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Newsletter Alpha1 Awareness

A Word from the Chairman

The Alpha1 Awareness Meeting held on the 21st April was attended by more than 70 people; Alphas, families and friends. We have received many emails from those who came to the Redcliffe Rooms at the Bristol Hilton. All are complementary about the organisation and the quality of the presentation given by our guest Professor David Lomas. Professor Lomas is an excellent speaker and makes complex scientific and medical issues clear to non-specialists. He is also an amusing and light-hearted speaker and the whole presentation was interspersed with banter and laughter from his attentive audience. Members were invited to ask questions at points throughout the presentation, there were many of these and it was greatly valued by the members even though it made the talk last over two hours.

After lunch we held an open discussion session moderated by Dawn Heywood-Jones. We concentrated on a number of questions that have been posed by the European Lung Foundation ahead of the European Respiratory Society congress in Vienna later this year. The general tone of the comments was that doctors are unresponsive to patients' needs and there were concerns that more and better information was not being provided by our medical specialists.

We were very pleased that a number of members of the Alpha1 Support charity came to the event.

The young children who came with their parents were entertained throughout the day by the four trained child-minders from the Alphabet Agency. This was a great success and one young boy was heard to say 'can I come back tomorrow?'

Following our tradition, we ended by giving Alphie teddy bears to our junior guests and for those regulars who already have one, a small present.

I should like to thank Lin and Dawn for planning and managing a most enjoyable and informative day.

Finally, I wish you all a healthy and easy-breathing Summer.

Alan Heywood-Jones

This edition of our Newsletter is slightly different to our usual editions.

Following our meeting in Bristol on the 21st April, we felt it was important to give you as comprehensive a report as was possible and will be focusing mainly on that. We also are including a report on the Rare Disease UK Meetings to discuss the Consultation on UK Plan for Rare Diseases.

Prof Lomas' talk about his research is important to all UK Alphas. With the help of 2 amateur voice recordings of Prof Lomas' 2 hour presentation, our chairman Alan has produced a verbatim report, transcribing it has been a marathon task for Alan. The finished report is 21 pages long, much too long to go into our Newsletter, so an extract will be produced here and you can access the full report, at the link, given in the report on the meeting. We are very grateful to him for all the hard work to produce the full report and also the extract for publishing here.

Meeting in Bristol 21st April 2012

It was a bright sunny, if windy day, when the Board of Alpha 1 Awareness UK, met at the Hilton Hotel, Bristol, early in the morning to prepare to host what to date was our biggest and most important meeting.

Members began to arrive early and there was a real buzz in the air, as old friends greeted each other and people who only knew each other from chatting on Facebook or on the AAW Forum, were able to greet each other face to face, there was a lovely friendly atmosphere with lots of chat and laughter. While all this was going on, the technical equipment was being set up in the meeting room and the group of Nannies, from the Alphabet Child Care Agency, were busy setting up a crèche for the children, in an adjacent room.

Personally I had a real thrill when I walked into the meeting room to see around 70 of our members gathered ready to hear Prof Lomas, after a few formalities like Fire Exits etc, Prof Lomas was introduced and he began his presentation. As Alan has said, Prof Lomas is an excellent speaker who is understandable and easy to listen to.

After Prof's presentation, we broke for lunch, a chance to chat, share experiences and knowledge and generally enjoy each others company. The children joined us for lunch, much to their great disappointment, because they were having such a lovely time in the crèche, they joined their parents more willingly, once they new they were returning when they had eaten!

After lunch, there was a chance for everyone to share their knowledge, understanding and comments, in response to 4 questions, Dawn, asked everyone. These are questions that she had been asked by the European Lung Foundation (ELF) to give answers, to the patient representatives committee, at the European Respiratory Society (ERS) meeting in Vienna, later in the year. These questions prompted some very useful interesting answers, although the questions where based on COPD, they proved as ever that Alphas are very aware and knowledgeable of the things that affect their health and well being. We would like to thank Dr Sandra Nestler-Parr for explaining about the GOLD system for COPD. Dawn was grateful

for all the answers you gave, which she will take with her to the ERS meeting, as a representative of all UK Alphas.

