a few days ago. It was fuzzy and full of waves for 30 minutes. It was suggested that she should take these chest pains seriously and get them checked out. The wavy and blurred vision sounds like the sort of eye problems that can be caused by migraine (and, just to be awkward, you don't always get a headache!!).