

## **Summary of ECD Global Alliance Internet Chat**

### **May 26, 2018**

4 Attendees

- In the days before the Chat a new chatter left a message. His 47 year old son has been diagnosed with ECD and lives in a group home in Pittsburgh. He has Diabetes Insipidus (DI) and Brain Stem Ataxia. He is on disability and Medicare. His treating doctor wants him to go to Memorial Sloan Kettering (MSK) to consult the doctors running a study. Mike asked whether there is any organization that could help out with the expenses of getting to New York.
- The Chat itself started with discussions about the weather. Putting toes in Lake Michigan was recommended.
- A member came on who had not been with us for a while. He is on vemurafenib (V) and doing "pretty well" although he is having some trouble with his hands and feet. This began just after starting V at eight pills a day, he is now on three a day. He gets his funding through the Veterans Affairs. His problem is very thick calluses on his feet and hard raised places under the skin you can see and feel.
- A member told us that she had also had problems with her feet, and then Dupuytren's contractures (when the tendons in the palms of the hand shrink) with dabrafenib. She also had surgery on both hands for trigger fingers. She was then switched from dabrafenib and then to mekinist. She said that her Anthem policy has always covered any cost, although it is a really expensive policy. "After 3K, I have no co pay. I hit 3K on January 15!" Her "next stop" is for a colonoscopy this week. Micralax has become her new "friend"!!
- A member is on Medicare, so getting hold of the medication is expensive. Although they cover the medicines, "the co-pay is huge." When he was still employed, he used to have much better coverage. There is one member, who is in remission after vemurafenib (V), who found it difficult to get the maintenance dose of one V daily covered. But he fought the case through and it is now approved.
- The topic of the next conference in Orlando was mentioned. A member and her husband, and another member are committed to attending. Another member isn't sure. The member from Norway, who has recently had a child, isn't sure either, as it will be a long trip for someone so small. The baby is eight-months-old now and doing fine. There is a direct flight from Oslo to Orlando which might be the best way to go. The member is going back to work in October if she can manage. She lost 20lbs after being pregnant, but is finally gaining some weight.
- The member who used to be a neonatal ICU nurse is going to do a little volunteering at the University of Kentucky Children's hospital. She will be helping in the waiting room.
- Members talked about the Natural History Study done at the NIH.
- One member said that she would be seeing an endocrinologist. She has gained about 20 pounds in three years, even with daily exercise and a low calorie diet. She is always off-balance. She drinks about 60 ounces of filtered water a day, and has a Berkey filter on her tap. She drinks a cup of black coffee in the morning, and unsweetened tea or water. It is hard to order unsweetened tea in the South.

When she had seen a doctor at home, her blood pressure was 200/100. That is when Dr.

Diamond took her off the dabrafenib and she now only takes Mekinist. She fell about six weeks ago and sprained her ankle. She had it x-rayed two days after the fall to be sure there was no break. There were signs of an old break there! She had to swallow her pride and start using her hiking stick regularly!