December 2022 | Vol. 5

RARE DISEASE POLICY NEWSLETTER

The Quarterly Newsletter of the UK Rare Diseases Forum

THE LATEST FROM RARE DISEASE POLICY

ABOUT THE NEWSLETTER

Welcome to the fifth edition of the UK Rare Diseases Forum Newsletter. This is a chance to provide you, the rare disease community, with the updates on rare disease policy from each of the four UK nations. Additionally, we'll highlight relevant news stories and upcoming events that you can get involved in.

This newsletter is part of the UK Rare Diseases Forum which is our primary form of engagement with individuals across the rare disease community. The online Forum platform offers an active discussion space for community members and a repository for the upload of governance structure papers.

This newsletter covers all four nations of the UK and is published to members of the Forum online platform. We welcome contributions and comments from members on any of the content posted here. For more information on the newsletter or the Forum, please contact gset@dhsc.gov.uk.

NEWS

Charities Partner to Strengthen antimicrobial discovery

Cystic Fibrosis Trust and LifeArc and Medicines Discovery Catapult have joined the Cystic Fibrosis Antimicrobial Resistance Syndicate. The Syndicate aims to address unmet patient needs by linking people with CF with leading experts across industry, academia, and the NHS. Together, this cross-sector consortium is identifying and tackling drug discovery hurdles to accelerate the development of CF antimicrobials and infection diagnostics, and the speed at which they can reach the people who need them. The development of frequent and persistent infections that are difficult to treat leads to a progressive decline in lung function for people with CF. Treatment failure due to the development of resistance is frequently seen in people with CF, and the consequences of this can be devastating.

Further information is available here.

Genomics in the NHS: A Clinician's guide to genomic testing for rare disease

On Monday 21st November, the Genomics Education Programme (GEP), working in collaboration with St George's University of London, launched the first of two practical courses for clinicians involved in ordering genomic tests: 'A clinicians guide to genomic testing for rare disease'.

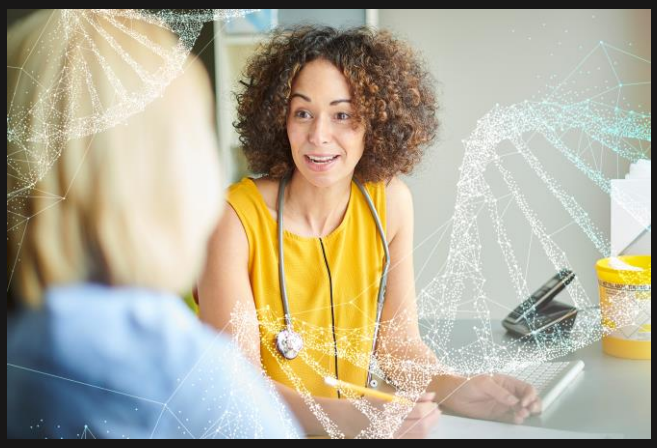
Hosted on the FutureLearn platform and set over two weeks, the course provides a practical introduction to genomic testing for rare disease and support clinicians in ordering tests and feeding back results to patients, including tutorials on how to use the National Genomic Test Directory and other resources.

(continued overleaf)

NEWS

continued

Learners will hear from a wide range of people, from genomics specialists to patients with lived experience of rare disease, to help contextualise the scope and importance of the service.



Learning is delivered through a variety of media, including video tutorials, interviews, animations, articles and discussion steps. Interactive elements, including quizzes and activities, allow learners to demonstrate their knowledge and further discussion is encouraged in the comments below each step.

For further details or to sign up, please visit the course page.

PROMS and PREMS

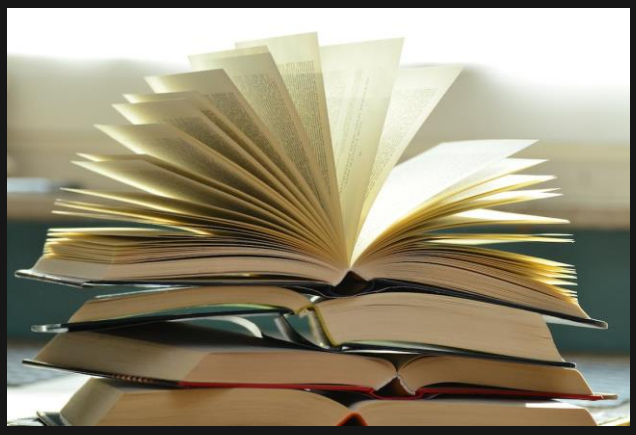
The launch of the Syndrome Without A Name (SWAN) clinic in Wales has afforded the Rare Diseases Implementation Group (RDIG) with the opportunity to develop patient reported outcome and expectation measures, which will be part of the evaluation of the service, which has been funded by the Welsh Government for two years. Laying the foundation for others to share learning on our pioneering service development, our Value Based Care team and core clinical and planning colleagues have submitted the first service evaluation application including patient-reported outcome measures (PROM) datasets for approval.

