

## Summary of ECD Global Alliance Internet Chat on 12/01/2013

6 Attendees:

- A number of members mentioned difficulties with swallowing. This varied from choking when swallowing foods, to choking on their own saliva. Some of the members have had their oesophagi ("swallowing tube") checked out. No blockages have been found. One member was told that his tube was oval in cross section, when it should be circular. It was generally thought that the problem was due to co-ordination difficulties.
- A member has been to M.D. Anderson Hospital, for a 6 month check up. The neurologist said that the cerebellum, which controls balance and speech, is small. The doctor said the small cerebellum has nothing to do with ECD, and that it is an inherited physical or biochemical change. There is no treatment for a small cerebellum. She, also, said that balance and speech has nothing to do with ECD! The NIH had seen brain lesions, but this neurologist did not see any. The NIH, also said, that there were lesions on the heart, and M.D. Anderson did not see any lesions. There is a visit to NIH, the end of February, and, the member will see what they have to say about all of this. (Editor's Note: There will be follow up provided on this over time. At this time, it is thought that swallowing difficulties can definitely result from ECD and a smaller cerebellum might also be a result of ECD.)

It was discussed as to whether any other members had been told that they had a small cerebellum, but no-one had been told this.

- A member, whose wife died recently, due to ECD, told us that her autopsy spoke of slight atrophy of the cerebellum. Also, there was an ECD lesion, extending into the cerebellum, as well as into the main part of the pons, so her speech and balance difficulties were clearly associated with her ECD. She had had problems with this, during 2011, and they arose, because of the ECD lesion putting pressure on the communication pathways, through the pons. She would frequently swallow the wrong way.
- A member has seen a urologist recently. At present, he needs to get up 10-15 times a night to go to the bathroom! This is all getting very difficult for him, and he is losing lots of sleep. Some further investigations are planned for this week.
- A member has received the paper work for the Spit For A Cure research project. A call will be made, next week, to get a registration number, and then kit will be mailed out. (Editor's Note: Any ECD patient with his/her 2 biological parents still living is qualified to enter this study. All it takes is a willingness by the patient and both parents to fill out some paperwork and spit into a vial! No needles, no travel is necessary. Please email [support@erdheim-chester.org](mailto:support@erdheim-chester.org) or Dr. Ken McClain or Dr. Carl Allen at Texas Children's Hospital for more information.)
- A member said that they were part of a study, in Amsterdam, that is studying the genetics of diseases involving the white matter in the cerebellum. The member had

joined this study before getting the diagnosis of ECD. This member has also received a mail from Dr Estrada. He would like the bone biopsy, that has been taken near the spinal cord. The last thing that this member told us is that a vacation, in Florida, is planned at the end of February!

- One member has been on vacation, for the last 2 weeks, and deliberately stayed, almost, completely computer free. He had had a great time, visiting family, and swimming at the beach, and seeing New Year fire works.

(Summarizer's comment; There are some perks, after all, to living in the Southern hemisphere!)

- A member has just started a course of chemotherapy. This is being given intravenously, and takes all of the day. There was nothing to occupy him, apart from his "beautiful wife", who had been there, with him. Another member pointed out that they could remember meeting people coming in to hospital for the day for chemo. They were REALLY pleased to have to stay just for the day, and not have to spend a night there.
- We were asked whether we might attend the Symposium in San Diego in November. Most members expressed a desire to go, and to meet each other, but it will depend, to a large degree, on members' personal circumstances. (Editor's Note: If you have not already done so, please go to <http://www.surveymonkey.com/s/QMSY63P> and complete the short and simple questionnaire found there. By doing this you will be helping the organization better plan all 2013 projects. PLEASE help us do the work of the organization by completing this survey.)