Summary of ECD Global Alliance Internet Chat on 28 Sep 2013

12 Attendees

- A member told us that she has had a spinal tap, and that this has helped get rid of her dizziness. She only occasionally feels slightly dizzy, nothing like before. Otherwise she is doing well, and has been doing a lot of work, clearing the yard, where a batting cage was, and painting fences. She wished us all a good week.
- A member told us that he had finally got to see a doctor (Dr Daniel) who, supposedly, has experience with ECD, and works with Dr Kurzrock, but not much came of the visit. Attempts were made to get Dr K to see him, but there wasn't a reply to any of the texts, that were sent. Dr Daniel said that he wanted the member to get involved in a study.
- A member has recently moved, and has "all new" doctors.
- A member is trying to schedule a visit to the NIH but needs to find someone to watch his children (one of whom has special needs). Another member said that he had just retired from foster care, and that he would look after them for a week, if they were dropped off at his place!
- A member has just returned from a vacation at the beach, and another member asked "What's a va...ka..shun?" He was told that "it's like a weekend, but every day".
- Dr. Estrada has asked the Doc of a member to get in touch with him. A new drug may be started because she has the BRAF mutation.
- A member has been stable on interferon for, almost, 6 years, and his Doc doesn't want to change treatment.
- A member heard from their Doc in the previous week, that most people that are on the BRAF medicine develop an immunity to it, relatively fast [an immunity is an allergic response, an unusual thing for a drug to cause. Maybe the Doc meant that the drug can, quickly, become ineffective]. [Editor's note: The BRAF-inhibitor trials that are being conducted will give everyone scientific data regarding the effectiveness of this treatment in both the short term and long term.]
- A member said that he had been told, by Dr Estrada Veras, that there is going to be a study involving testing for the BRAF gene mutation. Those patients who

test positive for the BRAF mutation are eligible for a BRAF inhibitor treatment study already occurring in New York at Sloan-Kettering (and other areas in the US and Europe) and another study that is scheduled to begin at the NIH after Jan. 1 2014.

- One member was very pleased to hear about a potential treatment trial opening up at the NIH, as she has been on the BRAF study drug for six weeks in New York at Sloan-Kettering. The drug, that is being studied there, vemurafenib, costs $9000/month, but the drug company pays it all, because it's a drug trial. She is seeing great improvement, but traveling to New York is financially crippling! Baltimore is just a couple of hours drive away. [Editor’s note: It may not be possible for a patient to move between studies as there are potentially two different BRAF inhibitor drugs that are being studied. If a patient starts on one trial with a particular drug, they may not qualify to enter another trial with a similar, but different drug. This would not preclude them from changing treatments, they just may not qualify as a trial participant.]

  This patient is down to monthly doctor’s visits, to check for any reactions to the medicine. In six weeks, her legs have gotten much better. They were huge from edema and it was difficult to walk. Now she is able to walk unaided, but still uses a walker when she is out of the house, because she is worried about falling. She has just passed the 50 pound weight loss mark, due to getting rid of all the excess fluid.

  She has had Diabetes Insipidus (DI) since 1990. DI is due to a lack of Anti Diuretic Hormone (ADH). ADH comes from the pituitary gland and many ECD patients have growth around the pituitary. One member said that he has had DI since 1982, and when he first went to the Doc, he was drinking 3 gallons of fluid every 24 hrs (which needs a LOT of visits, to the bathroom, to deal with). Another was up to 5 gallons a day!! Treatment is with DDAVP pills and we were told that it is an easy condition to treat. And the pills take care of the thirst and bathroom problems! DI had been present for some years before one member got her ECD diagnosis. The ECD is now in remission. She only takes a small dose of DDAVP now.

  A new member was advised to get her ADH levels checked, to see whether she has DI. Dr Estrada Veras called last week and said that she had the BRAF mutation.

  A member, who has been to the NIH, has also just been told by Dr Estrada that she has the BRAF mutation.

  One member went to the NIH 9 months ago, and still has no BRAF test result. Dr Estrada has been asking for her biopsy, which was taken in Norway, to be sent to the NIH. She has also been told that they want to do the testing in Norway but, in the mean time, nothing is happening!

- A member recommended a particular doctor, who is involved in her care. Dr Sunita Nasta is on the professorial staff at the University of Pennsylvania Hospital, in Philadelphia. It was said that she "has patience as well as patients", and answers emails and even phone calls.
- A member is hopeful that some papers may be interested in doing an article. One of these papers is USA Today!
- On the last update of the ECD website, a review article about ECD, published by Orphanet, was signposted. This article didn't mention methotrexate, which is a treatment being used by a member in the UK. The member e-mailed the author, and got replies back from Tel Aviv in Israel, straight away. The author had heard of one woman who had had methotrexate, but hadn't mentioned it in the review. Another UK patient is going to start taking methotrexate very soon.

  For a review of the article in question, please see: http://www.ojrd.com/content/pdf/1750-1172-8-137.pdf.
- A member who has been troubled with leg pains has started walking again. The right leg still hurts, but, he can do a little work with weights, push ups & crunches. The Doc thinks that he has tendinosis. His wife is now working days, so that he can't get to the Doc for a referral to sports medicine. Another member told us that he doesn't have "any cartilage" in either of his knees. He keeps his cane close by, and manages to get around the house OK. The cartilage problem isn't due to ECD, he has had knee problems since the age of 11.
- A member told us a BAD joke about walking canes. He should know better (jokes)!
- It was noted that it is only one month until the San Diego meeting. Members said that they were looking forward to meeting each other, and some mentioned their travel schedules.

  For information about the event, please see: http://events.r20.constantcontact.com/register/event?oeidk=a07e73ss7vic308c2f1&llr=vxnuk4lab
- Difficulties driving were mentioned. One member had managed to get a friend to volunteer to drive him to San Diego.
- A member is being "worked up" for significant varicose veins, which became much worse after ECD was diagnosed in 2009. She had severe leg swelling for

months before her diagnosis, and the Docs think that this may be contributing to her leg pain.

- Finally someone from Nepal came onto the Chat, but we had all gone by then.