In addition, we are excited to be co-producing patient-reported experience measures (PREM) questions alongside the SWAN UK Welsh members to support continued service redesign to improve the patient experience. Our continued conversations internationally are supporting this development to ensure collaboration is at the heart of improving service delivery for patients with Rare Diseases.

NEWS

Education

Raising awareness of Rare Diseases as a core component of the Wales Rare Disease Action plan has been a priority for RDIG since the plans launched in June 2022. We are luckily to have been able to partner with Medics4RD, in developing two primary care focused educational events in Wales in early 2023. Raising the profile of Rare Diseases in primary care, highlights the frequency that clinicians may expect to see individuals in their practice, but also where they can find help to manage these circumstances to support their patients. Combining patient stories and generic genomic education, we are utilising Medics4RD's experience in these training opportunities, and alongside the support of Health Improvement and Education Wales (HIEW) we will be able to offer these resources out across Wales.



Techniquest

Genomics Partnership Wales took over Techniquest, an interactive science centre, in South Wales in October for an evening extravaganza – Genomics After Dark. The event showcased a range of activities to improve awareness and understanding of the fascinating world of genetics and genomics.

The event included DNA extractions, public talks focussing on ethics and genomics, interactive stands as well as the opportunity for attendees to experience Techniquests' own hands-on exhibits and star tours in the planetarium.

Over 300 attendees took part in a diverse range of engagement activities; organised by Wales Gene Park and bringing together research groups and organisations from across Wales. There was some great feedback about the event, one attendee commented; 'the whole event completely blew my mind'.

Genetic Alliance UK and Office for Rare Conditions

The Scottish Government is providing funding to Genetic Alliance UK and the Office for Rare Conditions to deliver a suite of events addressing the Awareness Raising priority. These events, a schedule for which will be available in due course, will seek to better understand the needs of healthcare professionals and develop corresponding resources.

NEWS

Genomics Delivery Plan for Wales 2022-25

The Genomics Delivery Plan for Wales 2022-25 was published on 1st December and sets out our approach to mainstream genomics within NHS Wales. The plan forms a key part of our ambitions for the transformation and redesign of diagnostic pathways, in line with the aspirations of the new Diagnostics Recovery and Transformation Strategy for Wales 2022-25 currently being worked up.

The Genomic Delivery Plan for Wales will follow four delivery themes:

- Delivery Theme 1: A Focus on People
- Delivery Theme 2: Clinical Services
- Delivery Theme 3: Research and Innovation
- Delivery Theme 4: Enablers

The plan aims to build on the work of the first genomics strategy and ensure that Wales can deliver our vision of 'Working together to harness the potential of genomics to improve the health, wellbeing and prosperity of the people of Wales'.

There has already been significant advancement in genomics highlighted in June 2021 in the Genomics Partnership Wales *Genomics for Precision Medicine Strategy - Four Years in Review* supported by increased investment from Welsh Government, they have seen new technologies adopted, delivering real benefits for patients, including:

- Wales Infants' and Children's Genome Service (WINGS) - the first in the UK to routinely provide screening for acutely unwell children with a likely underlying genetic cause;
- Cymru Service for Genomic Oncology Diagnoses (CYSGODI) – using genomic testing to deliver personalised cancer treatments;
- Pathogen Genomics Unit (PenGU) - Sequencing and sharing more than 200,000 viral genomes during the pandemic, placing Wales in the top ten globally and having a direct impact on patients and our wider society.

NEWS

Screening

The UK National Screening Committee has started work on reviewing the case for screening for spinal muscular atrophy (SMA).

After its most recent review of the evidence in 2018, the UK NSC recommended not to screen because at the time there was:

- limited evidence on how well the test predicted SMA
- no evidence to show how effective a screening programme would be
- no evidence for effective treatments for people who did not show symptoms of SMA
- limited information about an emerging treatment

Since the 2018 review, there have been significant developments that the UK NSC will consider. These developments include that several drugs can now be prescribed for SMA on the NHS. For example, in England the drugs are available through a National Institute of Health and Care Excellent (NICE) managed access agreement (MAA). This agreement enables patients to access treatment while NICE collects more data before making a final recommendation on their use.

The new evidence review project has 3 objectives:

- **Objective 1:** Conduct a review of available decision analytic modelling studies and cost effectiveness evaluations which address newborn screening for SMA in the era of new treatments
- **Objective 2:** Develop an evidence map of published studies and evaluations of polymerase chain reaction (PCR) based screening and treatment in pre-symptomatic SMA
- **Objective 3:** Propose (scope) a plan for a future modelling study with clinical and cost effectiveness outcomes

The School of Health and Related Research (ScHARR) at the University of Sheffield has been commissioned to carry out this work and is keen to engage with stakeholders. Some key stakeholders will play an active role in workshops as part of the model scoping part of the project. This is a substantial project and work is likely to continue throughout 2023.

