Cross Party Group on Rare, Genetic and Undiagnosed Conditions

Tuesday 5th September 2017

Committee Room 5 – 1pm to 2.30pm

MINUTE

- **Welcome and introductions**

Apologies were received from Lynne Shields (Tuberous Sclerosis Association), Sue Rees (UK PIPs) and David Newman (Alexion).

Attendees were invited to introduce themselves and the organisation that they were representing.

- **Review of minutes from last meeting**

The minutes of the last meeting have been approved. The action to investigate the outcome of the Specialist Nursing Fund (announcement from 2015) has been carried over.

- **Update on workplan and collaborative work with other Cross Party Groups**

At an earlier meeting, there was lengthy discussion on the priorities for this group. Several topics raised were not necessarily specific to rare, genetic and undiagnosed conditions and may be more appropriate for the work of other cross party groups. Natalie has been in touch with the Chair/Secretariat of a few of the groups to set up meetings to discuss possible collaborative working. Natalie has written to:

  - The Cross Party Group on Disability – Natalie will be meeting with the Chair, Jeremy Balfour MSP at the end of September.
  - The Cross Party Group on Learning Disabilities (as education needs were raised as a priority)
  - The Cross Party Group on Children and Young People (as there many of the rare, genetic and undiagnosed conditions affect children)
  - The Cross Party Group on Sport (as access to sport opportunities were raised)

The priorities for our group will be:

  - Co-ordination of care and specialist nursing
  - Scottish Plan for Rare Diseases
  - Data Management and Sharing
  - Access to Medicines
  - Cross Border Healthcare
  - Transition
  - Benefits, welfare and social care

- **Topic One: Access to New Medicines**

  There was discussion at the last meeting regarding a number of the outstanding recommendations from the Montgomery review on access to medicines.

  Attendees were invited to provide further feedback on the Scottish Medicines Consortium (SMC) and Individual Patient Treatment Request (IPTR)/ Peer Approved Clinical System (PACS) process.

  Key points from the discussion included:

  - Many positive improvements since changes to SMC were introduced in 2014 – particularly with reference to patient and clinician engagement. It is too soon to comment on success of Montgomery recommendations and several recommendations are still outstanding.
  - With regards the Patient and Clinician Engagement Process (PACE), several attendees noted the importance in having representation from the right specialist clinicians and that such expertise may exist outwith Scotland. There is a need to have patient confidence in the expert clinical opinion. It was suggested that for some patient groups, establishing a medical advisory board may be beneficial although it was also noted that for some, very small and/or under resourced organisations that this may not be possible. There was also ambiguity over what may or may not constitute a conflict of interest (for both patient groups and clinicians) and clarity was sought, particularly in recognition that many specialists in rare conditions are likely to be involved in the trials of medicines they are being asked to represent at PACE.
  - A question was raised about new medicines and homecare. Although this is not a matter covered by the Montgomery review, it was considered a topic of interest and one which the CPG may wish to consider in the future.
  - There remains confusion regarding the IPTR and PACS process – regarding when each is used and whether IPTRs will ultimately be completely replaced. It was noted that there is little publicly available information on the status of the IPTR to PACS transition. It was also raised that on many Health Board webpages the need for 'exceptionality' remains in IPTR policy (this was modified by the 2014 access to new medicines recommendations to remove the requirement for exceptionality). Concerns were raised that there may have been a return to an issue identified a number of years ago, where IPTRS and PACS are not being applied for because of uncertainty over which process to use and clinicians not applying on behalf of patients where they feel there is a high chance of rejection – when IPTR/PACS are not applied for in this instance, it will not reflect in the Scottish Government statistics regarding access to medicines through these processes.
  - A query was raised regarding the suitability and robustness of the proposed changes with regards to emerging gene treatments.

- A general point was raised that, in the absence of a clear and understandable policy on IPTRS/PACS, there is a risk of postcode lotteries emerging.

Action: The Cross Party Group will write to the Cabinet Secretary for Health and Wellbeing to ask for clarification on several matters raised during the discussion and to request an update on the implementation of the Montgomery recommendations.

- **Topic Two: Care Coordination and Specialist Nursing**

A frequent issue raised by those affected by rare, genetic and undiagnosed conditions is that there is nobody to coordinate the multiple elements of care and treatment that their condition, or conditions, necessitates. This can mean that they do not receive the information and support that they need, that they have to tell their story over and over to health professionals, or that they feel lost on the health care system. CPG attendees were invited to share their experiences of good and bad practice regarding coordinated care and to suggest improvements.