As many of you are aware we have a tradition at the end of our meetings that when babies or children under 8 years attend one of our meetings for the first time, we like to give them a gift of a teddy, known affectionately as Alphie, at this meeting we had 3 babies who we had not met before, it was therefore a pleasure to be able to give them a teddy each, there were also 6 children who we had met before, at previous meetings, we do not like any off them to feel left out, so we gave each of them a game suitable for their age group.

There was tea and coffee available for people before they left and it was great to see people standing around chatting over a cup of tea, for almost an hour after the meeting ended, this to me was the sign of people having enjoyed the day and the company.

Thank you to all of you who were able to attend and for the very kind comments and thanks you sent to us, without you the meeting would have not have been the success it was. I know there were many of you who were unable to attend due to prior commitments or in some cases poor health; I hope the report will help to give you a taste of the day.

Lin Daniels

Meeting in Bristol 21st April 2012

Extracts from the presentation by Professor David Lomas

This is a verbatim report of some sections of the presentation. The sections of the presentation on the pathology of alpha-1, the history of it's discovery and hypotheses on the possible survival advantages and the death of Chopin have been omitted in this version. The slides used during the presentation are not shown here. Comments and notes are shown in show in square brackets [so]. The symbol -> is at a point where Professor Lomas is highlighting a picture or text on the screen.

You suffer from a very important disease it is a disease that I have worked on for 22 years I did my PhD on your disease where we described the fundamental mechanisms and my ambition is that by the time I retire is to give a talk which says:

- this is mechanism that I described when I was a graduate student in Cambridge,
- this is the pathway of the disease our understanding of the liver and lung disease,
- this is the cure and
- This is the patient that we've cured.
- Thank you and Goodnight.

That is my career plan. That is my goal and I suspect that it is aligned with your goal as well so I think we're on the same page here.

Recently as many of you will have seen we've had a big breakthrough in alpha-1. Alpha-1 has been mentioned on the Press wires and at the top of the publicity – even being on the Today Programme and in the Telegraph, the Times, the Guardian – all the big ones. Your disease was up there and to all those people who say *I've never heard of it* – just say *it's on the News*. You disease is a model for a variety of other diseases and it's first up there for a cure by genetic processes.

What I want to do this morning is to have an interactive session. I don't want to stand and give you a lecture for an hour – I can do that to my medical students at any time.

I really want you to understand what I've done and what I'm trying to do. So the questions and answers are really important in all this and what I'll do is break my talk into a series of chunks. So I'll do liver disease, lung disease, why you are so important and why carrying this mutation actually helped you a few hundred years ago. Then I'll talk about various strategies for a cure. I'll stop after each and take questions. Please, please have questions! If you think *Gosh I don't want to ask that it may make me seem a bit stupid* – it doesn't matter: I'll turn it into something sensible. So please, please engage.

I want you to leave here with a few snippets of information whereby you really understand this disease and I understand where he's going and what he's trying to do.

A special welcome to the people who see me in clinic and have been involved in some of this research – some of your blood – some of your **type** of blood. Those of you who come to the clinic on Wednesday know that the question is '*. . . and now can we take some blood for research?*' And you'll see why.

[INTERJECTION] You do get a free cup of tea!

Whenever I give a medical talk I start off with full disclosure and I'll do that for you today: so that you'll all know where I'm coming from and so I disclose my conflict of interests. It's the sort of thing in medicine so that if I say something you can say '*well hold on a bit . . maybe he's giving me a biased picture.*' It is very important.

For many years I've worked for GSK, GlaxoSmithKline, which is a major pharmaceutical company. I've been a consultant with them. I've received grants from them and three years ago they asked me to join the Board that controls the whole of drug discovery for respiratory [indistinct]. And this week they have asked me to be the chair of the board that controls the whole of the discovery pipeline for GSK.

So that's my disclosure of interest, I'm a university professor at Cambridge and I work for GSK and as you'll see I've rather used my position to help you.

[Sections omitted]

And finally, the reason why you all came. But as I had a captive audience I wanted to do all the rest first – just to do a bit of brain-washing before we got to this bit.

