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RARE DISEASE POLICY NEWSLETTER

The Quarterly Newsletter of the UK Rare Diseases Forum

THE LATEST FROM RARE DISEASE POLICY

News – 2
Policy updates - 11
Events - 15

ABOUT THE NEWSLETTER

Welcome to the sixth 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.

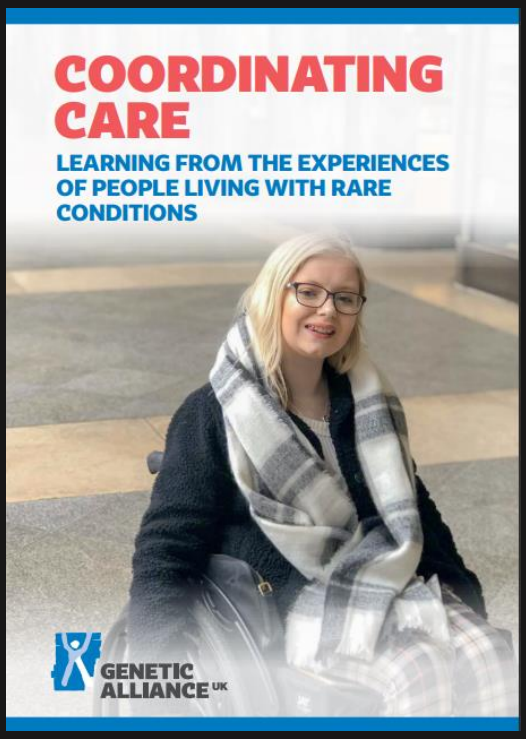
This newsletter covers all four nations of the UK and is published to members of the Forum online platform. We welcome contributions and comments from members on any of the content posted here. For more information on the newsletter or the Forum, please contact gset@dhsc.gov.uk.

NEWS

Genetic Alliance UK

Genetic Alliance UK hosted Parliamentary receptions in Westminster, Holyrood and the Senedd to recognise Rare Disease Day. The events were attended by over 280 members of the rare community in total. We were joined by Eluned Morgan MS, Minister for Health and Social Services, Maree Todd MSP, Minister for Social Care, Mental Wellbeing and Sport and Helen Whately MP, Minister for Social Care in their respective Parliaments where they addressed the rare community.

Genetic Alliance UK also launched the Coordinating Care report which highlights the lived experience of people with rare conditions, shows the value of care coordination and the impact of its absence. This report has been shared with key decision-makers including Scotland's DCMO Graham Ellis who met with Natalie Frankish, Policy and Engagement Manager for Scotland, on Rare Disease Day.



On social media, Genetic Alliance have 46 different individuals and organisations doing takeovers on their Twitter account to raise awareness for Rare Disease Day. 31 MSPs have also shown their support for rare conditions on Twitter.

NEWS

NICE (National Institute for Health and Care Excellence)

For Rare Disease Day, NICE ran a social media campaign on Twitter and Linkedin highlighting their published guidance on 20 treatments for very rare conditions as part of their Highly Specialised Technologies programme. They also focused on the priority in last year's action plan to improve access to specialist care, treatment and drugs for people with rare diseases by highlighting the June 2022 launch of the Innovative Medicines Fund with NHS England, fast-tracking the most promising, cutting-edge medicines. Throughout NICE used #RareDiseaseDay and #RareDiseases Action Plan.

NHS England

NHS England (NHSE) shared a video from Eleanor, Atsushi and baby Reuben from their Twitter and Facebook accounts highlighting how the world's first rapid whole genome sequencing service helped them get a diagnosis for Reuben's rare condition and has allowed them to look to the future with hope. This performed well with almost 8k views and over 700 engagements. The NHS Genomic Medicine Service also shared a video from Professor Dame Sue Hill on Twitter highlighting the second Rare Disease Action Plan and the vital role genomics will play in it to deliver better care for patients. This has had more than 7k views and over 100 engagements. NHSE also shared a video from Sarah Wynn, CEO of Unique, highlighting the impact genomics can have on patients living with a rare condition and the role of the Rare Disease Action Plan in supporting this. This had almost 1k video views and 50 engagements.

