

## **Summary of ECD Global Alliance Internet Chat on 21 Sep 2013**

### 7 Attendees

- The first members to log on talked about where they lived. They were from Ohio, Las Vegas, and Central Arizona.
- The NIH research program was discussed. Some members had already been; one had just received an E-mail to schedule his trip. He needs to find someone to take care of his boys while he is away, as he is a single parent. Those who have already been to the NIH, agreed that they had been well treated while there.
- Some of the members who have already been to NIH have had their BRAF test results back. 2 were BRAF positive. One member had not heard from the NIH yet. A member, whose doctor has taken bone biopsies to test for the BRAF mutation, told us that, although his stains show positive, they want to do another biopsy for sequencing to verify which mutation it is.
- The members discussed whether they were going to San Diego, and said that they were looking forward to meeting each other.
- Members expressed some sense of confusion about the BRAF mutation. It is a gene mutation that is found in about half of patients with ECD, and is also found in some patients who have malignant melanoma. It seems to be associated with diseases involving proliferations of unhealthy tissue. Drugs that are used in these conditions can cause other complications, such as skin growths that have to be removed monthly.
- The member, who is a single parent, has, unfortunately, not got any caregiver. 2 other members said that their wives were their carers.
- A member has had a low testosterone level diagnosed, and has had a testosterone capsule implanted this week. The capsules are inserted in 2 incisions the doctor makes in the buttock, and has to be done every 3-4 months. The member told us that side-effects were "a sore bum" and the possibility of infection. The low testosterone is probably due to the ECD. He has some abnormal growth around his pituitary gland.
- One member has a monthly shot, and another member uses a testosterone replacement gel. He has a thickening around his pituitary gland too. Diabetes Insipidus is also due to failure of the pituitary gland and this member has had DI for a while.
- We were told that a female member has also been found to have a low testosterone!

- Members told us about their various deficiencies found in blood tests. Low iron, vitamin D and calcium were mentioned.
- A member, who is 52, and his wife (who is 13 years younger) wish to start a family by adoption. Other members thought that this was an ambitious plan, but were encouraging. They advised that the member, and his wife, will need both "big hearts" and lots of energy. The member, who is a single parent, has raised both of his sons, himself, and "wouldn't trade it for the world".
- A member told us that he is going to write a book that will, hopefully, be inspiring for those who have been given a scary diagnosis.
- It was mentioned that ECD adds stress to a relationship. This was said to be even worse when your partner is in the medical field, yet still doesn't understand. In the case of one member, her partner is in the medical field, and this has helped, because he can act as an advocate on Doctor's appointments [a member, who was not on the Chat this evening, was, himself, a family physician before becoming ill].
- The member who has had the testosterone implant fitted, has fallen recently. He was taking clothes from the dryer, on the previous day, and "my world just fell from under me". He fell "right where doc cut me".
- One member always takes a cane when he leaves the house because he quickly becomes weak. It was suggested that it might be useful to have an assistive device, like a walker, available for when you have a rough day.
- After the Chat had finished, a member left a message wishing everyone well, and apologizing for missing the Chat. She has been having very bad dizzy spells, and had fallen that day because of them. She is going to have a spinal tap in the week to try to help, and is also getting some glasses. Apart from this, she said "I seem to be doing pretty good".