BADGEM Informatics Group Meeting – Tuesday 25th February

11am-2pm. Willan House, 4 Fitzroy Square, London

Chaired by Neil Rajan and Irene Leigh

Minutes

In attendance: Neil Rajan (NR), Irene Leigh (IL), Celia Moss (CM), Sinead Langan (SL), Keith Milburn (KM), Marilyn Benham (MB), Sagir Hussain (SH), Jem Rashbass (JR) and Louise Stanley (LS).

Apologies: John Dart

Welcome

As way of introduction to BADGEM's registers project, Neil Rajan presented his vision for the rare genetic skin disease database. Covering topics such as potential uses of the register (research, grant applications, clinical trials, lobby for funding, clinical use), integration of existing and potential registers (e.g. EB, EU), data input (clinicians, nurses, patients), type of data to collect (minimal data set vs deep phenotyping) and challenges to overcome (e.g. engaging support and participation from community, securing CRN support, database design).

Disease registers and the NHS - Jem Rashbass

Jem Rashbass (National Director Disease Registration, Public Health England) gave a very informative and insightful overview of the work to date on disease registers in the NHS. The aim is to create disease registers (in England) for congenital anomalies and rare diseases building upon the model implemented and successfully used for National Cancer Registration (total cases registered 275 600, minimum data required for registration. N.B currently excludes non-melanoma skin cancer). Ultimately hope to collect high resolution and quality phenotyping data to complement genomic data generating clinically useful datasets for congenital anomalies and rare diseases.

Important lessons learned from the National Cancer Registration include:

- Do not try defining extensive datasets – time consuming, agreement difficult and often become obsolete
- Need patient and public support; local knowledge and links across NHS
- Driven by partnership (patients, carers, clinical teams, 3rd party sectors, research and "Government"
- **Data quality** driven by **timely feedback** - clinical and to patients (patient portal – PROMS)

National Cancer Register achieved through team >200 individuals, large operation with dedicated data liaison teams to persuade participation and gain vital data feeds (key to success). Dedicate analysts to resolve data mismatches.

National Cancer Registry has made use of section 251 of NHS Act 2006 which allows the common law of confidentiality to be set aside in specific circumstances where anonymised information is not sufficient and where patient consent is not practical.

Rare disease challenges:

- Numbers much lower requiring flexible infrastructure making automation difficult.
- Way to identify non-index cases e.g. terminated fetuses (option - maternally).
- Record cases that do not have diagnosis with "SWAN" – and record what is known. What disease definition system to use – BAD codes, Orphanet, ICD11 (when released).

Jem expressed that BADGEM can join the forthcoming Public Health England initiate if we would like. Interaction will be important, particularly for England.

ERN and European Registers, Geneskin Patient Groups – Irene Leigh

Irene Leigh gave an overview of the European projects (active and historical) with the aim of placing a funding bid in April 2015 as a consortium (BADGEM included).

Three major projects:

- Geneskin (founded 2005 – only 5 year funding by European Union)
- TAG (Together Against Genodermatoses – hosted by the Rene-Touraine Foundation as a pilot project for European Reference Network, 2007 now adopted by EADV). ERN has various sub-groups with team leaders. A few BADGEM steering group members sit on some of these sub groups. Rare disease register group of the ERN met and has decided to focus on EB as pilot project as a lot of work has already done for this group.
- ClinetEB (2012)

Overlap between all activities. Irene suggested use of disease classification as defined on Geneskin (and subtypes) and refine/reclassify as necessary.

Disease Specific Registers

Discussion occurred around existing disease datasets: NIRK (Germany), PC Project (USA), ClinetEB (Austria) and Scottish EB (Helen Horn). Celia Moss presented an overview of the Epidermolysis Bullosa Data System in which the four EB centres of expertise enter data live in a bespoke electronic patient record – accessible within NHS net. These patient have not consented for data to be used as a research tool. Range of fields can be/are collected including prescriptions, dietary, dental, follow ups (less info collected on simplex and milder patients as not necessarily followed up).

Health Informatics Centre and provision of infrastructure – Keith Milburn

Keith Milburn the Health Informatics Centre (HIC) at the University of Dundee gave an overview of the services that they can offer. It operates on a non-profit basis (£508/day programmer, £140/day data entry) and they can assist on all aspects of projects, from design, data entry, software development and recruitment for clinical trails. They provide secure data storage and can provide a safe haven whereby researchers can access anonymised data but are unable to remove it.

Keith advised the group to:

- Decide on what BADGEM is trying to do and achieve
- Base register on an evolutionary structure
- Scope project out but not for too long; better to get something up quickly with proof of concept – keeping data set minimal.

Sinead Langan raised question of consent and advice is to keep broad as possible to minimise repeat requests to patient resulting in fatigue and lack of engagement.

Minimal Data Sets:

Names for consideration for register: Badgem-R or BADgeR

Group progressed to discuss what the minimal data fields would need to be collected. Minimising free text boxes important. Data entry envisage by dermatologists – BAD can act as portal for promotion.

Diagnosis coding: Neil Rajan suggests that the system is developed that allows for suggestions as the name is typed or allow for search of the respective databases and delivers suggestions based on clinical entry

Data entry: Possible mechanism for the hesitant participants to add the data - molecular and phenotype, but choose not to add unique identifiers and spine data. We should discourage this but it would allow partial participation, and could be merged with the patient file later.

Unique local identifier (NHS number, CHI number)
Basic Demographics (name, d.o.b., gender, address)
Diagnosis (coding system to use – ICD10 (inadequate?) BAD codes (potentially redundant), Orphanet, ICD11 when released)*
Molecular/genetic data
Lead Physician/Dr. of contact
Centre in which data entered
GP

* for cases with no definite diagnosis suggest SWAN (syndrome without a name) category and description

Discussion about promoting this initiate to patient support groups occurred (e.g. at BAD meeting) with the general agreement to focus on the more neglected patient groupings e.g. ectodermal dysplasia, ichthyosis, PPK rather than XP and EB.

Will be important to have both clinical and patient portals as part of infrastructure but initial phase of project is likely to primarily focus on clinical portal and data collection.

Point of discussion around EB database and integration – this is likely to be addressed nationally and group will await outcome.

Action Points:

- Circulate minimal dataset suggestions arising from meeting for comment and consideration by sub-group committee. **Deadline 14th March 2014 – ALL**
- Collate responses for minimal data set and liaise with HIC to generate quotation for project proposal. LS/NR/KM
- Buy url domain for BADGEM website (public facing). LS
- Subsequent mock ups of front-end interface with deadline for 1st July. KM
- Present (NR/IL) BADGEM draft interface and purpose to patient groups at BAD meeting (suggested date/time 1st July pm). MB to compose letter once more details clarified to inform patient groups point of contact.
- Date of Next Meeting - 1st July 2014 at BAD conference, Glasgow. Afternoon time slot to be confirmed by MB for suitable integration into conference programme.