This -> is from the 12th of October when we published our paper in Nature. Using cells from a patient of mine with alpha-1 to try and produce liver cells and in doing so try to correct the genetic defect. It caused a huge, huge interest in the press.

It was published in Nature and other journals picked it up. There was an interview on the Today programme – which is very scary. It's still available – it's online – but my mother's got a copy if you ever want it. I went to the BBC Radio Cambridge studio – do you remember The King's Speech? I was sitting in a room with a huge brass microphone – the rest of the room was shrouded – no one else in there. They put headphones on me and said Edwin Davis will be in this ear and you will be in that ear. They don't tell you what they're going to ask. And another thing about the Today programme is that the closer you get to 8 o'clock the more high profile they think your work is.

So I arrived at quarter past seven and they said you've been bumped to 20 to 8 – that's good. SO you listen to the preamble – they got Tom Field in to do the preamble because they thought that he would be able to explain what we'd done to a lay audience. Then it went across to me talking to Edwin Davis. I looked at this microphone and I thought if I make a complete Horlicks of this, this will be the end of my career. All I have to do for the next few minutes is not to say anything stupid – and in the way of medicine get all the caveats in front. That was my plan. I sat there facing the microphone with this strategy. No matter what the first question was going to be I was going to say 'Yes, but the caveats are - - - '. [long pause]

But of course it caused a real stir in the alpha-1 community, here and abroad. It's just been absolutely terrific.

Let me explain – very simply the strategy was a hugely ambitious project. You cannot imagine how scientifically how ambitions this was.

We took skin cells from a biopsy from a person in this room. The biopsy is tiny – a tiniest piece of skin – turn it into normal liver cells and then put it back into a mouse to show it works. That was our plan.

The person who did this – I suggest that at some stage you get him to come and talk to you – he's a liver Fellow. He saw me give a talk on alpha-1 at the Royal College of Physicians. He said he wanted to do a PhD with me and said he wanted to do it with stem cells.

So I said that's great. I said here's your plan. I want you to take a skin biopsy. I want you to turn it into stem cells. I want you to correct the genetic defect. I want you to produce normal liver cells. I want you to put them into mice and I want you to show that they're normal. And because he's a PhD student he said Yes that's OK [tug of forelock].

[Question] All in three years?

Three years. He went to his mates and said they laughed at me. They say it is not possible in that period of time to do that work. He did it.

Actually the real star of this is this PhD student. He now has his PhD and he was the joint first author on the Nature paper. I don't take the credit for this; I want to give the credit to the doctor who actually did the work. He pulled together all the strands that were required.

[Question] How old is he? [laughter]

He's about 28 [laughter] He's the future – he will be an alpha-1 person. He is into metabolic diseases. He will be a liver disease doctor. He's already been offered serious positions in the liver units in the UK – which I've stopped him accepting because I want him to grow a bit more scientifically. He's had a terrific success but scientifically he needs to do a lot more.

[Question] The Sorcerer's Apprentice.

The Sorcerer's Apprentice. He will get there and he's a really likeable bloke and he must come and talk to you. He's a liver doctor, I just make it up but he's the real deal.

This -> is the scientific slide – I'll take you through it.

So on our Wednesday clinic we said 'and can we take a little skin biopsy while you're here?' We had to go through ethics approval – round and round until we got all the leaflets – because we can't do anything without ethics.

So we take the skin biopsy, grab the skin cells, turn them into stem cells that produce all the cells in the body. Then we turn them into liver cells.

We did the skin biopsies, took out the cells and we grew those cells on plates and we turned them into stem cells. Stem cells are the fundamental cells that regenerate – produce all the cells of the body. They are the starting point of all growth.

We know that they are stem cells because these -> different markers light up. They've all got names but they don't matter – if we have all four lit – it's a bit like if you pull the handle of a slot-machine – when all the things come up you've got what you want. You've got a stem cell.

[Question] how long did it take to get to that stage?

They'd already done that when he came to the lab. It was using a reprogramming factor.