NEWS

Haemophagocytic Lymphohistiocytosis publication

A landmark paper has been published on Haemophagocytic Lymphohistiocytosis (HLH) in HemoSphere.

This is the first nation-wide population-based estimate of the incidence of HLH across all ages. This is also the first time that results of linked National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) and National Cancer Registration and Analysis Service (NCRAS) data have been published. HLH is associated with high mortality rates in all age groups, particularly those with underlying malignancy.

The study found that the incidence of diagnosed HLH in England has quadrupled between 2003 and 2018. There were 1674 people with HLH diagnosed in England between 2003 and 2018. Substantial variation in the incidence occurred with inflammatory rheumatological diseases/ inflammatory bowel disease-associated HLH, increasing more among the younger age groups, whereas in older age groups, the largest increase was seen with haematological malignancy-associated HLH. These findings highlight areas in which early diagnosis or preventative measures could be developed.

Northern Ireland All Party Group (APG)

The NI All-Party Group (APG) on Rare Disease held their first face to face meeting at Parliament Buildings in Stormont on Monday 10th October. The function of the Rare Disease APG is to provide a much-needed voice at the NI Assembly for this much under-represented group, raising issues affecting the community and, additionally, increase awareness of rare conditions within the NI Assembly.

Mark H Durkan MLA (Member of Legislative Assembly) chaired the meeting, and a letter of endorsement of the APG was shared from the NI Health Minister Robin Swann, “This is a much-needed forum to give a voice to our rare disease community at the Northern Ireland Assembly, and to promote the needs of people living with rare conditions and their families”.

MLAs also had the opportunity to listen to guest speakers giving an overview of the NI Rare Disease (RD) Action Plan, a ‘Spotlight’ on the Rare Condition PKU (Phenylketonuria) and a ‘Spotlight’ on the Cost-of-Living Crisis and how this is impacting members of the rare disease community. The MLAs in attendance were very engaged and agreed that the APG would be a very positive vehicle where all parties would work together to highlight the issues shared at the meeting to support the rare disease community.

NEWS

Northern Ireland All Party Group (APG) (continued)

At future meetings it was agreed to continue to 'Spotlight' an individual rare condition to help the MLAs gain an understanding of what is like to live with a rare condition. It was also agreed to 'Spotlight' current issues affecting the rare disease community and to provide updates on the DOH led NI Rare Disease Action Plan.

UK Rare Disease Research Platform (MRC-NIHR)

The UK Medical Research Council and National Institute for Health and Care Research (NIHR) call for outline proposals to form the UK Rare Disease Research Platform closed on 8 November. The outlines will now be assessed by an expert panel and successful applicants invited to submit a full proposal to the next round. Around ten nodes will be funded with work getting underway by mid-2023. The Platform will support the coordination of UK rare disease research and address tractable research challenges, with the ultimate aim of a step change in the mechanistic understanding, diagnosis and therapy of rare diseases. The Platform will fully engage with the diverse range of crucial stakeholders in rare disease research, including patients and families, academic, clinical and industry researchers, policy makers and regulators.

POLICY UPDATES

Wales

The recent RDIG meeting was well supported by clinicians and planners across Wales. The continued direction of the group is now focused on the implementation of the Action plan following the successful launch in June 2022. Current priorities include developing an oversight of service provision of those patients with Rare Diseases, which we are collating with the help of a reporting template created and based on the outcome measure defined in the plan. To date, this has prompted two out of our seven health boards to develop strategic groups to focus on local gaps and drivers for improvement in service delivery.

Mapping data of those with rare disease is an immediate challenge, and essential for health boards to understand the challenges they face, as well as enabling patient to access research studies to further their ability to manage their quality of life and treatment options. However, conversations with the Congenital Anomaly Register and information service (CARIS) in Wales and the SWAN Clinic have provided the opportunity to consider small scale pilot, and the potential of a more robust registry in Wales.

Collaborating across the world, in particular with Australia and the US, has enabled the lead clinical team in RDIG to share learning regarding education, research and service developments, which in turn is highlighting the examples of good practice that will forward clinical decision making and improve patient care.

Several opportunities to collaborate nationally and internationally have been developed as part of the MRC/NIHR Rare Disease research node funding, and we look forward to sharing more details of this in the next newsletter.

An adult clinical fellow for rare diseases has been appointed, and with over 135 applicants the successful clinician will be involved in the evaluation of the SWAN clinic, research opportunities and potentially support the **R**are D**IS**ease **E**valuation (RISE) survey UK wide on Rare Disease knowledge in medical undergraduates.