NHS England Digital focused on supporting with social media output for Rare Diseases Day, including a quote retweeting DHSC's press release post, and sharing content from the Westminster #RareDiseaseDay2023 Reception, which was attended by Mary Bythell and Peter Lanyon, Rare Disease Lead and Clinical Lead for the National Disease Registration Service. The post received 4,400 impressions and 80 engagements, including 2 replies praising their work and that of the NHS.

NEWS

Genomics England

On 28th February Genomics England (GEL) marked Rare Disease, which aims to raise awareness amongst the general public and decision-makers about rare diseases and their impact on patients' lives.

10 years on from the launch of the very first initiative, the 100,000 Genomes Project – GEL are continuing to drive diagnoses and research into rare conditions.

6,625 participants with rare conditions received a diagnosis which had not been possible by routine care through the 100,000 Genomes Project. By supporting the NHS Genomic Medicine Service, the first healthcare system in the world to offer whole genome sequencing as part of routine clinical care – GEL are still helping to provide answers for people with rare conditions.

But GEL's work doesn't stop there, their clinical research interface provides a safe and effective way for insights and data to be exchanged between researchers and NHS clinical teams, helping to bring new diagnoses, explain symptoms and support onward care for participants.

NEWS

New course: Clinician's Guide to Genomic Testing for Rare Disease

On Rare Disease Day 2023, the Genomics Education Programme officially launched its new online course: Genomics in the NHS: A Clinician's Guide to Genomic Testing for Rare Disease.

This free two-week course provides a practical introduction to genomic testing for rare disease, taking clinicians through the process of using the test directory, ordering tests, and feeding back results to patients. Learning is delivered through a variety of media, including video tutorials, animations, and discussion steps.

Find out more about the course on the dedicated webpage.

NIHR BioResource

To mark Rare Disease Day and publication of the second Rare Diseases Action Plan, the NIHR BioResource published a case study of a family living with a rare eye condition. The family are participants in our Rare Diseases RNA Phenotyping project, one of two NIHR BioResource projects featured by DHSC in this year's Action Plan. The project aims to better understand rare genetic conditions by combining extensive genetic and clinical data and looking for patterns amongst patients, relatives and healthy controls.

Continued overleaf

NEWS

Chloe was diagnosed with Leber Hereditary Optic Neuropathy (LHON) on her 19th birthday and is now registered blind. Just last year, her older brother Tom was also diagnosed with LHON. Chloe, who joined the NIHR BioResource as a patient volunteer in 2016, believes that research is the way forward to better understanding the condition so it can be detected early and quality of life can be improved. Tom is grateful to his younger sister and all others involved in research for their role in developing treatment options such as Idebenone, which has already helped Tom's vision, but was not available to help Chloe at the same stage 12 years ago.

Anyone can join the BioResource as a research volunteer, and researchers can access samples, data and participant recall by genotype or phenotype from over 200,000 consented volunteers by making an application to the NIHR BioResource.

SWAN clinic

The Adult Rare Disease Fellow commenced employment in December and has been busy supporting the (Syndrome without a name) SWAN clinic, in particular focusing on collating outcome data (patient reported outcome measures -PROMS and patient reported experience measures -PREMS) to help us to understand the impact of the improved coordination of care for those who are without a diagnosis. The Fellow is also supporting a proof-of-concept project between RDIG (Rare Disease Implementation Group) and CARIS (Congenital Anomaly Register and information service), looking at how we can create meaningful clinical and condition specific workstreams to improve pathways of care for those with Rare Diseases.

