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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 third 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 meeting 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, including if you have any contributions for our next newsletter, please contact gset@dhsc.gov.uk.

NEWS

Genetic Alliance UK Good Diagnosis report

Genetic Alliance UK set out to better understand people's experience of diagnosis and to identify what matters most to people on their diagnosis journey through a series of workshops in November 2021. Key themes from the discussions, together with the findings of the Rare Resources (Scotland) and Rare Experience 2020 projects, were used to identify principles of a good diagnosis. The Genetic Alliance UK Good Diagnosis report was published to coincide with Rare Disease Day (28 February 2022) and makes four recommendations to improve the experience of people at the three key stages of the diagnosis journey: the search for a diagnosis, receiving a diagnosis, and following diagnosis. The report is available on the Genetic Alliance UK website.

Health Education England Rare Disease Education Hub

To mark Rare Disease Day 2022 Health Education England's Genomics Education Programme (GEP) launched a new education hub for rare disease, aimed at supporting health professionals to better understand rare disease and support patients and families in their care. The hub features an introduction to what rare disease is, explores how genomics and new genomic technologies can help, and provides guidance for giving the best care. The resource features stories from patients and signposts to other useful educational resources. The GEP is thankful to partners and stakeholders who have contributed to the education hub and welcomes feedback and contribution to inform its evolution in line with the action plan.

NEWS

Timothy Syndrome Alliance and Pulse Infoframe collaborate on new registry for rare CACNA1C-related disorders

Timothy Syndrome Alliance (TSA) is creating a global patient registry for the CACNA1C gene, which encodes the alpha-1-subunit of a voltage-dependent L-type calcium channel highly expressed in the heart and brain. To better understand the impact on individuals with variants in the CACNA1C gene and the natural history and disease progression, the organisation has selected Pulse Infoframe's real-world evidence platform to study the gene that causes CACNA1C-related disorders including Timothy Syndrome and LongQT8. The registry is expected to be live by the end of April. More information is available on the Pulse Infoframe website.

Academy of Medical Sciences workshop on clinical trials for rare and ultra-rare diseases

Attendees discussed innovative ways to overcome barriers to clinical trials for rare diseases at a workshop held across 24 and 30 March 2022 by the Academy of Medical Sciences' FORUM and the Faculty of Pharmaceutical Medicine. Themes covered included barriers to patient participation and involvement, developing meaningful outcome measures, innovative clinical trial designs, and the use of real-world evidence. The workshop included perspectives from researchers, patients, carers, regulators, health technology assessors, industry representatives and more. A full workshop report will be published by the Academy in the coming months exploring how these innovations can be harnessed to improve the future of clinical research delivery in rare diseases.

LifeArc and CSO work together to drive Scottish research into rare diseases

LifeArc and the Chief Scientist Office (CSO) within the Scottish Government announced on 24 March 2022 the joint awarding of a total of £300,000 to researchers at The University of Edinburgh to progress research towards helping people living with rare and complex health conditions. Further information can be found on the CSO website.

