DERMATOGENETICS – a new special interest group linking the BAD with the BSGM

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Why?

'Dermatogenetics' is a newly established special interest group. We aim to develop links between **Dermatologists** with an interest in genetics and **geneticists** with an interest in Dermatology, to facilitate the sharing of knowledge and exchange of ideas in both clinical medicine and research.

Dermatogenetics has been accepted as a special interest group within the **British Society for Genetic Medicine (BSGM)**. We are currently the second sub-group to be established in BSGM (after cancer genetics), reflecting a real interest amongst colleagues in genetics and their awareness of the importance of Dermatology in the fields of genetic medicine and at the forefront of genetic research.

How does the Dermatogenetics special interest group fit with other Dermatology networks?

Dermatogenetics is a sub-group of the BSGM. The 'BAD-Dermatology and Genetic Medicine' group (BADGEM) is a clinical network led by Prof Irene Leigh; BADGEM functions to link clinical Dermatologists with an interest in genodermatoses and genetic skin disease. The Dermatogenetics group aims to link clinical and academic Dermatologists with clinical geneticists, including medical, nursing and laboratory-based scientists, and will work closely with BADGEM.

BADGEM and Dermatogenetics are supported by the BAD and by the Wellcome Trust-funded centre for Dermatology and Genetic Medicine (DGEM), Dundee, led by Prof Irwin McLean and Prof Irene Leigh (see **Box 1**).

Box 1 DGEM and BADGEM

The **Centre for Dermatology and Genetic Medicine (DGEM)**, located in Dundee, is supported by a strategic award from the Wellcome trust and is aimed at translating basic science discoveries in genetic skin disease into clinical application. The Centre brings biologists and clinicians together with physicists and chemists to tackle the major challenge of developing new medicines for skin disease and delivering these novel molecules into the skin. DGEM is fully equipped with high throughout, cutting edge genome sequencing technology (see Genomic Sequencing Unit; http://gsu.lifesci.dundee.ac.uk/) to facilitate resolution of the remaining unsolved skin conditions. The unit is also expanding its dermatology drug discovery programme and any individuals with potential projects are welcome to contact Dr Andrew Woodland, DGEM Drug Discovery Portfolio Manager for more information (a.woodland@dundee.ac.uk).

Professor Irwin McLean is the scientific director leading the programme with Professor Irene Leigh acting as the clinical director.

The **BAD-Dermatology and Genetic Medicine (BADGEM)** is a company set up by the BAD to act as UK-wide network for genodermatoses. BADGEM is chaired by Professor Irene Leigh and is governed by an executive committee to determine it's projects, monitor their performance and coordinate activities for the best outcomes for UK patients with genodermatoses. Three working sub-groups have been established; bioinformatics to guide development of a clinical database, diagnostic signposting and clinical trials.

Professor Irwin McLean
DGEM Scientific Director

Professor Irene Leigh
DGEM Clinical Director

Opportunities…

Genetics is a field of active research in Dermatology, with the potential for early application to clinical practice. The use of current DNA sequencing technology offers the promise of identifying genes and genetic mechanisms responsible for many of the Dermatological syndromes and signs which remain incompletely understood. The highly ambitious 100K Genome Project, announced by David Cameron in 2012, aims to sequence the entire DNA code of up to 100,000 NHS patients (see **Box 2**). This will represent a uniquely powerful resource for the research community, including rare disease research in Dermatology.
The field of genetic research is moving forward at such a pace that it is difficult to keep up. Dermatogenetics aims to help by providing clinically relevant and up-to-date information for clinical Dermatologists as well as those with an academic interest.

Specifically, *Dermatogenetics* aims to:

- Improve the recognition and phenotyping of cutaneous manifestations of genetic disorders, by closer working between Dermatologists and geneticists.
- Provide a forum at the annual BSGM to discuss cases that have cutaneous manifestations at a multidisciplinary meeting involving Dermatologists, geneticists and scientists.
- Offer opportunities for the education of specialty trainees in genetics and Dermatology via seminars held at the annual BSGM and/or BAD meeting.

Upcoming meetings

Our first meeting is a half-day event that will be held at the BSGM annual meeting in Liverpool on the 22-24th September 2014. This will comprise of

Dermatogenetics


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lectures on genetics and dermatology, including talks by Professor Celia Moss and Professor John McGrath.

We invite you to join us at this meeting and to submit abstracts, which, if accepted, will be published in the *Journal of Medical Genetics*. Registration and abstract submission will be via the BSGM website http://www.bsgm.org.uk/. Abstract submission is expected to open on 7th April and to close on 24th May 2014.

How to become a member of Dermatogenetics?

Membership is open to individuals with an interest in the clinical and basic science of the hereditary aspects of skin disease. Expressions of interest should be directed to Sara or Neil (contact details below). Dermatogenetics membership fees of £30 will be administered via the BAD in line with other Dermatology special interest groups. Non BAD members will join via the BSGM.

Contact us

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Box 2 100K Genome Project

In 2012 the **Human Genome Strategy Group report recommended a major investment in the application of genomics to medicine**, recognising the opportunity to redesign healthcare and build on the UK's leading role in the development of the field. In December of that year the **Prime Minister announced a plan to invest £100 million** to pump prime the production of 100,000 whole genome analyses within the NHS, focused on finding the causes of rare diseases and contributing to the care of patients with cancer and infectious diseases. The last year has seen a rapid adaptation to this bold decision; a company called **Genomics England Ltd owned by NHS England** has been created with Sir John Chisholm as chair and Professor Mark Caulfield as its chief executive. The Board includes the Chief Medical Officer, Sally Davies and former chair of HGSG, Sir John Bell. A scientific advisory committee, of which I am a member, has met twice and is developing a business plan based on input from three committees looking at the disease groups, an ethics committee and a team addressing technology and informatics.

An early decision was to **invest in building the country's bioinformatics capacity** and a Genomics Advisory Board within Health Education England is addressing this challenge. What will not happen is the purchase of multiple expensive machines which will rapidly be overtaken by new technology. Instead, the major manufacturers have been invited to demonstrate that they can produce high quality sequence and contracts will then be constructed to allow them to run facilities within the NHS firewall and be paid for completed sequences. The leading company is Illumina who have recently announced their HiSeq X (pronounced High Seek Ten) which can generate a whole genome for $1000.

A **pilot programme is collecting 2000 samples** in the NIHR Bioresource from Newcastle, Cambridge and London where an appropriate consent system was already in place. At the time of writing, over 700 consented samples had been collected in under 2 months.

There are many challenges ahead but it is already apparent that this bold initiative to build whole genome sequencing into routine NHS care is having a galvanising effect. **All clinicians need to consider how to participate in this venture.** The Dermatogenetics group is well placed to help identify targets for the all categories, especially the rare disease cases. There is a real prospect of **Whole Genome Sequencing becoming a core investigation, allowing therapy to be more targeted and rare adverse effects predicted.**

It remains unclear whether the NHS informatics can cope with storage and safe access, or that we will move towards cheap testing and discarding unwanted data, or developing simple disposable devices for each application once the key drivers have been discovered from pooled whole genome data.

Genomics england

Prof Sir John Burn

Useful links

British Society for Genetic Medicine (BSGM) http://www.bsgm.org.uk/

Dermatology & Genetic Medicine (DGEM) http://dgem.lifesci.dundee.ac.uk/

Genomics England http://www.genomicsengland.co.uk/z