Bevan Commission exemplar programme

One of the clinical champions in Wales has been successful to the Bevan Commission exemplar programme. This programme supports the clinical leaders and influencers in Wales to collaborate and engage across Wales and internationally to harness expertise and experiences that challenge the status quo and inform policy and practice. This funding will enable a new Rare Disease clinic to be funded, expanding to a multidisciplinary team set up across paediatric services in the Cwm Taff Morgannwg University Health Board. It will be tested using patient reported outcome measures (PROM), patient reported experience measures (PREM) and ongoing audits.

NEWS

Care & Respond Patient passport webapp

The 'Care & Respond' Patient passport webapp was launched on Rare Disease Day. Developed in Wales by Science & Engineering Applications Ltd, in collaboration with various patient groups and partners, it was supported by RDIG and received funding from the Welsh Government via the Welsh Health Hack and Welsh Social Care Hack. The webapp aims to support the coordination of care for those with Rare Diseases, highlighting key information (chosen by patients) to provide accurate information in emergency situations to clinical teams regarding their condition and care needs.

Health Education and Improvement Wales

On 2nd March Health Education and Improvement Wales (HEIW) hosted an online Reframing Rare Disease in Primary Care Webinar in association with Medics4RareDiseases. This webinar was aimed at primary care physicians across Wales. The Wales Rare Disease Implementation Group Clinical Lead introduced the webinar and gave an insight into the Rare Disease Plan in Wales. Colleagues from Medics4RareDiseases presented on RAR Disease 101—tips and pitfalls for caring for those with Rare Disease. A valuable insight into living with a rare condition was also given by Same But Different. The event was well attended, and further primary care educational events will be planned. On 4th April, a Welsh Health Circular which provides a clinical guideline for the Early developmental impairment or intellectual disability (WHC/2023/03) was published. The guidance supports clinical decision making for paediatricians who assess children and young people referred because of early developmental concerns.

Cystic Fibrosis

On 3rd March, Wales hosted the 5th Conference 'Cystic Fibrosis trials: building on the new norm' bringing together international researchers looking at ways to address research priorities, public and patient involvement, equity of access to research, research delivery in rare diseases and novel gene therapy research.

A team lead by Welsh researchers, funded by Research for Patient and Public Benefit, have published their latest research in the Journal of Cystic Fibrosis on Sharing decisions on reproductive goals: A mixed-methods study of the views of women who have cystic fibrosis, part of the CF Prosper, funded by Welsh Government, looking at using registry data to improve knowledge of outcomes of women with Cystic Fibrosis choosing to become mothers and what information needs are and how we can improve shared decision making. This work drives forward the body of knowledge of those women living with rare disease contemplating starting a family.

NEWS

Medics4RareDisease

Every year Medics4RareDisease run an annual essay competition along with **Beacon** known as the **Student Voice Prize**. Every year, on rare disease day, we have a short online event called "**Beyond the Student Voice Prize**" where we hear from a few medics who are passionate about rare disease. The aim of the evening was to highlight to current medical students the wide range of opportunities available through a career more focused on rare disease and the exciting further opportunities available.

Hereditary Brain Aneurysm Support

Hereditary Brain Aneurysm (HBA) Support is a new not-for-profit organisation that aims to provide information and support for people affected by hereditary brain aneurysms.

A patient-led organisation, Rebecca Middleton founded HBA Support after her battle for diagnosis and treatment following the discovery of her own family's history.

In September 2022, HBA Support published a targeted literature review carried out pro-bono by Costello Medical. Available to download for free, the review demonstrates the need for more research into prevalence and incidence rates and the genetic factors behind the condition. It also highlights the need for consistent screening guidelines for people at risk.

HBA Support is currently seeking specialists to join a new Advisory Panel. No previous trustee or consultative experience is required to join the panel. However, experience in the following is preferable to help HBA Support to fulfil its vision.