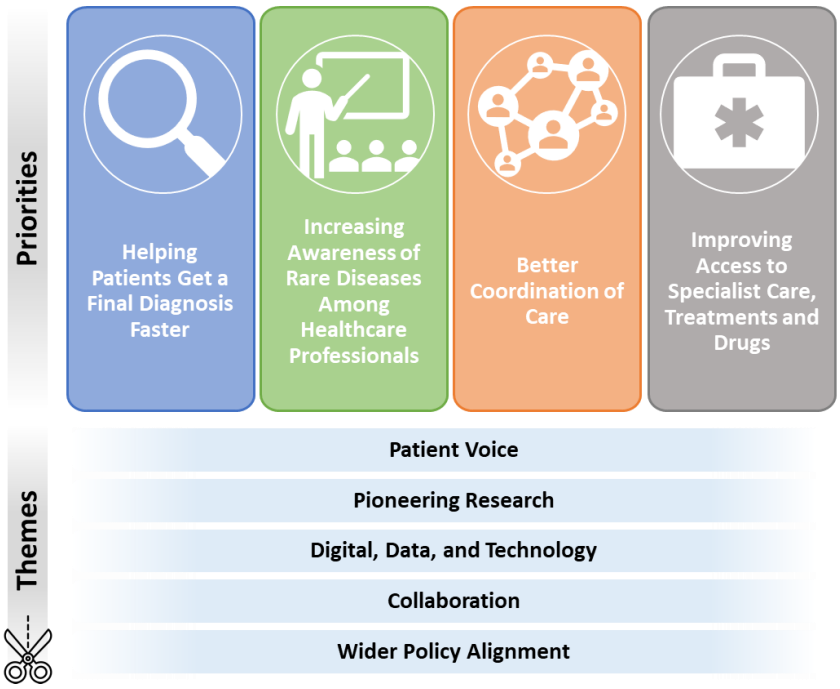
NEWS

MELODY study to improve understanding of vaccination in people with immunosuppression

The pioneering research study MELODY (Mass evaluation of lateral flow immunoassays in detecting antibodies to SARS-CoV-2) opened to recruitment in January. Using data held by the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) at NHS Digital and NHS Blood and Transplant, the study aims to recruit 35,000 people who are immunosuppressed as a result of a rare autoimmune rheumatic diseases, a rare blood cancer or a solid organ transplant. The study will determine immune responses to Covid-19 vaccination and future risk of infection, hospitalisation rates and survival over six months. It will inform vaccination strategies and who could benefit from other interventions.

This ground-breaking study is the first time that people with rare diseases registered with NCARDRS have been invited to take part in research. The study is led by Imperial College and is being funded by the Medical Research Council in collaboration with several health charities including Kidney Research UK, Blood Cancer UK, Vasculitis UK, and the Cystic Fibrosis Trust. People in the UK who have had solid organ transplants can opt into the study. A random sample of people with blood cancer or rare autoimmune rheumatic disease who reside in England are being invited via letter.

The UK Rare Diseases Framework



POLICY UPDATES

Northern Ireland

On 22 March 2022, Northern Ireland's Health Minister, Robin Swann, published the Northern Ireland Rare Diseases Action Plan for 2022/23. The Action Plan, which has been developed by the Northern Ireland Rare Diseases Implementation Group (NIRDIG), sets out 14 high-level actions aligned to the strategic priorities of the UK Rare Diseases Framework, that will be taken forward over the next year, working with healthcare professionals and educators, researchers and commissioners, as well as rare disease patient organisations and those personally affected by rare disease.

Prior to publication of the Action Plan, a stakeholder Focus Group, organised by the Northern Ireland Rare Disease Partnership, met on 3 March 2022 to consider and discuss the Action Plan and whether it met stakeholder expectations. Eleven rare disease conditions were represented at the Focus Group; and feedback was positive, with stakeholders' noting content with the Year 1 actions and encouraged by implementation progress made so far.

In January 2022 the Northern Ireland Rare Disease Partnership (NIRDP) led on a campaign to establish an All-Party Group (APG) for Rare Disease at the NI Assembly to provide a voice for the Rare Disease Community at the highest political arena within Northern Ireland. The campaign led to hundreds of members of the NI Rare Disease Community reaching out to their local MLA's to get involved and NIRDP were delighted to announce on Rare Disease Day that the cross party APG was approved with Chairperson Mark H Durkan MLA and with NIRDP providing the secretariat. For information on the APG please contact engage@nirdp.org.uk

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POLICY UPDATES

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On 1st April 2022 the University of Ulster Interprofessional Module in the School of Health Sciences delivered a problem based learning workshop with people living with Rare Disease to Allied Health students studying Occupational Therapy, Physiotherapy, Podiatry, Diagnostic Radiography & Imaging, Radiotherapy & Oncology, Speech & Language Therapy, and health care science students. The session, with expert patient input supported by the Northern Ireland Rare Disease Partnership, looked at the rare disease journey through interaction with service providers from pre-diagnosis, diagnosis, managing treatment, radiography imaging, and different types of therapies. Students worked in interprofessional groups to generate solutions to enhance patient experience.