Key points raised:

- There are some excellent examples of good practice – including Bardet Biedl Syndrome specialist clinics which provide coordinated, multi-disciplinary care with the support of a specialist clinical nurse. The benefits include better care planning for the patient, less emergency admissions and better emergency planning, easier condition management, time and cost saving for the patient and, ultimately, cost savings for the NHS.
- It was noted that specialist nurses, where they exist for certain conditions, offered great benefit. Specialist nurses are well informed, well networked and an excellent point of support and advice to patients and families. It was also noted that specialist nurses have a key role to play in coordination of multidisciplinary care and managing data collection.
- For other conditions, such as sickle cell, the lack of a specialist nurse and appropriate care coordination is frustrating. It was noted by a representative for the condition that no specialist nurse for sickle cell exists in Scotland, despite there being evidence from post holders in England that specialist nurses offer piece of mind to families, saves families time and money, reduces emergency care and promotes better symptom management at home. **Action: Bob Doris to raise this matter.**
- The £2.5million specialist nursing fund, which was announced in January 2015 was raised and it was noted that there was little in the way of public information on how this fund had been allocated (only £700,000 allocations to motor neurone disease was known about). **Action: Bob Doris to ask the Scottish Government about the status of this fund.**
- It was noted that for many, care coordination may exist for a short time near diagnoses, but ongoing support is not available. For some,

care coordination led by a health care professional or patient group representative does not exist – 'the person who coordinates my care is me".

- Coordination of care, if an individual moves between health boards, is a challenge. Living rurally in Scotland was also considered a challenge for effective care coordination and there was a recommendation that specialist centres be supported to provide training for local staff.
- Travel to specialist clinics based in other parts of the UK can be valuable when there is access to multidisciplinary teams – however, this often comes at a burden to patients and families (financial and time). There can be frustration when care could be delivered in Scotland because the component parts exist, but there is no coordination.
- Managed Clinical Networks were viewed positively as a way to manage coordinated care, but it was noted that for many conditions such networks do not exist. For some, experience suggests that developing specialist services for rare conditions when can be made more difficult when the condition is considered 'not rare enough' or only effects one or two patients in Scotland.
- Cystic Fibrosis specialist nursing is an example of good practice – where there is an established model, standards of care and regular review.
- Centres for rare diseases in England, such as the centre in Birmingham, were cited as good practice and it was considered that a rare disease centre for Scotland should be on the horizon.
- Specialist nurses, with knowledge of rare diseases in general, rather than a specific condition focus, would be beneficial. The former single gene complex needs service could be a basis for a rare disease service, where patients have access to a rare disease specialist nurse who would be responsible for coordinating care, referring to appropriate services and providing support and information. It would not be necessary for the specialist nurse to have in depth knowledge of specific rare diseases, but they would be expected to have knowledge of the rare disease landscape, where services exist and where to source appropriate information – there would also be an opportunity for data collection.
- Funding for specialist nurses for rare conditions often rests with third sector organisations – which can be challenging for small organisations with limited resources. An example of third sector funding was shared, where nurses for myasthenia were funded by a charity – however, after a few years NHS took over funding and the role changed in a manner which reduced the focus on myasthenia – this is a challenge for NHS funded or joint funded specialist nursing models. Further, it was noted that recruitment for specialist nurses for specific conditions can be difficult as often it is not perceived as good for career progression.
- It was considered that specialist nurses for rare conditions could lead to improved patient outcomes and cost savings for the NHS. It would be valuable to consider mapping existing specialist nursing roles for rare conditions in Scotland.

- There was a general consensus that a pilot for a rare disease specialist nurse may be valuable (Roald Dahl Nurses and the Cambridge rare disease networks were cited as examples to consider).

Next steps – Natalie will invite further comment from CPG members by email and will post a call for evidence in the Genetic Alliance UK newsletter. The topic will be revisited at the December meeting.

- **Any other business**

  No other business was raised

- **Date of Next Meeting**

  The next meeting will be held on Tuesday 5th December in Committee Room 3 between 1pm and 2.45pm.

Attendees and Apologies

Salena Begley MBE	Family Fund	Attendee
Amy Caffrey	British Liver Trust	Attendee
Harriette Campbell	Representing Sickle Cell	Attendee
Michelle Conway	Alexion	Attendee
Bob Doris (Chair)	MSP	Attendee
Elizabeth Dougan	Office of Rare Conditions, Glasgow	Attendee
Jane Ferguson	Ettrickburn	Attendee
Natalie Frankish (Secretariat)	Genetic Alliance UK	Attendee
Simon Flynn	Genzyme	Attendee
Sam Graham	Biogen	Attendee
Yvonne Hughes	Cystic Fibrosis Trust	Attendee
Lynn Laidlaw	Vasculitis UK	Attendee
Lesley Loeliger	PNH Scotland	Attendee
John Miller	Action Duchenne	Attendee
Susan Rees	UK PIPs	Apologies
John Scott MSP	MSP	Attendee
Lynn Shields	TSA	Apologies
Arlene Smyth	TSSS	Attendee
Lynn Stewart	MyAware	Attendee
Rebecca Stewart	Teddington Trust	Attendee
Margarita Sweeney-Baird	BBSUK and Inclusive Skating	Attendee
Sandra Thoms	Fragile X Society	Attendee
John Wallace	NLRP12	Attendee