We'd got these stem cells and by cooking – you've got a whole series of recipes – bit of this, bit of that, milk, sugar - - -. Then over 25 days the stem cells turn into liver cells. As they go through the process of turning into liver cells the genes get switched on and we recognise them as being liver genes. When you get to about day

20 they produce albumen, albumen is a big protein in the liver and by about this stage they produce antitrypsin – they are now producing your protein.

So from a skin biopsy we have turned it into a liver cell. That's really clever. We've now got liver cells from people with alpha-1 and we've got liver cells from who they're married to (because they came to the clinic at the wrong time) and we use them as controls. A husband and wife will do fine. We'll take the skin biopsy from the alpha and we'll take a skin biopsy from their partner who doesn't have alpha-1.

The liver cells produce antitrypsin -> the red staining. We then need to say are there any polymers in these cells? Remember what I was telling you, it's all about polymers. We have an antibody marker – a way of detecting polymers in cells. We screened 10,000 cell lines to make that antibody, it's very close to my heart that. It was in my prayers every night. It revolutionised the whole field. So there you go!

Here are cells from you lot and we've made liver-like cells and here -> is a big green signal of polymers. We took skin parts from husbands and wives turned them into stem cells, we made liver cells and we produced antitrypsin. Those liver cells look just like those in your liver. Terrific achievement.

We published that in 2010. We used our antibodies to show those polymers were there. We have human lines now that look just like you livers and I can use these in drug screening. I can use those with my small molecules to see if they will work. If you go back to 2010 that caused a big stir as well – that's another press release.

What we've got is a whole load of human cells that produce polymers. Not great – why would you want me to give you back cells that produce an abnormal protein? N You'd be no better off than what you were when you started. We now need to correct the genetic defect.

So I said to him 'very good, very good my boy.' That was the first year of his PhD. Got a very nice paper – we were very impressed. 'Now do it all over again and along the way when you've made the stem cells I want you to correct the genetic defect in the stem cell – I want you to change two base-pairs out of six billion. I don't want you to change anything else – if you change anything else you'll be in trouble. And I want you to show me that it works.' And he said 'Thank you David.' {tug forelock] That's how it works at Cambridge.

Make the stem cells and correct the genetic defect and to do this he needed – and this is why he was brilliant – he needed to phone a company in the US and persuade them to give him the correcting material, free, as opposed to $30,000. He needed to get hold of the Sanger Centre to do all the genome correction stuff, and he needed the patients and he needed the stem cell technology. This is a huge feat of just pulling people together – he's a very engaging chap, he works very well and then reprogrammed they're the skills you need to get people to pull together. He did it.

Back to where we started. Skin biopsies – skin cells – re-programme them into stem cells. Back to the slot machine – all the colours come up – red, green, red, green – the markers we want; these are stem cells. But these stem cells have your genetic abnormality, they have the Z abnormality.

So we said to him 'Fine but now I want you to cut the DNA next to the mutation.' This -> is the DNA all lined up. These are you base pair lined up as a sequence of four different letters. 'I want you to cut only at one point close to the mutation,'

So he used what is unfortunately known as a FOK 1 enzyme and that cuts the DNA -> breaks it. Then we needed a scaffold to repair it. We got the scaffold from the Sanger Centre. The scaffold lines up next to the break with the correct sequence. So that when the break is repaired the correct sequence is now in there. We choose those cells which have the correct sequence. Five percent of the cells have one gene corrected and point three percent have two corrected. So we could choose the ones which were corrected. Then we said 'fine – that's terrific. Now turn those corrected stem cells into liver cells.' WE went through the recipe – milk, eggs, sugar, - - -. And a month later we look at the cells that we get and we say can you produce antitrypsin and are there any polymers any more?

These -> are the stem cells from the people, like people in this room, and these -> are the stem cells from their partners. And when we corrected the genetic defect there are no polymers ->. We had corrected the genetic defect. We said 'that' fine, that's very clever – but we have to show that we haven't wrecked the genome. If we've wrecked the genome that's not good.'

In Cambridge there's the Sanger Centre and it seems that there's a big button at the Sanger Centre. You put in DNA, push the big red button and out comes the sequence. We sequenced the genome of the cell 30 times and out of the base pairs we looked at there had been 27 changes from start to finish – starting from the skin cell to get to the liver cell and the genetic correction was absolutely clear – beautifully clear. So we had corrected the genetic defect at the stem cell level and we'd produced normal liver cells.

