

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

**24May2014**

10 Attendees

- A member who is in the vemurafenib ("V") trial being run by Memorial Sloan-Kettering Cancer Center (MSKCC) has appeared in a video. MSKCC recently received a \$100M gift to fund a molecular genome lab to help people, and asked her to tell her story for it. The final result is heavily edited and doesn't contain a lot of the info that she hoped would make it, but she was glad to do it, if it will help others with ECD or other rare diseases in the future. A link to the film has been emailed out to all members of the ECD Global Alliance and will be made available on the [www.erdheim-chester.org](http://www.erdheim-chester.org) website in the future.

The patient said she had ECD for 30+ years, and in the past two or three years has been taking "a fast slide downhill." She spent most of 2013 as an invalid, and was unable to walk more than a few steps. She had many issues to deal with, and knew that things were serious. When she got to MSKCC the doctor gave her 2-3 months to live. With V, she feels that her "life has been given back" to her. She still has "issues" every day "but don't we all?" She had huge improvements in the first month or two, including losing 85 pounds of fluid, and everything has been stable since then. A whole new wardrobe is expensive, but "so worth it and so much fun!"

- One member on V would like to lose some weight! His doctor tells him that his weight gain is typical of ECD.
- Another member taking V said that she was "doing great". She has a lot of energy and has had some "really great" workouts lately, getting really sweaty!
- A member who has woodworking as a hobby has been managing to get out to do some. He has some projects underway. He told us that ECD has taken all of his energy and it restricts his breathing. A member, who had severe problems with his breathing, has had a great improvement since starting V.
- A member told us of her experiences at the NIH. She has been there twice. She was "so nicely taken care of" and saw specialists who knew what they were dealing with. She was checked body from head to toe. The schedule is very full, so make sure to order your meals in advance! A driver came and met her at the airport. She went out a few nights, and had a night or two at the hotel, with her mom and sister. Most days her mornings started so early that she needed to stay at the hospital.

She would definitely recommend going to the NIH to anyone who hasn't been there yet.

- The timetable for this year's conference was discussed. The Sept. 17/18 meeting is for members of the medical community, and the meetings on Sept. 19/20 will be for patients and family members. Representatives from the medical community will stay over, to present the information that was discussed in the previous day's talks. This year the Medical Symposium (on September 18) will be on the NIH campus. The room where it is to be held will be smaller than the room that was used last year. This will prevent members of the patient/family community from being there. However, there will be volunteers there, taking notes, so that we can share the information discussed with everyone.

The morning of Sept. 19 will be devoted to panel discussions with the doctors. They will be able to tell the audience what they had said, and heard, and what they are doing relative to research and treatment. They can also say what they will be doing as a result of the Symposium.

Members said that they would need some help to make all the medical "gibberish" understandable! One said that they needed it to be converted into "cornbread language". The member who used to be a family physician said that he would try to turn it into "understandable stuff"!

- A member has been having difficulty leaving messages in the Chat Room, at times other than at the main Chat. It should be possible to type in, like normal, at any time, and then it should appear. The Chat room is always open, and nothing has been set to schedule the chat. It is just scheduled so that people will arrive at approximately the same time. Anyone with similar issues is encouraged to email Kathy at [support@erdheim-chester.org](mailto:support@erdheim-chester.org).
- Our Turkish member would like to come to the conference but, in Turkey, school begins in September and "we must be at school". There were some difficulties in swapping information about treatments and investigations. The Turkish ECD patient has some nodules in her lungs, but she has had TB in the past. This is now cured. It was asked whether the ECD Global Alliance could do anything to help, such as give contact information so that the doctor can contact the knowledgeable doctors in other countries. Perhaps another doctor could confirm the ECD diagnosis. Once there is a confirmed diagnosis, then the doctors might be able to get her started on a treatment, to ensure the ECD is controlled as best as possible. There are experts who speak French, English, Spanish, and other languages. Unfortunately, neither she, nor her doc speaks any languages other than Turkish. It was suggested that her doctor contact Dr. Julien Haroche in Paris or Dr. Lorenzo Dagna in Milan, Italy. They both know a lot about ECD.
- A member has some busy days ahead. He has a PET scan one day, cardiac MRI another; regular appt. one day, dermatology another, and "kidney stuff" along the way. Another member recently had all of that in one day at Sloan-Kettering (and it was her birthday!).
- We were told that a member who has been having back pain had some injections to try to relieve it. These helped until she started cleaning her car yesterday, but there is still less pain than before the shots. She made a trip to Arizona the other week, and it had been great to see the family. The weather helped with the pain, but the drive was horrible. The pain was intense but her chiropractor helped.
- Finally a member (non-ECD patient) came on who had been out walking a puppy. He has a painful right foot due to a broken bone. It doesn't hurt much, but it's uncomfortable. It's called a "Jones Fracture", and is one of the hardest fractures in the body to heal.