

Summary of ECD Global Alliance Internet Chat 23 February 2013

5 Attendees

- A member new to the Chats came on. His father, 66 years old, was diagnosed with ECD in December 2012. He had always lead a healthy, active life. His symptoms and sites of involvement are classical for ECD. The ECD is affecting both legs, sacral bones, left kidney, lung linings, aorta, and just outside the brain, in the meninges (this is the largest growth).

He has been on dexamethasone since late November, and has just been started on interferon (Pegasys). The first injection was on last Friday, and unfortunately he became ill 24-48 hours afterwards. He was extremely lethargic and almost unresponsive. It is believed he aspirated food, and this lead to aspiration pneumonia. He was taken to the local ER, after consulting with his doctors in the city, and was hospitalized. His pneumonia is, now, under control, and he has been sent to re-hab to get him walking again. Hopefully he will be home this week.

On the recommendation of his treating doctor, his interferon shot, this week, will be skipped. When the pneumonia has cleared, it will be restarted. The family hopes that his next Pegasys injection goes well. He is currently on Pegasys 135 micrograms.

He has tested positive for the NRAS mutation, and the medical team want to research this. We were asked whether any of us had tested positive for the B-RAF mutation. No-one, on the Chat, was able to answer this.

The member also wanted to add that the family is very happy with the care he is receiving.

- A member told us that, they have had 3 shots of 1MU interferon a week, since Nov 2005. It leaves him totally exhausted, all the time, and these side effects had not lessened with time. Tramadol (a painkiller) had also been used for a short while, before the interferon was started.
- Another member had been on interferon since July 2007, and is taking 3 MU three times a week. Side effects include shortness of breath, and fatigue. Hopefully, this means that the drug is working! These effects have improved over time. Initial growths found were in the lower legs, mid-section, pituitary, carotids, and optic nerves. NIH found more sites in the lungs, right atrium, and humeri. The largest growths are on the Tibia & Fibula. The interferon has helped to get the growths stable.

- The member, who has been having balance problems, has had tests at the Ear, Nose, and Throat department. He had warm, & cool, air blown into each ear. Then, he had to try to track dots, of light, across a bar on the wall. This was not a problem, until he stood up! He is "still getting" his balance back. The results from this test show that the problem is "deeper than mid ear", and nothing can be done to repair it, or to help with medication. The wife of another member also had this test, and she had also had trouble standing after the air in the ears.
- A member, whose son has ECD, told us, that since taking 2CDA for 6 months, he seems to be staying stable. He has good days, and not so good days. His balance is horrible, and he walks with a cane. When he is tired his speech is bad. His teeth are a big problem. They just seem to crumble. He is now on Gleevec, and leaves, tomorrow, for his second visit to NIH. In April, they hope to match his medication to his DNA.

His mother had been a dental assistant, and he has always taken good care of his teeth, seeing a dentist, every 6 months. since the age of 3. He has had only 2 fillings in his life. Now he goes every 3 months. His teeth seem to start to crumble for no good reason. He has had root canal work, and some have been saved. The other day, he was eating a piece of candy (a Milk Dud), and a perfectly good tooth broke off at the gum line!

During his last visit to NIH, the NIH dentist had said that the hygienist was "poor". When he got back home, the dental office sent NIH X-rays, from a few years back, showing that he had been taking excellent care of his teeth. He will be visiting the dentist again at NIH this week. The new member, whose father has ECD, then told us that he was a dentist, himself. He suggested, from his own experience, the use of Prevident , and MI Paste Plus, if the caries rate for the member is high. These products should not interact with his medication, but that should be checked with his treating physician. The "dentist member" said that he had had a pretty good success with MI Paste Plus.

- The member, who has recently suffered the death of a much-loved dog, got a Yorkie/terrier mix about a week ago. The member needs a companion while his wife "brings in the bacon".