Genetic Research | Genetic Counselling | Neurosurgery | Neuro Nursing | Medical Research | Medical Devices | Charity or Not-for-Profit Organisational Governance | Fundraising/Funding | Health Policy

For more information about the Advisory Panel, including how to apply, or to read the targeted literature review, visit https://www.hbasupport.org/

NEWS

North South Rare Disease Day Celebration

Mark H Durkan MLA and Deborah Erskine MLA sponsored a Rare Disease Day Celebration in the Long Gallery, Parliament Buildings, Stormont on 28th February on behalf of the Northern Ireland Rare Disease Partnership (NIRDP) and Rare Diseases Ireland (RDI) and was supported by the Department of Health NI.

At the in-person event, there was the opportunity to connect with various members of the rare disease community, including people living with rare diseases, rare disease advocates, healthcare providers, researchers, and policy makers over a networking lunch.

The formal part of the event then offered an opportunity for members, unable to attend in person, to listen in via a Zoom Webinar to presentations which included hearing an update on rare disease policies and plans; an introduction and overview of all-party groupings on rare diseases in the NI Assembly and the Oireachtas; and to hear a review of the work of RDI and NIRDP.

The event closed with the launch and first public viewing of a short film from the NIRDP Rare Stories project - a project which worked over 2022 to capture real life stories of people in the NI rare disease community, supported by esc films with funding from the Rank Foundation. The link to the launch film can be seen here.

The successful event saw some 160 people participate dual in-person and virtual event.

Rare Disease Day in Scotland - 28th February 2023

The Minister for Public Health, Woman's Health and Sport, Maree Todd published an open letter to the rare disease community re-affirming the Scottish Governments commitment.

The Scottish Government lit up St Andrews House and Victoria Quay in honour of rare disease day in conjunction with the 4 nations.

The Deputy Chief Medical Officer (DCMO) visited the Royal Hospital for Children Glasgow to participate in a rare disease stand operated by Genetic Alliance UK and the Office for Rare Conditions (Glasgow). The DCMO also took part in a presentation to hear from people with rare conditions and be updated on Genetic Alliance UK's new report on care coordination.

NEWS

15th March 2023

The Minister for Public Health, Woman's Health and Sport, Maree Todd spoke at an online reception to mark rare disease day on 15 March 2023 which was hosted by Bob Doris MSP and run by Genetic Alliance UK and the Office for Rare Conditions in Glasgow.

Publications

December 2022 – Publication of Initial finding of an economic evaluation of Whole Genome Sequencing for Rare Disease - Continuing the sequence? Towards an economic evaluation of whole genome sequencing for the diagnosis of rare diseases in Scotland.

December 2022 – Publication in the European journal for human genetics - NHS genetics centres in Scotland sought to investigate the Genomics England 100,000 Genomes Project diagnostic utility to evaluate genome sequencing for in rare, inherited conditions.

POLICY UPDATES

Wales

Rare Disease Day reception was held at the Senedd on 14 February, hosted by Genetic Alliance UK. The Minister gave a video address at the event. The reception focused on progress, implementing the new Welsh Plan and also raised awareness of the flagship SWAN Clinic pilot led by Dr Graham Shortland OBE.

To celebrate Rare disease day (28 February 2023) there was a global chain of lights to show solidarity for the worldwide rare community. Cathays Park, Cardiff was lit in purple from dusk until midnight.

A written statement on improving services for people living with rare diseases in Wales was published on Rare Disease Day.

Guidelines for the investigation of the child with developmental delay have also been developed and are due to be published very shortly.

The Welsh Rare Disease Implementation group (RDIG) meeting, held on 8 March, was well supported by clinicians and planners across Wales. We had presentations on the transition of RDIG into an Implementation Network in the new NHS Wales executive and were reassured that the level of support will remain the same to support the implementation of the Wales Rare Disease Action plan. Examples of innovation in Wales was also covered with discussion on the Care and respond webapp and the research nodes successful to the second stage of the MRC/NIHR grants. We now will start to look forward to the evaluation of the current implementation of the Wales Rare Disease Action plan and the refresh for the 2023/24.

POLICY UPDATES

Scotland

Scotland published the first iteration of the Action Plan for Rare Disease on 19th December 2022.

