

ECD Global Alliance

2013 Patient and Family Gathering Summary

November 1, 2013

The morning began with a presentation provided by Dr. Timothy Vollmer which focused on the role of diet and exercise with neurological diseases. The take home message was to optimize your dietary habits, and make a plan to stay physically and intellectually active as these types of activities can provide reparative therapy for neurological disease.

Directly after, there was a panel discussion of ECD treatment options. Panel members included Dr. Augusto Vaglio (Parma, Italy), Dr. David Hyman (Memorial Sloan-Kettering, NY, USA) and Dr. Lorenzo Dagna (Milan, Italy). The quick take home message was that if your current treatment is working, continue with it. If not, perhaps being tested for the BRAF mutation would be beneficial. If you have already been tested and found negative for the mutation, it may be beneficial to be re-tested. Current treatment options included (in no particular order):

- Interferon (still the most frequent first line treatment for most patients)
- Vemurafenib
- Anakinra
- Gleevec
- Methotrexate
- Sirolimus
- Cladribine

The next panel discussion was about ECD research and studies. Panel members included Dr. Juvianee Estrada-Veras (NIH, Bethesda, Maryland, USA), Dr. Omar Abdel-Wahab (Memorial Sloan-Kettering, NY, USA), Dr. Lorenzo Dagna (Milan, Italy) and Dr. Augusto Vaglio (Parma, Italy). Most of the questions focused around the

BRAF mutation testing. Particulars about the current studies and research can be found on the ECD-GA website at <http://www.erdheim-chester.org/ECDStudies.html>.

A round table discussion was held with the panel members and the audience was able to ask general questions about ECD. Interestingly, it has still not been decided if ECD is an immune disorder or a cancer, as it can have characteristics of both. The panel members reiterated that if you have already had a biopsy and it has shown negative for the BRAF mutation, then perhaps a second biopsy should be performed at an institution with experience in testing ECD patients.

An important outcome of the medical symposium, and was discussed during the round table discussion, was that several doctors/medical institutions from both the U.S. and Europe have agreed to work collaboratively to prepare a paper that will outline diagnostic, laboratory, and treatment recommendations on ECD. The goal of the paper is provide an updated review on the subject from a diverse and multidisciplinary perspective on the disease. This paper will be submitted to peer reviewed medical journals for publication. Once published, the ECD-GA will ensure that the paper will be available on the website. A publication of a paper such as this will make it easier for patients, local physicians, and insurance companies to understand the current status of ECD knowledge. It is hoped that this will help in many ways, including making it easier to obtain necessary approvals from funders for necessary tests and treatments.

The attendees were also able to hear about different types of non-traditional treatments as presented by Dr. Caroline Bjonback, to include acupuncture, Reiki and meditation. Attendees also learned about mindfulness and relaxation techniques to aid in the healing of the body as well as the mind as presented by Allan Goldstein and Cassondra Graff.

A break-out session concerning the management of emotional and stressful aspects of caregiving was provided by Martha Ranon.

The day of rounded out with a presentation from Cindy Livings, a spouse of an ECD patient, who guided the group on how to communicate with medical professionals and keeping your own medical records. The take home message was to respect your clinician's time but also keep in mind that you are your own self-appointed advocate. Be concise when speaking with your physician and if you have questions about your or your loved one's medical care or something doesn't "seem right" then ASK the question. Having copies of your medical records to include testing and scans can speed the process when trying to puzzle out treatments or a path forward. Keep these records up to date and available to prevent re-testing should your records on file become missing.

November 2, 2013

The day began with a personal account of having a spouse diagnosed with ECD as presented by Gadi Bachman.

Another presentation, given by Dr. Gordon Saxe, focused on the 5 pillars of a healthy diet which included descriptions of whole and unprocessed, plant based, local and seasonal and organic foods while creating a balanced diet with the types of nutrients available.

Round table discussions throughout the day concerning (1) treatments, (2) caregiver coping techniques, (3) exercise and independent living experiences, (4) spiritual life, and (5) navigating the health system allowed for attendees to share their personal experiences by communicating their story in a group setting.

To finish out the discussions for the day, the attendees had a group discussion highlighting different initiatives and activities that could better support ECD-GA's mission as a patient advocacy group. Here is a quick snapshot of the ideas presented:

- Ask medical experts to help get ECD on health news programs or other visibility media
- Send more emails with updates about the organization
- Upgrade the website to be user-friendly, have search capabilities, links, etc.
- Provide resources to clinicians in other countries concerning the diagnosis and treatment of ECD
- Every member should contact their local media to raise awareness using the hook "living with a rare disease" from a personal perspective. Tell your story, make it interesting and bring awareness to the public about rare diseases.
- Share the Medical Symposium Program flyer with your treating physician to pique interest and attendance in next year's symposium.
- Develop a lecture route for medical students, Fellows, etc. to raise awareness of the disease.
- Provide updates about ECD to the big Pharma companies via web postings.
- Make ECD information available in different languages in order to reach out to our non-English speaking members.
- Track where medical events take place and ask for volunteers to "man a booth" in their area
- Utilize social media outlets for outreach, awareness and connections
- Give your (or any) doctor a copy of the ECD documentary
- Use the ECD form letter to ask for donations
- Perform fundraising in creative ways – and share your ideas with the group!

This is just a quick summary of the topics covered in the two day Patient and Family Gathering. Most of the presenters and round table moderators were volunteers - both as ECD-GA representatives or representatives from other organizations. Many thanks go to those that dedicated their energy, knowledge and time for the momentous event!

ECD Global Alliance
2013 ECD Medical Symposium ECD Investigator
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2013 ECD Patient and Family Gathering Attendees (not all shown).