England

The first England Rare Diseases Action Plan was published on international Rare Disease Day, 28th February 2022, co-launched by Secretary of State for Health and Social Care Sajid Javid and Parliamentary Under Secretary of State (Minister for Patient Safety and Primary Care) Maria Caulfield. It sets out 16 specific, measurable actions for the next year under each of the 4 priority areas of the UK Rare Diseases Framework. To ensure delivery and accountability, each action lists an owner, desired outcomes and, how we will measure and report on progress.

The England Rare Diseases Framework Delivery Group is the group responsible for developing and subsequently monitoring action plans in England. Over the course of the last year in developing the first Action Plan, the group has focused on the development of actions and metrics, and on the discussion of community feedback and the recommendations of stakeholder reports. The Delivery Group continues to meet regularly on an 8-weekly schedule, with the next meeting on 19 May 2022. Future meetings will include reporting on the progress of our existing commitments as well as continual development of new actions in preparation for the next Action Plan, to be published in early 2023.

The Department of Health and Social Care (DHSC) has actively engaged with the rare diseases community during the development of the England Action Plan, including bringing the draft Action Plan to UK Rare Diseases Forum for comment, a targeted online questionnaire, two community roundtables, and a community workshop focusing on health inequalities (full details on engagement is available in Annex C of the Action Plan).

On 29 March 2022 Secretary of State for Health and Social Care Sajid Javid hosted a roundtable at Great Ormond Street Hospital (GOSH) to discuss the Action Plan as well as ongoing work and challenges for the rare diseases community. The roundtable was attended by 15 stakeholders including those with lived experience of rare disease (invited in partnership with Genetic Alliance UK), representatives of the pharmaceutical industry, researcher-clinicians and representatives of public research bodies. It focused on the themes of health inequalities and research and innovation. The event was reported via blogs on the GOSH and Genetic Alliance UK websites.

POLICY UPDATES

Wales

The Welsh Rare Disease Implementation Group (RDIG) held a workshop on 25 March to agree contents of the new Wales Rare Diseases Action plan with a wide range of stakeholders. A final draft action plan has been circulated to a wide range of stakeholders for informal engagement and feedback. A final workshop with Genomics Partnership Wales Sounding Board will be held as part of stakeholder engagement on 14 April. The next Welsh RDIG meeting is taking place on 22 April 2022 when we are hoping the plan will be signed off for publication by the end of May. A new chair for the Welsh RDIG, Professor Iolo Doull is taking over from the role from 1 May 2022 replacing Dr Graham Shortland who has been in the role since 2014. Professor Doull is currently the Medical Director for Welsh Health Specialist Services Committee having had extensive experience in paediatric respiratory medicine (Cardiff) and involvement in UK-wide initiatives. He has made a major contribution to research in the area of paediatric respiratory medicine and published his research findings widely.

There has been continued work to develop a pathway for the investigation of global developmental delay and intellectual disability and ensure "sign-up" across Wales. This is now nearly complete and there are educational events planned to role out this initiative with paediatricians across Wales.

As a consequence of discussion and engagement at RDIG, Webinars are being established initially in Hywel Dda University Health Board between genetic and general practice services to enhance referral processes and co-ordination of care between specialist and general practice care. If successful this will be rolled out across Wales

RDIG is working with a Welsh University Health Board, industry and Welsh Government (the Welsh Hack) to develop a patient passport for patients with rare diseases. This will be hopefully introduced and be available to all Welsh rare diseases patients in the next six months, working across all relevant NHS IT systems in Wales and hopefully also across UK NHS providers.

A National clinical lead for rare diseases has been appointed from April 2022, Dr Jamie Duckers who is a Consultant in respiratory medicine specialising in cystic fibrosis with Cardiff and Vale UHB. He will play a major part in supporting the Chair of the Rare Diseases Implementation Group (RDIG) and liaising with other "leads" in the UK. Key areas identified for initial work of the clinical lead include development of exemplar pathways for a small number of disorders that can be applied across rare diseases generically. Also he will work with other specialist leads as part of the National Clinical Framework in Wales.

