COMMUNITY CHECK-IN NOTES

Tuesday 29 September: Impact of Covid-19 on roll-out of genomic medicine

Organisations represented:

- Duchenne Family Support Group
- Duchenne UK
- Hereditary Spastic Paraplegia Group
- Huntington's Disease Association
- Pitt Hopkins UK
- PKD Charity
- SWAN UK
- Wolfram Syndrome UK

Genetic Alliance UK staff attending:

- Jayne Spink (JS), Chief Executive
- Amy Hunter, Director of Policy
- Lauren Roberts, Director of Policy
- Nick Meade, Director of Policy
- Natalie Frankish, Policy and Engagement Manager – Scotland
- Emma Hughes, Policy and Engagement Manager – Wales
- Sophie Peet, Policy and Public Affairs Officer
- Izzy Rundle, Communications and Support Officer

JS welcomes all in attendance and welcomes our invited guest, Alex Pickard (Policy and Strategy Lead, Genomics Unit at NHS England), Dr Vinod Varghese (Clinical Geneticist at the All Wales Medical Genomics Service) and Professor Zosia Miedzybrodska (Service Clinical Director of Genetics for NHS Grampian).

Presentation from Alex Pickard:

- Introduction on NHS Genomic Medicine Service in England
- Impact of Covid-19 on delivery of program – they did see an impact across genomic testing service, mainly redeployment of staff/equipment and staff absence. In order to manage this they worked to develop a prioritisation of testing during the pandemic. Prioritisation is no longer in

place and back to delivering service as normal, but conscious that increase in cases may impact service again, so continuing to plan for this.
- Introduction of new boundaries, rare disease specialist testing and test directory – currently on pause, using existing boundaries and providers.
- NHS Genomics Laboratory Hub Mobilisation – have been able to continue to progress on most of this, including work to consolidate the existing laboratory infrastructure. Have done post-Covid planning for delivery of specialist testing model (97% transition in Autumn 2020 and 3% in April 2021).
- Development of large pan-cancer next generation sequencing panel supporting more streamlined testing and work towards reducing turnaround times.
- Funding for cancer genomic testing used to be part of national cancer tariff, that funding has now been unbundled for standardisation and efficiency
- Whole Genome Service is at beta stage of testing has been undertaken (early 2020) – over the last couple of months using acceptance testing which commenced on 20 July 2020 – not actual patient samples, but test samples. Front end of pathway working smoothly, some issues to be worked through. Hoping testing will be completed by end of October 2020, a checklist needs to be completed before it goes live.
- 100k Genome Project – ramped up the return of findings during Covid-19 lockdown. 91% of rare disease results have been returned to clinicians, asked that 100% be returned by end of September/early October. Huge amount of progress made over last six months.
- Genomic Medicine Service Alliance – working to put in place seven alliances to be attached to the seven Genomic Medicine Hubs. Provider selection process has been impacted by Covid-19.

Questions:
- Participant asks whether hubs will share their data to form incidence of a particular gene.
- AP says they will be gathering this information on an anonymous basis and will look at how they can cascade this appropriately.
- Participant asks for clarification on 'geography'
- AP describes how the country will be carved up.

Presentation from Dr Vinod Varghese:
- All Wales Medical Genomics Service (AWMGS) is a national service for the whole of Wales, common protocols and pathways across Wales and funded by specialist commissioners.
- 100K Genome Project – Wales a bit late joining this study. Recruited 439 samples from 154 family units – diagnosis in 21%.
- Since the Test Directory was published last year, AWMGS commissioned to offer all tests approved for test directory.
- Since August 2020 have offered foetal anomaly gene panel for pregnancies.
- WINGS (Wales Infants and Children's Genome Service) – rapid whole genome sequencing for seriously unwell children in neonatal/paediatric intensive care units. 12 cases so far with a diagnostic rate of 42% to 50%.

Covid-19 impact:
- Laboratories never stopped
- Face-to-face clinics stopped towards end of March and replaced with telephone and video consultations — well received by many.
- Since August there has been one counselling room for face-to-face clinics where necessary.

