BADDGEM Clinical Diagnostic Signposting Committee.
Monday 23rd March 2015, Willan House, 4 Fitzroy Square, London
11.30 am - 1.30 pm

Attendees
Edel O'Toole [Chair]
Mandy Aldwin
Liz Jones
Nicholas Lench
Mike Simpson
Sagair Hussain

1. Welcome and apologies

Apologies from David Kelsell, Jemma Mellerio and Veronica Kinsler

2. Previous meeting minutes and matters arising

The minutes were accurate reflection of the meeting and there were no subsequent comments.

3. Update on clinical signposting

Edel presented (PowerPoint) the updated list of geneticists. Subsequent discussions were based on this list. The committee felt the following individual should be added to the list:

Andrew Carmichael, dermatologists

Liz mentioned Southampton has a big genetics centre and there maybe individuals there that could be added to the list, Edel will investigate this further. Liz will e-mail Glasgow to identify geneticists that could be added.

The discussion moved onto whether Northern Ireland should be included and the committee felt it should and the following individual was suggested

Keith Armstrong

Liz will investigate geneticists based in Northern Ireland.

The discussion moved onto the inclusion of Alex Anstey, the committee felt he should be asked if he's interested in being added. Although, it was unknown whether he was in the midst relocation to North Wales.

Edel felt the list could be added to the webpage and updated as necessary. The committee felt the named individuals on the list should be notified of their inclusion to seek consent. In addition, they should be notified of the purpose of the list, the reason for their inclusion as well as the implications of inclusion. Although, Liz highlighted on the Tubular Sclerosis (TS) Association website there is a list of all the UK TS specialists and a contact person.

Action Point 1: Edel to add to the list further and e-mail everybody

4. Update on diagnostic (genetic) signposting

No further update on the diagnostic signposting.

With regard to commissioning, Liz felt things were up in the area as it varies depending on the budget for genetic testing in each centre. Each genetic testing centre is commissioned differently, for example there is a separate budget for internal testing and those that are sent externally outside of Manchester. Each centre has a different funding model to meet testing demands. The committee felt the current set up was very confusing and sometimes it was difficult to know who and or where to send the samples for testing.

5. Update on DDD and 100K Genome Project

DDD

Liz mentioned, generally DDD study includes developmental delay plus dysmorphic features or developmental abnormalities. If they have multiple developmental abnormalities some of them are still eligible. Recruitment is aimed to finish at the end of March 2015 but they will keep doing the analysis for the next five years. They are only sequencing genes that are involved in development and are not sequencing everything.

GEL

Liz mentioned, they had a pilot phase which has just finished although is awaiting results from this phase. The range of disorders submitted to this phase was very broad. Manchester is now recruiting to the main project although the list of disorders is much more limited and the list can be found on the main GEL website.

The Genomics England Clinical Interpretation Partnership (GeCIP)

Edel gave an introduction into the application for skin GeCIP and subsequent discussions were based on the document provided to the committee. The steering committee for the application consists of, Edel, John McGrath, David Kelsell, Neil Rajan, Richard Warren (Manchester), Mike Arden-Jones (Southampton) and Irwin McLean (Dundee).

John has proposed 3 sub-domains; monogenic subdomain, neo-plastic subdomain and inflammatory subdomain. John is keen in doing recessive Ichthyosis, all the EB's were there are no mutations, keratodermas and ectodermal dysplasia whilst Neil is keen on cancer. Edel is keen on TEM which has a genetic component as well eczema variants that don't have SKN 5 mutations. They will get feedback from GEL on the application soon. The plan is for the clinician that actively sees the patients to recruit into GEL and ideally to have deep phenotypic data as well as trios.

6. Feedback from ISG

Mandy is continuing to work on the BAD newsletter article summarising the results of the BADGEM survey of ichthyosis patients and families regarding "Genetic diagnosis testing".

7. AOB

N/A