BADGEM Informatics Meeting 1st July 14.30pm – SECC, Glasgow.

Attendees: Irene Leigh, John Kernthaler, Mozheh Zamiri, Neil Rajan, Celia Moss, Marilyn Benham, Vinzenz Oji and Louise Stanley.

A brief meeting was held to discuss the progress made towards creating a national register for genodermatoses.

Neil gave the committee an overview and update, namely:

(i) Aim to create register to capture all rare skin disease patients in UK
(ii) Collect minimal data (not deep phenotyping)
(iii) Host on secure NHS server and development by HIC in Dundee
(iv) Initial specification and quote provided by HIC has been accepted- Test system developed by HIC should be ready by mid-late August for comment/trial
(v) BADGEM data can feed into national disease register in England being developed by Jem Rashbass (this is not in competition with this project)
(vi) Possible suggestion for consent to carbonised and then to be sent centrally to attach to patient entry (who to do this?) Alternative would be to have the form downloadable as a PDF. There also needs to be a mechanism for recording level of consent on the register.

Discussions were held around:

(a) Disease codes: The list of disease codes (whether based on geneskin, ICD9/10/11 or BAD) and method of entry were discussed. Potential options including look-ups, drop down menus and a "google" approach were also discussed. The number of BAD codes are significantly more than the geneskin codes and will influence the functionality of the site. Neil has asked for feedback on the system used by the Human Phenotype Ontology (http://compbio.charite.de/phenomizer/). Two separate data entry areas may be required, depending on whether the clinician knows the diagnosis or if only the phenotype and a disease grouping is known. This would help capture undiagnosed but rare genetic conditions.
(b) Only rare conditions with a skin phenotype (single gene) will be included and others which are common, such as eczema, or without a skin phenotype, but say neurological phenotype are out of scope. List of genes known to be associated with disease codes is required – request from Diagnostic Signposting group. Disease codes and entries need to be decided upon as this affects development by HIC and to be provided as soon as possible.
(c) The possibility of patients registering themselves into the database (in addition to consultant led entries) was discussed. The advantages and disadvantage of this approach were extensively debated but it was agreed that, at least in the initial phase of development, that this would not be supported. Patients would be encouraged to seek registration by discussion with their specialist if appropriate, possibly via the informatics register website. BAD would retain ownership of the data in the event of the register ending.
(d) Access to data entries, tiers of access rights/viewing capabilities, record of "quality" of entry were also discussed at length. User names and passwords for access to the register will be processed and authorised by the BAD. It is anticipated that very few, individuals requesting access will be non-BAD members. It is proposed that an audit process where data is sampled and independently reviewed validates the data. Access to patient entries would be restricted, at least in the initial stages, to the consultant that entered the data. However, it is thought to be a good idea to produce reports (e.g. once a year) on how many patients have been recorded against each disease code/category and publish this information on the website. It may be useful to have a discussion of data visualization outputs at the next meeting with HIC. One example by Vinzenz used in NIRK was knowing the locality of patients with similar phenotypes and therefore potentially an algorithm using postcode data to determine distances between addresses would be useful. An information governance committee will need to be set up to authorise data access requests for research purposes. Any requests from Pharma should have a charging mechanism to act as an income stream.
(e) Incentivising patient entry was discussed. It was agreed that a financial gain could not be supported nor be encourage. An alternative incentive such as some possible measure of activity that is creditable to the department could be used. This would need to be actioned by registering the informatics register as a project to capture the prevalence of rare genetic disease in the UK,

with a view of it being a portfolio project on dermatology CLRNs nationwide. Recruitment would then count as activity and influence departmental CLRN annual budgets.

(f) Ethics proposal and forms for recruitment should capture both new patients, and be suitable for taking over existing patients from other registers. A discussion of whether one form or 2 were needed was held and it was felt that 1 single form would be appropriate. Taking over more complex registers (E.g. EB with extra phenotypic data) would need the additional phenotypic data to be stored as a linked second page to the primary record. This would need to be discussed with HIC, once a sample entry from the EB register is seen, to determine the complexity of this data.

(g) Increasing visibility of the project needs to occur. The aim is to have a working solution in place by Feb 2015 for the SDS (Scottish Dermatological Society) meeting.

(h) BADGEM is to have its own website (www.badgem.org.uk) which will have sections for diagnostic/clinical signposting, clinical trials and also for the register – with a link taking consultants to the secure login page.

Date of next meeting: Tuesday 14th of October 2014 10am Willan House, London