We then said 'that's really clever – but do they work?' You can see it's pretty tough in Cambridge. We had some mice and we put some cells into the livers of the mice and we killed the mice six weeks late. Six weeks later we found cells – can you see that -> that produced human antitrypsin. So the cells that we'd made work. We'd put them in a mouse liver and they function – and the genome is clear. We'd corrected a genetic abnormality.

This can now be done for every genetic disease that involves a single-point mutation. The technology is terrific – it just happens that your disease was the first one through. It's a super way of correcting the abnormality at the genetic level. Technologically terrific but for your disease even more important.

This was published in the New England Journal of Medicine. It's the biggest medical journal in the World. Sandy Sandhaus, who some of you may have met, wrote an editorial to accompany our paper which was published in Nature and once again [new slide} poor old John Walsh ->. The medical fraternity got it – this caused a huge stir in the science community as well as in the patient groups. Scientifically it's a tour-do-force because the graduate student pulled together these different people with different resources to get this done. I can't praise him highly enough as you will have gathered.

In conclusion. The genetic abnormality that you've got forms polymers. This is a disease of polymers. The polymers form in the liver that's why we get liver inclusions and liver disease. Lung disease is largely deficiency – no doubt – and you can also get polymers in the lung. I still think that despite the terrific science that I've just shown you, the cure will be a small molecule. I still believe that and whoever does it good luck to them.

Some drugs may clear polymers in liver cells, at least in mice, and those studies have started. If this works it may negate what we want to do. Good luck to them. Someone has to make progress. This trial is available, it's in the public domain, it's started.

Stem cells may allow the growth of new liver cells. We can put healthy liver cells back into patients.

Finally. People who gave us money; MRC, the Medical Research Council have funded me now for 22 years from when I was a young, spotty graduate student. This is the hardest money to get in the country – they had funded largely on the basic science of your disease.

I'll stop now and just take questions.

[Question] Obviously the first question with stem cells is – when?

Before I retire [laughter]

Let me give you the answer I gave to Evan Davis – has to come with caveats – when you go from stem cell to liver cell I need to be absolutely comfortable that they don't cause harm. You will read in the papers about people doing some very dodgy stem-cell work. In my world this will not get into man until I'm really happy about it. The last thing in the world is for it to go wrong.

Second thing is that the liver cells that we made look relay like liver cells – you will see in the papers I don't call them hepatocytes, we call them hepatocytes-like cells. And that's deliberate because they're not really the finished article. They're like a baby's liver cells and we would hope that if you put them in a liver they would then grow.

Cambridge has just got some money from the Government via the Biomedical Research Centre to set up a big stem-cell initiative. At the end of five years, I, and I will only be peripherally involved in this, will try to get stem cells, liver cells, into man. We have a development plan to be able to do it. The simple way to do it is not to do the genetic correction – the correction adds a whole level of complexity. The first thing is that it will take, probably, primary biliary cirrhosis, with this you get a stable but progressive disease – and I can see a system whereby you can take a skin biopsy from those people, make liver cells, put them into mice – they must be absolutely safe – say for six months and then put them into a patient a couple of years later. Then I would feel comfortable.

[Question] Do the cells have to come from a donor and go back into the same person?

The same person – the beauty of that is that you overcome all the problems of rejection. If I made cells from me and I gave them to you then you'd reject them.

[Question] It'll be very expensive.

Yes. We've answered that for the press – it's actually cheaper than a liver transplant – cheaper than a liver transplant and then long term therapy after transplant. If you have to have a liver transplant, or a lung transplant, you have to be on agents to stop your body rejecting it for years and years. It would be expensive at first and then get cheaper because everything gets cheaper.

[Question] Costs versus augmentation therapy?

One may work.

Do you know when the Guthrie test for all babies at birth will come?

