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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 second 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

UN Resolution on Rare Diseases

The United Nations Resolution on Addressing the Challenges of Persons Living with a Rare Disease and their Families was adopted on 16 December 2021. The UK was supportive of the resolution and actively involved in negotiations on the text. The NGO Committee for Rare Diseases, Rare Diseases International and EURORDIS-Rare Diseases Europe campaigned actively for this resolution to recognise the challenges facing the global rare disease community and protect and promote their human rights at a global level. The full text of the resolution can be found on the United Nations website and more information can be found in the announcement on the NGO Committee for Rare Diseases website.

Genetic Alliance UK and ITN productions working on a programme on Rare Conditions

Launching on Rare Disease Day, 28 February 2022 – Genetic Alliance UK and ITN Productions Industry News are co-producing *Shining a Light on Rare Conditions*, a news-style programme raising awareness of people affected by rare, genetic and undiagnosed conditions. The programme will offer honest accounts from medical professionals as well as people living with rare conditions and their families, exploring the link between increased awareness and quicker access to diagnosis and treatment and successful medical developments including genome sequencing. The programme will also highlight what more needs to be done to provide a better future for people with rare conditions, outlining how their quality of life can be improved from birth through developments in screening and early diagnosis.

NEWS

Genomics England sets out vision for newborn genomes research pilot

Genomics England has announced its vision for a pilot research project to examine the potential for using whole genome sequencing in newborn health screening programmes to find and treat rare genetic diseases in newborn babies. The pilot, which will be run in partnership with the NHS, will look for a set of childhood-onset genetic conditions, which may affect their health in early years, that we can do something about. Work has already begun to explore the types of genetic conditions that would be looked for, and Genomics England will be working with the rare disease community and clinical and scientific experts to develop principles for what conditions will be included. More information can be found on the Genomics England website.

Consultation on the Innovative Medicines Fund

NHS England and NHS Improvement and the National Institute for Health and Care Excellence have launched their consultation on the Innovative Medicines Fund, a fund designed to enable patients to benefit from early access to promising new medicines. The fund builds on the success of the Cancer Drugs Fund by supporting patients with any condition, including those with rare and genetic diseases, to get early access to the most clinically promising treatments where further data is needed to address uncertainties. The consultation is open until 11th February and more information including a link to submit feedback online are available via NHS England's website.

NEWS

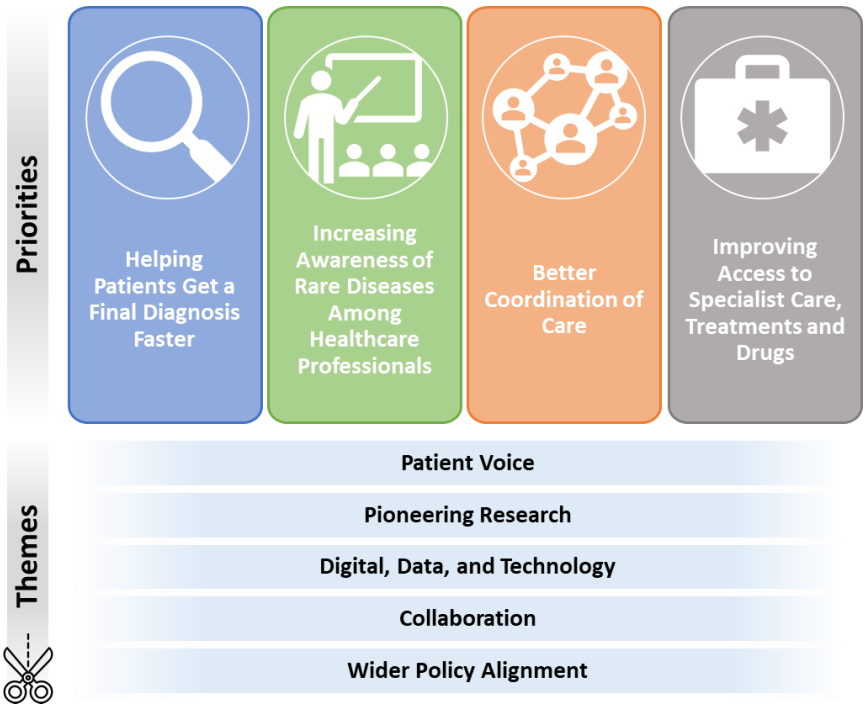
New paper on the benefits of whole genome sequencing for rare disease diagnosis

A world-first scientific study has shown that whole genome sequencing can uncover new diagnoses for people across the broadest range of rare diseases investigated to date. The pilot study of rare undiagnosed diseases involved analysing the genes of 4,660 people from 2,183 families – all of whom were early participants in the 100,000 Genomes Project. The ground-breaking Project, led by Genomics England and NHS England, was established in 2013 to sequence 100,000 whole genomes from NHS patients and their families.

