

Summary of ECD Global Alliance chat held on Saturday 25th July 2009

9 Present:

- One patient has recently started on the drug Gleevec. They feel that, in spite of some side effects they are making an improvement and have more energy; the improvement seemed to start almost immediately after commencing Gleevec. Side effects vary from day to day, but the most frequent are 'once a day' leg pains, these are usually effectively dealt with by taking ibuprofen. The abdominal pains that were present in the early days have now gone.
- One patient has had a nephrostomy, which they described as a mechanism that provides an extra way for the kidneys to evacuate. Tubes from each kidney come out of the back and these are connected to tubing leading to collecting bags.
- One member who has been struggling to get an appointment with an ECD knowledgeable doctor has finally gotten an appointment for mid August.
- One patient described that prednisone has not proved effective in treating pericarditis. They will be starting methotrexate next week and asked if anyone had any experience of this drug. There have been people in the group take methotrexate with at least one positive result. It is described in Wikipedia as 'an antimetabolite and antifolate drug used in the treatment of cancer and autoimmune diseases. It acts by inhibiting the metabolism of folic acid.'
- A patient has been taking physiotens for blood pressure for the last 6 months but has now stopped it as it is thought that it may be causing an irregular heart beat.
- A member is going to see an ophthalmologist in a few weeks and is taking some information learnt from the group. The benefits of being your own advocate when you visit a doctor were discussed. With rare diseases, the more information you have, the better able you are to participate in your care.
- Two members recently attended the Genetic Alliance conference in Maryland.
- Vision problems were discussed. One patient described their vision problems caused by masses in the eye, another patient described how they find it difficult to focus. For another patient, the first symptoms of ECD were vision problems in both eyes; the pressure on the optic nerve caused fluttering/waviness in the peripheral vision, this has now gone. One patient on interferon described how their vision has improved throughout the illness. The literature does seem to indicate that interferon works well with optic issues related to ECD. Several patients in the group have described in the past that masses in and around the eyes have decreased in size following treatment.