Guthrie test for alpha-1 – Robert Stockley, some of you will know him, when I was a young doctor I worked for Rob. Rob and I worked with Sue Hill to try and get the genetic monitoring group in the UK to have antitrypsin measurement but they threw it out. They said why test now – what's the advantage of testing now when you can wait until people are 12, 13, 14 and make their own decision? Clinically it makes no difference.

[Question] Surely things like cystic fibrosis are tested for?

That's because early intervention makes a difference.

[Question] Are there not cases – children have been lost because doctors did not recognise liver damage?

Good point – but when a baby presents with liver problems then the paediatrician would test for alpha-1.

It's a cost benefit calculation. They screen for under-active thyroid, used to be called cretinism in the old days, because giving babies thyroxine changes their lives. The test for phenylketonuria for leaving the absence of a particular amino acid in your diet allows the brain to develop. The same for CF because early diagnosis may prevent the lungs deteriorating. And so on.

There is a panel for what they screen for. Alpha-1 doesn't make it. Now if we had a small molecule and that has to be implemented early we may get it through. We tried it was an impassioned plea but - - - the logic of science is rather cold.

[Question] The liver cells put back in a patient – is the assumption that they would grow at the expense of the existing liver cells.

That's my idea.

[Question] [contd] Is there any evidence for this?

In mice, yes. The reason for that is that the existing liver cells have a burden which are the polymers. Simplistically, if you put back cells without that burden they should have a survival advantage.

[Question] Going back to screening. Were the panel that you spoke about in favour of a milestone at the age of 12?

It's difficult. If you have a diagnosis of alpha-1 it halves the smoking rate. The Government would say that the smoking rate of 30% is falling (only rising in young girls) – and we tell everybody not to smoke anyway.

[Question] Can we go back to the small molecules?

The vision, the promise, the sunny uplands –

[Question] I've heard that the problem is the degrades.

I see it as more an accumulation and the cell dies – we're in hand waving territory again. Let's assume that you actually reduce polymerisation because you increase degradation – [hand waving]

Ultimately it requires a lot of resources – lost of people.

[Question] Very exciting.

It is very exciting. For me to engage a pharmaceutical company is great. I've been trying for years to do that. With more people engaged we're more likely to get a cure. But we will get one – and before I retire. With that I close.

[prolonged applause]

To read the full verbatim report of Prof Lomas' presentation, please go to http://www.alpha1awareness.org.uk/bristolmeeting.htm, if you do not have access to a computer and would like a copy of the full report please contact me, Lin Daniels, at the PO Box address at the end of the Newsletter.

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Consultation on the UK Plan for Rare Diseases

Myself, Meryl and Jeff Darkins (our membership secretary & treasurer) attended the Rare Disease UK meeting in Cardiff on Tuesday 1st May, to hear about and discuss the UK Government's Consultation Plan for Rare Diseases, below is the report Meryl has produced about the meeting. You have until 29th May to respond to the document, you can see the full document on the website http://www.dh.gov.uk/health/2012/02/consultation-rare-diseases/

There is a response form at the end of the document. You do not need to answer all the questions only the ones you feel are relevant to you, if you prefer you can put

your views into a document and return that instead of the response form, just make sure you include your name and address etc as required on the form.

Thank you to Meryl for her report on the RDUK meeting in Cardiff.

Report on the RDUK meeting at Maldron Hotel Cardiff on 1st May, to advise on responding to the Consultation on the UK Plan for Rare Diseases
The Consultation Document can be downloaded from:

http://www.dh.gov.uk/en/Publicationsandstatistics/PublicationsPolicyAndGuidance/DH_132880

We heard from

1. Dr Annie Procter Consultant Medical Geneticist, All Wales Medical Genetics Service
2. Hayley Cleaver, Chair of Turner Syndrome Support Society
3. Dr Stephen Jolles, Consultant Clinical Immunologist, University Hospital Wales
4. Prof. Julian Sampson, Head of Institute of Medical Genetics, Cardiff University
5. Michele Matherson, Senior Public Affairs Officer, Wales Council for Voluntary Action.

The first four speakers addressed the specific consultation questions and the final speaker gave general guidance on responding to any consultation document. My comments concentrate the remarks of the first four speakers; Michelle's advice is available separately.