The pilot study, led by Genomics England and Queen Mary University of London and undertaken in partnership with the National Institute for Health Research (NIHR) BioResource, found that using whole genome sequencing led to a new diagnosis for 25% of the participants. Of these new diagnoses, 14% were found in regions of the genome that would be missed by other conventional methods, including other types of non-whole genomic tests.

The paper was published in the New England Journal of Medicine. More information can be found on the Genomics England website.

The UK Rare Diseases Framework



POLICY UPDATES

Scotland

The second meeting of Scotland's Rare Disease Implementation Board (RDIB) was held on 14th October 2021. The minutes will be published in due course once approved. RDIB will next meet on 27th January 2022.

A workshop led by Public Health Scotland was held on 30th November 2021 to discuss the work to date and the future of Scotland's Congenital Anomalies and Rare Diseases Registration and Information Service (CARDRISS). CARDRISS are on track towards becoming a fully operational 'live' registry in March 2022, and will be able to start registering some cases of interest from this January, including a year's worth of retrospective cases. The CARDRISS team are currently looking at how we expand the use of the registry to support the data priority in the Framework and we expect to see a business case for this by the end of this financial year.

The first meeting of our Patient Voices Advisory Group was held on 6th December 2021, and heard views from those with lived experience of rare diseases and professional interests. This useful discussion will help ensure that patients' lived experiences are at the heart of our Action Plan for Rare Disease. The Group will meet again in February 2022.

To further help shape Scotland's Action Plan, two Virtual Involvement Events are being hosted by Genetic Alliance UK in association with the Scottish Government, aimed at people with lived experience of rare diseases. The first is being held on Thursday 20 January. For further details of the second event, taking place Wednesday 16 February at 7pm, please contact natalie@geneticalliance.org.uk.

POLICY UPDATES

Northern Ireland

An online Engagement Event on 27th Sep 2021 had a good turnout, with a mix of voluntary groups, patients, public sector, pharma/industry, carers and healthcare professionals. Of those attending, 27% identified as having a rare disease.

The Engagement Event presented each Priority from the UK Framework in turn and proposed actions for each, based on previous research by the Northern Ireland Rare Disease Partnership. Breakout rooms facilitated discussion, and participants identified their 3 top ranked actions per Priority and fed back to the meeting. Feedback was pooled and analysed to determine the overall stakeholder endorsed top ranked actions for each Priority.

The stakeholder-endorsed top ranked actions provides the Northern Ireland Rare Diseases Implementation Group (NIRDIG) the basis for developing our Action Plan. Over the next number of meetings, NIRDIG will focus on developing metrics, costings and timescales for these top ranked actions, to be included in the draft Action Plan for Northern Ireland. NIRDIG last met on 6th December 2021 when Priority 3 (better co-ordination of care) was discussed. Priorities 1 and 2 were the focus of a NIRDIG meeting on 8th November. Further NIRDIG meetings are to be arranged for early January 2022.

Our key partners have formed a practical scoping group for a NI rare disease registry (NIRADCAR), which is one of the top ranked actions. Colleagues from across all four nations and a wide range of specialties and experience joined the first meeting in November, with a further meeting planned in late December. Quarterly rare disease registry meetings have also been planned to share knowledge and best practice.

An All Ireland network for prenatal and newborn screening has also been formed.

On 12th November the Northern Ireland Rare Disease Partnership (NIRDP) along with Ulster University (S Duguez) delivered a STEMNI Session for Key Stage 5 students (ages 16-18yrs) which had a focus on employability. The session was very well received with over 70 classes across Northern Ireland signing up and over 600 teachers and students participating.

On 23rd November NI Rare Disease Partnership presented at a Pharmacy Forum NI webinar entitled ‘Dealing with disability in pharmacy: Are we doing enough?’ The session was a unique opportunity to underscore amongst the NI Pharmacy community the necessity for awareness and education of rare diseases to facilitate better understanding.

On 29th Nov 2021 Queens University Belfast (AJ McKnight) co-hosted with colleagues at University College Dublin a public webinar on "Executive dysfunction in the classroom: the impact on children with rare diseases".

On 30th Nov 2021, the Northern Ireland Rare Diseases Partnership (NIRDP) hosted with Medics4Raredisease and Queens University Belfast (AJ McKnight) with the Royal College of General Practitioners Northern Ireland to present ‘Top Tips for Managing Rare Disease’.

