

## Summary of ECD Global Alliance Internet Chat 22 Dec 2012

### 7 Attendees

- Before the Chat started, a new member from the UK came on, having been diagnosed with ECD, in August, at the Sheffield Metabolic Bone Centre. The disease is affecting the long bones in the arms and legs, and the member also has had Diabetes Insipidus, for some time. This member commented that patients feel so isolated, as it is such a rare disease, and so few of the doctors, in general practice, have even heard of it. The member asked whether there were trials going on in the UK, or any patient groups? (Editors note: No known ECD trials are going in the UK at this time, although there are studies underway in France and Italy. There are also no known ECD patient groups in the UK currently. However, with 8 ECD patients living in the UK, perhaps those members might like to try to connect. If so, please send an email to [support@erdheim-chester.org](mailto:support@erdheim-chester.org).)

Prednisolone had been used, as a treatment, for three months. The dose was tapered off at the end. This did stop the extreme pain, but left the member very weak and wobbly. A month later, the pains returned with a vengeance, and walking was very difficult. Prednisolone has been started again at a slightly lower dose than before. The benefit is, already, noticeable, and "the fat face side effects will have to be put up with."

- Many of the Chatters mentioned the current weather situation in their area of the world. The Northern hemisphere was mostly snowy and cold, and (surprise, surprise) the member from Australia was expecting a "scorcher" of a Christmas. No snow, for the kangaroos, this year!
- We were all reminded that the "Spit for a Cure" campaign is getting started, and specimens of spit, are being asked for, from patients, and their biological mother and father.
- A member told us that the autopsy examinations, of his wife's donated brain had, sadly, not shown anything definitive. The analysis had not been able to definitively diagnose ECD, but the doctors still feel, strongly, that the only reasonable diagnosis is ECD, and that the High Dose Methotrexate was effective in dramatically reducing the ECD lesion in the brain. Dr Vaglio still wishes to see her case history for his study.

An interesting comment, from the doctors who had performed those examinations, was that, there are actually a range of similar histiocyte diseases - all with specific characteristics and all rare. This makes it even more difficult to make a clear diagnosis, and, seriously, complicates the process of identifying which treatment is most effective in which case.

- Dr. Estrada would, also, like to have tissue samples as well. A member, who has been to the NIH, told us that spinal cord lesions, had already been sent to him.

- One member told us, that their son, who has ECD, is doing well, but the cold weather "freezes up" his legs, and he has missed some work.
- A member has been going "up north", with members of his family, a lot longer than ever before. They are deer hunting and he thinks that he is invited along, to "keep the cabin warm". He enjoys their company. There was a blizzard this past Thursday, and this may well affect the hunting. One of the party is paraplegic, which makes it a lot easier for him to get snowed in.
- A member then told of us of a female friend who was "house-minding" in the mountains of Colorado. She got "snowed up" for three months! But she was well prepared and provisioned (and was with a new boyfriend, and, happily, they "got along" just fine!).
- The member who, last week, suggested that an "inspirational book" should be written, to help people trying to come to terms with a difficult illness, asked us whether we thought that it would be best done as individual stories, or should all of them be brought together.
- A member told us that their nephew has Reiter's syndrome. This is a syndrome in which the patient (usually men aged 38-40) gets inflammation affecting many parts of the body. It is "just thrown in" with autoimmune diseases, and no one is doing research into it.
- A "baclofen pump" has been suggested to one of the members, who has been having severe problems with muscle spasm. The "pump" would deliver small amounts of the anti-spasm drug, baclofen, just under the skin. Administering the drug in this manner can help reduce side-effects from the medicine.

No other member had had any direct experience with a "baclofen pump", although one member had had intrathecal treatment (directly into the fluid that surrounds the spinal cord) with cytarabine chemotherapy.

- The member, who has recently been ordained as a deacon, said that he is doing lots of diaconal work.
- A member, who has been having a lot of problems with dizziness, now has an appointment to see an Ear/Nose/Throat specialist. Hopefully, something will happen that will help him regain his balance. He has also been having pains radiating down his arm. He has an appointment to get treated by a masseuse. She is fresh "out of school", and will want to make a good impression. She has already said that he will probably "need a few sessions". Lucky him!
- A member, who has recently moved apartments, is "loving it". The member was congratulated.
- Our attention was drawn to the recent e-mail asking for information about chronic ECD issues. It is hoped that members of the organization will be able to respond. This is a great opportunity for a number of doctors to hear the "patient voice". It is a chance to focus on the things that are most difficult for patients and families.