

## Summary of ECD Global Alliance Internet Chat on 10<sup>th</sup> March 2012

### 9 Attendees

- A European member left a message saying that she would be away that night, and then going to Florida for 2 weeks, so she would have to miss a few Chats.
- A new member came on from the UK, and was welcomed aboard. He has had ECD diagnosed at the end of last year. He says that he is managing. "Could be better, could be worse." His symptoms include bad balance, slurred speech, skin and brain lesions and brittle bones. He fractured his spine late in 2010. He is taking prednisolone at present.
- The NIH study is still progressing, although no more patients will be seen until the end of summer. However, lab work is being done on the first 10 patients seen. The research team is working hard, and looking at the disease from many different angles. Dr. Kurzrock and her team continue to treat ECD at MD Anderson and feel that progress is being made there. In addition, the organization has many projects on the table that we hope will help to raise awareness and set the stage for more research.
- The organization has a volunteer who is going to try to redesign the look of the website. The volunteer has requested pictures of members of the organization, that we could put on the website. If you have a picture, or two, that you would like us to look at, for, possible, inclusion on the website, can you send them to Kathy, please? There will be forms to sign before they can be used, but at least our volunteer would be able to get an idea of what might be possible.
- The member, who has had injection treatment for back problems, said that his back was "pretty good". He is going to have a radiofrequency treatment next week. This will involve inserting 3 needles on either side of his spine, and then applying some sort of current to them. This has been offered because the member is moving away.
- And another member, who has been having back problems, is also feeling better.
- A member said that her son, who has ECD, was making pizza from scratch. She was thrilled that he felt well enough to do this.
- The new member was asked whether he had seen any doctors with experience of treating ECD. No one had been seen yet. An MRI scan showed a small shrinkage of the lesion around the pituitary gland. He was asked about visual involvement. He said that glasses had helped, but that fever, in particular, affected his vision. He said that he is 38 going on 83!! He has also had to cope with diabetes insipidus. He was drinking 12 litres of water a day! He had desmopressin treatment for a while but the DI has eased. A member, who had also had DI, recommended desmopressin treatment, having found that it solved the problem.

- Yet another member with DI takes DDAVP. She had needed excessive amounts of fluids for some time. Her parent wonders whether this could have helped to get an earlier diagnosis.
- Another member was asked whether his current doctor had ever treated another ECD patient. The member was pretty sure that he is the only ECD patient in Los Angeles. The principal doctor involved is an oncologist, operating out of the hospital cancer center. He had been visited, because he had experience with interferon, which is mainly used as a treatment for certain types of cancer. The member thinks that his “main” doctor, and the others that are involved, “enjoy” trying to deal with something out of the ordinary. The doctor had spotted the more recent article about French doctors’ success with kineret in treating ECD.
- A member had been unable to go to the NIH, as planned, because she had had a dreadful week with kidney problems. This wasn’t an infection, just a lot of pain, but it has improved now.
- Our new member has had a thyroid problem for 3 years. His thyroid medicines have been slowly increased and his levels now are good. (it wasn’t made clear as to whether the thyroid was overactive or underactive). It has all been thought to be due to the brain lesion interfering with the functions of the pituitary gland and the hypothalamus.
- Other members have had both overactive and underactive thyroids.
- A few people had found that steroids (prednisolone etc) had caused a deal of weight gain. One member had had to have a hip replaced because the steroids had affected the bone in the hip joint.
- Xanthomas (yellow collections of cholesterol, usually under the eyes) were discussed. The new member has them, as do some of the existing members. They don’t affect vision.
- Other members have had problems with tumours in the orbit, sometimes causing severe problems. In one case, the optic nerve was crushed, and has caused blindness.
- We were told that Kathy had completed her Half Marathon charity run. Well done!!