

Report of the meeting held on Tuesday 28th April 2015

Present: Board members: Mark Arthur, Caroline Richmond, Chanell Pritchard (secretary) Kit Sandford, David Webster, Sary Workman, Gary Narborough and 35 members. Apologies were received from Jose Drabwell (chairman), Andrew Symes and several members. The meeting was chaired by Kit Sandford.

Membership Kit Sandford thanked Caroline for bringing the membership to above 200 by attending the Royal Free on clinic days and persuading potential members. Caroline reported that there were few refusals, and now that the majority have joined it was no longer time-effective to continue recruiting in this way. However, as members change their email addresses and new patients join, she will continue intermittently.

Annual Patient day, this year in the hospital Atrium this autumn. Members were asked whether they would prefer this to be on a Saturday or a Sunday; Sundays were more readily available. There were few objections to either day. Caroline proposed that lunch tables at the meeting could be marked by districts so that members could, if they wished, meet others who live near them. Organisational help from members will be much appreciated. A member asked for a brief session on the day about vaccines “ which ones are recommended, and which are unsafe? This would be a useful topic for the website.

Research: Mari described how important it is to involve patients in research design and implementation. For example, a recently-introduced treatment in children with serious genetic immunodeficiencies is to give them a new immune system by doing a stem cell transplant. This originally had a high death rate and was therefore offered only to the severely ill. It has steadily become safer and so it is used more widely. However, little is known about recipients' long-term quality of life. This is what we want to find out. To pilot the project, Mari assembled a focus group of patients, parents, patient organisations' representatives, and members of the clinical team. They discussed their experience of transplant and treatment, and the aims of the project. Everyone involved found it helpful in finalising the topic and prioritising what should be addressed. This improved the study design and made it more focused on patient need. It went so well that the team hope to run more focus groups in the future.

Individual consent to participating in research Sary Workman (research nurse) told us that there is a new 'umbrella' consent form “ that will enable those who sign it to have their data used, confidentially, in a range of research projects. Those who have already signed consent forms will be asked to sign again. As she is trying not to overload us with too many forms at once, she has asked Chanell to send the forms out to us a week before our clinic appointments. This will give patients time to read and consider them. There are eighteen research projects under way in the department.

New treatments in the pipeline Sary told us that there are new ways of giving immunoglobulin coming on stream in the foreseeable future. Small doses can be given subcutaneously daily, or huge doses once a month. The clinic staff are trying to individualise treatments, and will discuss this with us when we come to our clinic appointments.

Dr David Webster reported that gene therapy is currently on trial at Great Ormond Street hospital and the Royal Free for Wiscott-Aldrich syndrome (the fourth type of PID treated in this way). The defective gene in bone marrow cells is replaced by a normal gene. This is the dreadful condition that affected Anthony Nolan; the International bone marrow bank is named after him. The disease causes severe immune-deficiency and a bleeding disorder.

Another important landmark is that drugs developed by the pharmaceutical industry to treat various "common" immunological diseases are sometimes turning out to be useful in correcting specific rare genetic defects causing PIDs; two such drugs are being tried in rare types of CVID patients. This underlines the benefit for patients in the UK research initiative to find the genetic defect in all patients with PID.

Treating viral gut infections David Webster said that norovirus is proving very hard to treat in some PID patients; this virus is a common cause of vomiting and diarrhoea in the community but persists and causes severe bowel problems in some CVID patients. There are drugs that are likely to kill this virus but hospitals have rules that make it very difficult for clinical consultants to use these. The Board recently wrote to the Royal Free committee responsible for using medicines for unlicensed indications suggesting that the rules be modified for patients with rare conditions like PID -- a reply is expected soon.

Exercises to improve the lungs: A member asked for advice on lung exercises that can be done at home. Sary replied that there used to be a physiotherapist attached to the unit but there is no longer any funding for this. She hopes to reinstate this, and to have a physio workshop at the patients' day. Caroline has since found some internet videos of breathing exercises which are being checked by the clinical team; if considered useful there will be a link flagged on our website. Sary reported that she and Andrew will be handing out flutter-valves to patients who need them. These are hand-held devices that use a combination of vibration and air pressure to make it easier to cough out any mucus.

Miscellany: A member asked for a beginners' guide to terminology and jargon on the website. Patients are finding it difficult to contact anyone when phoning in from outside. Sary will look into this but advised that either herself or Andrew can be bleeped via the switchboard if infusion appointments need to be changed or there is an urgent medical problem.

Next meeting Thursday 2 July, 2015, 5.30 -7.30 pm