POLICY UPDATES

Wales

The Rare Diseases Implementation Group is responsible for the development and implementation of the Rare Diseases Action Plan for Wales. The RDIG will work with a dedicated programme manager funded by the Welsh Government, who will be appointed shortly. The RDIG last met on 5th November 2021 and reports to Welsh Government.

There is now a programme of regular meetings of RDIG planned into 2022 to deliver an implementation plan for the new Rare diseases Framework around April 2022 including the development of Task and Finish Groups to discuss each of the four key priorities.

Key highlights reported at the meeting included:

- Welsh Government have approved a business case for the development of a, state of the art, Genomics site in the north of Cardiff. It is an existing facility which will be upgraded, three stakeholders will be joining together in this property.
- The Wales Infants and Childrens Genome Service (WINGS) won the Bypass Award for Innovation in Health Care at the Healthcare Awards 2021.
- Development of All-Wales Guidelines for children with developmental delay. Progress is being made in agreeing the final document.
- Progress on the Syndrome without a Name Clinic (SWAN Clinic).

The Welsh Minister for Health and Social Services has previously agreed to fund a 2 year pilot of a SWAN clinic. The SWAN clinic will offer an all Wales service with physical and virtual outreach services provided on a regional basis in south and north Wales. The key objective of the SWAN clinic would be to shorten the time to diagnosis. Lead Clinicians for the service have now been appointed for Paediatrics, Adult and Genetic Services. The clinic is planning to see new patients in the very early part of 2022.

A framework for outcomes and referral criteria is currently being developed.

In November 2021, two consultation workshops were organised and coordinated by Genetic Alliance UK and the Wales Rare Disease Implementation Group. The aim of the workshops was to engage people affected by rare and genetic conditions in Wales and involve them in the initial phase of the development of a Welsh Action Plan, to implement the Framework. Both sessions were held virtually via Zoom, engaging over 50 people affected by or representing those affected by rare, genetic and undiagnosed conditions across Wales. Following the meetings recommendations in relation to the four priority areas of the UK Rare Diseases Framework, have been developed based on data collected from the workshops to inform the development of the Welsh Action Plan.

There has been a series of meetings held to understand the importance of research and as part of the Welsh Action Plan. The meetings have taken place with the idea of forming an Undiagnosed Research Hub in Wales which could also collaborate on a UK wide basis.

POLICY UPDATES

England

The first England Rare Diseases Action Plan is rapidly approaching completion. The plan is currently undergoing a series of consultation, review and editing stages ahead of publication to coincide with Rare Diseases Day at the end of February 2022.

The England Rare Diseases Framework Delivery Group is the group responsible for developing and subsequently monitoring the action plan for England. The Delivery Group has met every 6 weeks in the lead up to the Action Plan publication, with meetings focussing on the development of actions and appropriate metrics; feedback and comments on the drafting of the Plan; and discussion on the recommendations of stakeholder reports. The next meeting will be held on the 26th January 2022 and will focus on building in further feedback and final revisions to the Action Plan as it nears completion. The group will continue to meet through 2022 to monitor progress on the first action plan and develop actions for the second.

The Department of Health and Social Care (DHSC) has undertaken a great deal of community engagement to feed into drafting England's first Action Plan. We have asked the UK-wide Forum for feedback as the actions have been developed and on a late-stage draft of the document. We have also received feedback through two community roundtables, an online questionnaire, and a workshop held in partnership with Breaking Down Barriers, which focused on health inequalities.

EVENTS

Community events

- Genetic Alliance UK will celebrate Rare Diseases Day with a virtual parliament event on 23rd February. Further details will soon be available on the Genetic Alliance UK website.
- JNetics, a charity for improving the prevention and management of Jewish genetic disorders in the UK, is hosting a series of events as part of their awareness month, GENEuary. A full schedule of events can be found on their website.
- The Rare Diseases Day webinar series has restarted ahead of Rare Diseases Day 2022. These webinars cover a range of topics and experts. More information can be found on their website.
- The Findacure International Rare Diseases Showcase, a celebration of innovative rare disease projects across the world returns for its sixth year from 1st – 3rd February. Free tickets are available on their website.
- On 9th February, The Royal Society of Medicine and Medics 4 Rare Diseases will host 'The unusual suspects: Rare disease in everyday medicine', an online meeting to discuss how rare diseases are relevant in clinical medicine. Tickets are free from the RSM website.
- A series of online events on the use of whole genome sequencing at birth are hosted by the Progress Education Trust in partnership with Genomics England and its Newborn Genomes Programme. Future seminars are to be held on the 16th February and 16th March.

Governance events

- Next Scottish Rare Disease Implementation Board meeting: 27th January 2022.
- Next England Rare Diseases Framework Delivery Group meeting: 26th January 2022.
- Next UK Rare Diseases Framework Board meeting: 3rd May 2022.