- There was a recognition/explanation of the difficulty in diagnosing rare diseases (RD) because of the rarity and variability in the symptoms, co-morbidity and the availability of data to inform diagnosis.

- There is a need to improve education about RDs

  (ii) in undergraduate medical training;
  (ii) in in-service training for GPs and consultants for common conditions, to recognise the need to refer on when confronted with a rare condition and of the acceptability and indeed, advisability to do so;
  (ii) in training for ancillary staff such as physiotherapists.

- There were mixed messages about neonatal screening; there is variability in the number and type of condition tested within the UK, and, despite the universality of the screening for a small number of conditions, this contrasts with the large number (hundreds) of conditions screened for in the USA and Australia.

- The "Diagnostic Odyssey", where years are taken to diagnose rare conditions was mentioned. This has cost implications: financial, where many expensive treatments are applied unsuccessfully, where the opportunity to make informed choices about lifestyle is lost with the consequent unintended exacerbation of conditions, where informed choices about further family planning are missed; emotional and psychological, where patients feel isolated and begin to doubt the veracity of their symptoms ("am I imagining it?").
- To facilitate the transfer of good research outcomes to good clinical practice there needs to be judicious use of IT; there is maybe a role here for RDUK to act as a clearing house for reliable, authoritative and up-to-date information on RDs for GPs and staff in District General Hospitals. There was much talk of a portal for information not only on research but on details of symptoms indicating RDs.
- Everyone agreed that there should be co-ordinated care, and that RDs pose a particular challenge in getting an adequate caseload to develop expertise and experience for development. Consultation questions 5 and 6 are specific about the basis for shaping future care. The document is rather complacent about the present situation. All the sufferers we spoke to complained of a <u>lack</u> of coordinated care. *There is particular problem in Wales, which has <u>not one</u> specialist care centre. Referral to a specialist centre in England has to be submitted to a Welsh Assembly committee for approval; a very bureaucratic and time-consuming process.*
- Question 7 deals with the need for (i) expert centres to know the network of hospitals they serve, and (ii) local hospitals to know their pathway to the expert centre. All speakers mentioned the fractured nature of the relationships at present. There is no level playing field, with variations within England and between the 4 UK nations. The proposition in the document seems utopian and masses of work is needed to bring about the necessary coordination, although Dr Stephen Jolles suggested one way of moving forward on this.
- Question 8. In England, from April 2013, Healthwatch England will provide a platform for making the NHS and local government accountable to their local communities. This will enable the collection of views of the users to influence policy and provide feedback – this should be a useful channel for people with RDs (*no such mechanism is envisaged for Wales)*
- Question 9: Should the UK continue in the Orphanet Project? Obviously YES.
- Question 10. Information to patients.
  (*personal note: I am appalled and aggrieved that, however imperfect the NSF in England, the Ips in England, the Information Standard in England, the NHS forum in Scotland, the ALISS in Scotland and the plans for a framework in Northern Ireland, there is NOTHING in Wales and NOTHING is planned - this is incomprehensible complacency by the Welsh Government)*

  Patients need reliable, authoritative, up-to-date information to cope with their conditions and to adapt to their condition. In the context of RDs this is very fragmented and is frequently provided by patient charities and groups. The role of the patient groups and charities is very much understated and undervalued by this document. There is a need for the Information Standard being developed in England to be applied across the UK – we all know of the

danger of misinformation and unreliability of information posted on the Net!!!!

There was little attention to the psychological impact of being given a diagnosis out of the blue, this an area where patient groups can make a real difference, especially with RDs

- Question 11 : the role of registers. The document suggests several ways of approaching registers, and speakers spoke about taking registers out of the narrow range of use by researchers alone. Some registers are for particular conditions, some administered by charities(e.g. Cystic Fibrosis register), some by Pharmas. In the Nordic countries, permanent residents have a unique number, which can be used in compiling registers (don't we each have a unique NHS no??) The Scottish Morbidity Record is a hospital activity database funded as part of care. It must be in the interest of RDs to develop registers at some level.
- Question 12 : Equality. One field where there is a lack of equality is in the way in which the transition from paediatric care (where the paediatrician coordinates care) to adult care, where no-one does. There are massive variations in the quality of care across the UK, and between the articulate and vociferous compared with the ignorant and timid. Loads of work to be done here.