A new rare diseases programme manager has been appointed and is taking up post in April 2022 to assist the RDIG Chair and Clinical Lead. Further, the Syndrome Without A Name (SWAN) clinic is now up and running and information on referral processes have been issued to all health boards in Wales.

POLICY UPDATES

Scotland

The fourth meeting of Scotland's Rare Disease Implementation Board (RDIB) took place on 12 April 2022 and featured a substantive discussion on the underpinning theme of research. Minutes of the meeting will be made available in due course following approvals. RDIB will convene again on 28 April for an extra-ordinary meeting to discuss Scotland's draft Action Plan for Rare Disease. The fifth full meeting of RDIB is scheduled for 17 June.

We will shortly be writing to NHS Scotland Medical Directors and Nursing Directors regarding our tests-of-change bidding process by which we aim to identify a suitable model of care co-ordination for wider implementation in Scotland, using a pilot or pilots from the bidding process.

Drafting of the first iteration of Scotland's Action Plan continues, towards publication in the week commencing 23 May 2022. We have continued our outreach to relevant third sector organisations and to facets of NHS Scotland to ensure we are making the right connections to resources that improve the lives of people living with rare diseases, and of course continuing to engage with the rare disease community. The latter is how we find out "what matters to you" in the context of living with a rare disease, and two further meetings of our Patient Voices Advisory Group have taken place, as well as further online engagement events led by Genetic Alliance UK. Maree Todd MSP, Minister for Public Health, Women's Health and Sport, gave a keynote speech at a virtual reception on 1 March to mark Rare Disease Day in Scotland, and the Minister also published an open letter to the rare disease community in Scotland renewing our commitment to them.

Our registry, Scotland's Congenital Anomalies and Rare Diseases Registration and Information Service (CARDRISS) is undergoing final internal IT approvals for its bespoke software ahead of becoming fully operational and able to register babies. We will continue to support CARDRISS' work on rare diseases in this financial year.

EVENTS

Community events

- Health Education England is hosting their annual #GenomicsConversation week for all health professionals from 20 – 24 June 2022. This year, the event will focus on helping healthcare professionals to feel confident when talking about genomics with both colleagues and patients. Each day of the week will be themed around different areas of genomics communication: from where conversations can happen, to common questions and the 'clinical clues' that may be cause for action, and where to find out more. You can find out more about the week via the Genomics Education Programme website.
- Salivary Gland Cancer UK is holding their next hybrid gathering on Saturday June 25th 2022. Open to all patients, carers, clinicians and researchers with an interest in salivary gland cancers, you can find more details and how to join on the Salivary Gland Cancer UK website.
- The Genomics England Research Summit 2022, taking place Wednesday 4 May in London, will be an exciting hybrid conference where scientists, industry leaders and policy experts will come together to showcase the latest discoveries and strategic direction in human genomics. With four tracks each focusing on a theme – rare disease, cancer, emerging tech, and policy and initiatives – there will be something for everyone interested in the latest genomic developments. This event is open to anyone curious about human genomics, including patients and participants. See the agenda and register for free now at the Summit website.
- The Office for Rare Conditions is hosting a webinar from 5:30pm – 7:00pm on Wednesday 27 April 2022, titled "The Patient Passport or Care Summary for those affected by Rare Conditions." Anyone wishing to register for this event is welcome to do so at the registration webpage.

Governance events

- Scotland Rare Disease Implementation Board – extra-ordinary meeting to discuss draft Action Plan: 28 April 2022
- UK Rare Diseases Framework Board meeting: 3 May 2022
- Wales Cross Party Group Meeting with Genetic Alliance UK, stakeholders and Welsh Assembly Members: 18 May 2022
- England Rare Diseases Framework Delivery Group meeting: 19 May 2022
- Scotland Rare Disease Implementation Board – fifth substantive meeting: 17 June 2022