- Department meetings and teams moved to Teams.

Presentation from Professor Zosia Miedzybrodzka:

- Zosia shares 'E's story, a patient that got a diagnosis through a targeted clinical exome testing. In this case they were able to find a very specific gene – family found it very useful because it helped future family planning and are also able to trial treatment for this child.
- Scottish NHS genetic centres – there are four genetic centres but distance between centres is much greater in other parts of UK. We have a service tailored to our geography and rurality. Notes different ethos NHS Scotland to NHS England.
- Genetics labs in Scotland – labs commissioned in each of four centres and funded by NSD – a distributed service model rather than hub and spoke.
- A lot of testing has been done in Scotland.
- Notes that at point England tripled genomics funding, nothing in Scotland. Strategy for Scotland has been delayed due to Covid-19.
- Standard genetic testing.
- In Scotland they are looking at the spectrum of testing available and how these can be used. Notes interpretation burden of whole genome sequencing. WGS through SGP - yield of about 20% in answers for patients/families.
- Four genetic centres.
- In Scotland currently funded for a 'bridge to a Scottish strategy' – expanding targeted panel testing to hundreds of genes for more people. Establish trio based exome tests as a standard service using
- Deciphering Developmental Disorders pipelines. Continue trio-based genomes with Genetics England. 100K Genome Project – caused a lot of paperwork and hold ups.
- Now funding NHS Scotland genomics data store. Covid-19 means a new bridge of funding until a new Scottish strategy.
- One question is how best to use clinical genetics staff in the future – asking about the follow up and ongoing support.
- Impact of Covid-19 was that staff and equipment were redeployed, Glasgow team were redeployed to lighthouse lab and some counselling staff redeployed to contract tracing programme, however urgent and as much non-urgent testing as possible continued. Were able to get reports back to patients and moved to approximately 80% virtual clinics and beginning to reopen the most urgent appointments/clinics. Now redesign of working with community hubs.
- Looking forward, hoping to develop a new strategy for Scotland.

Discussion:

- Participant asks what is your experience of giving results via video/phone consultations?
- ZM notes some patients prefer video appointments, some don't – admin staff call the day before to test system, ask questions and explain process – this allows to triage those who want to continue, those who want phone call and those who want face to face.
- ZM notes having had to give very bad news over video/phone even before Covid-19 and notes how the comfort of being in their own home can help. But notes that it is a very personal choice

and typically preferred to be face to face (especially when child has a learning difficulty - is the focus of urgent appointments).

- ZM notes some hubs in place in Scotland where patients can be assisted in using technology and notes confidence in using it in Highlands because it has been used there for a while.
- VV notes the positive nature of virtual/phone clinics, but has challenges and notes does not replace a face to face meeting. Video conference much better than telephone because of non-verbal cues. Also notes the difficulties with technology not always being reliable.
- AP agrees with what has been said – feedback is that a mixed model is preferable and must reflect different perspectives. AP says anecdotal evidence that video conferences have benefited families that have multiple appointments as reducing travel time.
- JS invites attendees to share their experience – one particpant notes her experience of receiving a diagnosis at home and how valuable that has been.
- ZM is clear of the importance of mixed model.
- One participant notes potential benefits of receiving bad news in the comfort of own home – but asks where is the support when families are on their own and left with their emotions?

A.O.B.

- JS reminds all that next week we will be discussing telehealth and care coordination.

Zoom chat discussion:

- (Participant) Is there a website for all the English GLHs? With patient info for referrals?
- (AP) More information about the Genomic Laboratory Hubs can be found here: https://www.england.nhs.uk/genomics/genomic-laboratory-hubs/.
- (AP) Many of the Genomics Laboratory Hubs also have their own websites which provide more information about patient referrals.
- (Participant) Zosia and Vinod, HSP is a rare disease. I estimate 909 Wales and 180 in Scotland. Different communities have different incidences as Zosia has mentioned. How will this be recognised difference and patients with common genes referred to the same service.
- (ZM) In some areas in Scotland we now have hubs where patients can be assisted to use the technology.