  (Wearing my Welsh hat again. The paucity of the Welsh Assembly input to the consultation document is staggering. Several speakers spoke of absence of centres of excellence in Wales, despite the existence of an outstanding genetic facility in Cardiff. The protocol for recruiting clinical and research staff militates against institutes in Wales. There are few opportunities to recruit senior, mature researchers (i.e. above post-doctoral fellowships. There are no mechanisms for overcoming the problems of the geography of Wales, with its attendant communication problems.)

Finally, all speakers were much more impressed by the RDUK Vision Document than by the United Kingdom Plan for Rare Diseases, the subject of the consultation. All speakers supported the idea of an information portal for disseminating information about symptoms and treatments to health professionals, in order to improve the identification of RDs as quickly as possible.

I am intending to respond in a personal capacity (in case it's quantity rather than quality they want)

Meryl Darkins

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As we told you in our last Newsletter, we have secured 6 places in the British 10K Run, for this year and the next 2 years. So far we have 4 definite runners, there are 2

places still available for this year's run. From what I understand, 2 people took registration forms at our meeting in Bristol, but as yet the forms have not been returned to Jeff Darkins, our treasurer, who is dealing with entries, so if you want to take part in what promises to be a great event, please contact either Jeff at jeff@alpha1awareness.org.uk or myself, Lin Daniels at secretary@alpha1awareness.org.uk, as soon as possible.

Could you, a member of your family or a friend raise money for Alpha 1 Awareness UK in the British 10K London Run on Sunday 8th July 2012? We know we have quite a few runners in our membership so we would love to hear from you.

The British 10K London Run is the UK's most prestigious and sought-after 10km road race which is staged on the world's greatest route through the heart of central London. 25,000 runners fill the streets of the nation's capital and get the unique chance to run past many of the country's greatest landmarks including Big Ben, The London Eye, St Paul's Cathedral, Trafalgar Square and Westminster Abbey. Abilities range from charity fundraisers and recreational runners right through to Olympic champions!

The 2012 event is staged just 19 days before the start of the Olympic Games and the excitement and buzz within London at this time should make for a truly special occasion and the best British 10k London Run in its 12 year history. http://www.thebritish10klondon.co.uk

This should be great event, with London looking it's very best following the Queen's Jubilee Celebrations in June and the start of the Olympics in July and it gives us a chance to really get Alpha 1 noticed.

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Chat online to other Alphas

Remember if you want to chat to other Alphas and you have access to the internet, there are 2 ways you can do it. There is:

The Alpha 1 Awareness UK Forum - to join go to
http://techno.demon.co.uk/a1aforum/index.php

We also have a Facebook Page – If you are on Facebook, search for "*Alpha 1 Awareness UK*" we have our logo next to our name, click "request to join", it is a closed group page so nothing on there is public, other than our Group description.

Shop Online and Raise Funds

Don't forget when shopping online to register with **easyfundraising.org.uk** and it won't cost you a penny more to shop and raise funds in this way. In fact you could even SAVE MONEY as many retailers give exclusive discounts, special offers and even 'e-vouchers' when you shop through the easyfundraising site.

Easyfundraising is a shopping directory featuring over 600 trusted online stores, including: Asda, Tesco , Argos, Amazon, the Body Shop, NEXT, Debenhams, John Lewis, Toys'R'Us, HMV, Virgin, iTunes, CD WOW, Marks and Spencer, Currys, Dixons, Staples, PLAY.COM, Pets at Home, Choices Direct, WH Smith, The AA, RAC, Direct Line, Churchill, The Carphone Warehouse, Ticketmaster and over 600 others...

Register for **Alpha 1 Awareness UK** and just by doing your shopping online you will be helping raise funds for
the Charity.

Also you can set up www.easysearch.org.uk, as your search engine page and register Alpha 1 Awareness as your chosen cause then every time you search a webpage you raise money for the Charity.

We would love to hear from you, if you wish to contribute in any way to future editions of the Newsletter, please contact at the address below.

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