The formal launch of the SWAN went ahead on the 25 October, which included national coverage on the BBC.

POLICY UPDATES

Scotland

Scotland's Rare Disease Implementation Board (RDIB) last met on 22 September 2022. Discussions included views on forging closer links between RDIB and the Scottish Genomics Network, and progress towards the publication of Scotland's Action Plan for Rare Disease. Dr Jonathan Berg, RDIB Chair, will shortly be stepping down to take up the role of Clinical Lead for the Scottish Genomics Network. A new RDIB Chair will be sought in due course.

We intend to publish the first iteration of our Action Plan by the end of 2022 and have now put in place task and finish groups within our Implementation Board to focus members' expertise on appropriate priorities. Our Action Plan will take into account wider work to reassess priorities across Health and Social Care in the Scottish Government as we work through our recovery from the pandemic. This will include making better strategic use of existing resources, and ensuring that actions are realistic and tailored to those who will benefit, resulting in a stronger and interlinked Plan. To discuss the implementation of our Action Plan and ensure that our actions will be prioritised with the rare disease community in mind, we are planning a Workshop to be held in the new year, date to be arranged.

Northern Ireland

The NI Rare Diseases Action Plan sets out 14 high-level actions that will be taken forward over 2022-23 and five working groups (WGs) have been established to take forward the agreed actions. Each WG has representation from the NI Rare Diseases Partnership (NIRDP) and policy support provided by Department of Health. Details of the areas covered by the WGs are as follows:

- WG1: Expert Centre, Information Hub, Patient Portal;
- WG2: Registry, Pathways, Access to drugs & treatments;
- WG3: Education & Training;
- WG4: Research;
- WG5: Rare Diseases Champion.

A regular update on progress from each WG is presented to the Northern Ireland Rare Diseases Implementation Group (NIRDIG).

NIRDIG meets every two months and, at each meeting, the DoH Policy Team provides a summary update of discussions of the UK Rare Diseases Forum, the UK Rare Diseases Framework Board and the online forum platform for consideration and discussion.

POLICY UPDATES

England

The England Rare Diseases Framework Delivery Group is the group responsible for developing and monitoring the delivery of rare diseases action plans for England. Following the publication of the first England Rare Diseases Action Plan in February 2022, the Delivery Group continues to meet regularly every 8 weeks to report on the progress of commitments from the first action plan, and work towards developing new actions for inclusion in future plans.

At each of the two meetings since the last newsletter, delivery partners have reported on their commitments. All are reported to be on track for delivery, although pressures associated with workstreams in NHS England, such as monkeypox, have been highlighted as a risk associated with delivery. Delivery partners are now working to finalise new actions and commitments for inclusion in England's 2023 Rare Disease Action Plan, which we expect to publish in February 2023.

In the September meeting of the England Rare Diseases Framework Delivery Group Professor Iolo Doull (Executive Medical Director, Welsh Health Specialised Services) gave a presentation on the Wales Syndrome Without A Name (SWAN) Clinic. Members of the Delivery Group discussed how future actions plans could better support people living with non-genetic and undiagnosed rare conditions. At the November meeting members discussed a very early draft of England's 2023 Rare Diseases Action Plan. The Delivery Group are next meeting on 14th December.

On Wednesday 2nd November, the DHSC team held the first meeting of the England Rare Diseases Action Plan Patient Advisory Group. This group has been established in partnership with Genetic Alliance UK to amplify the voice of people with lived experience of rare conditions in providing feedback on drafts of England's 2023 Rare Diseases Action Plan. This initial meeting was attended by over 20 stakeholders, who provided value insight and feedback into the initial outline of the 2023 Action Plan. There will a second meeting of this group in early December, followed by a third meeting after publication of the 2023 England Rare Diseases Action Plan to seek further reflections.

EVENTS

Community events

UHC Day 2022 – Universal Health Coverage for Rare Diseases. 12th December

Whole genome sequencing for newborns: updates, developments and future perspectives. 20th January 2023

The Festival of Genomics and Biodata – 25th and 26th January 2023. This is the UK's largest genomics event and is a place for the global genomics community to meet, learn and get inspired. **25th and 26th** January 2023

Rare Disease Day – 28th February 2023 - Keep a look out for organisation events leading up to the biggest day celebrating and raising awareness for people affected by rare, genetic and undiagnosed conditions at https://geneticalliance.org.uk/events/rarediseaseday/

Governance events

Wales Rare Disease Implementation Group meeting: 6th December at 12.30pm.

England Rare Diseases Framework Delivery Group meeting: 14th December at 3pm and 25th January 2023 at 10am.

UK Rare Disease Forum meeting: 9th January 2023 at 10am.

UK Rare Disease Framework Board meeting: 10th May 2023 at 10am.