The goal is for this Action Plan to evolve over time and be reflective of the changing rare disease landscape. Our Action Plan will be reviewed regularly with actions refreshed and renewed. We will also continue to engage with the rare disease community throughout this process and beyond to hold us to account on our actions, ensuring we are ambitious in our delivery for people with a rare disease.

The Scottish Rare Disease Implementation Board will continue to oversee the implementation of our Action Plan and will soon resume, following the appointment of a new Chair and deputy Chair. The Scottish Government policy team, now bolstered by additional resource until at least the end of 2023, will continue to provide secretariat support for RDIB and have started work on supporting working groups as well as scoping work across Health and Social Care Directorates to link in to relevant workstreams.

The Scottish Strategic Network for Genomics Medicine launched in August 2022 with the Oversight Board and Steering group meeting on a number of occasions since.

Most recently, a number of working groups kicked off looking at a range of areas to be captured under the forthcoming Genomics Strategy for Scotland.

The Germline working group is being chaired by Dr Jonathan Berg, previous Rare Disease Implementation Board Chair and current clinical lead for the genomics network for rare disease.

Furthermore, arrangements are ongoing to establish a patient panel which would come under the Genomics Network. Discussions have been had with Genetic Alliance UK on how best to ensure the rare disease voice is not lost in this crucial piece of work.

Rare Disease specific publications;

- 24 March 2023 – Publication of Genomics in Scotland: Building our future. This is the first genomics publication for Scotland setting out our strategic intent for our future genomics strategic being published in the summer of 2023.
- 14 February 2023 - Congenital conditions in Scotland - This release by Public Health Scotland (PHS) focuses on congenital conditions. It provides current best estimates of the number of babies with serious congenital conditions among pregnancies ending in Scotland up to 2020.
- Congenital Conditions and Rare Diseases Registration & Information Service for Scotland (CARDRISS) – webpage and associated information/video.

Continued overleaf

POLICY UPDATES

continued

General publications for which are relevant for the Scottish rare disease community is as follows;

- 22 March 2023 – Publication of Voice of the Infant: best practice guidelines and infant pledge. The intention is to provide guidance on how to take account of infants' views and rights in all encounters they may have with professionals in statutory or third sector services, or in public spaces such as shops, libraries or galleries.

- 27 February 2023 - Transitions to adulthood for disabled young people: literature review. Scottish Ministers have committed to delivering a National Transition to Adulthood Strategy during this parliamentary term "to support disabled young people as they make the transition to adult life, and provide them and those who look after them with joined-up guidance and support to unlock better educational and employment opportunities and health outcomes.

- 22 February 2023 - Health and social care: data strategy. This first Data Strategy for Health and Social Care lays the groundwork for transforming the way we as members of the public access and use our data to improve our own health and wellbeing.

Northern Ireland

The NI Rare Diseases Action Plan sets out 14 high-level actions that have been taken forward over 2022-23. A review of Year 1 progress is currently underway, which will inform and help refine the plan for 2023/24.

The Northern Ireland Rare Diseases Implementation Group (NIRDIG) continues to meet every two months. At each meeting, the DoH Policy Team provides a summary update of discussions at the UK Rare Diseases Forum, the UK Rare Diseases Framework Board and the online forum platform for consideration and discussion. The 5 NIRDIG Working Groups which have been established to take forward the 14 actions in the Action Plan continue to meet regularly and provide updates on progress to NIRDIG.

To facilitate and drive forward the implementation of the Action Plan, recent appointments have been made to Adult and Paediatric Rare Disease Clinical Lead consultant posts. The RD leads will also have a key role on NIRDIG and will work closely with the rare disease community through the NI Rare Disease Partnership (NIRDP).

POLICY UPDATES

England

England's second Rare Diseases Action Plan was published on 28 February 2023 on Rare Disease Day, reporting on progress since the 2022 Action Plan and introducing 13 new actions for the year ahead.

We are pleased that the 2023 Action Plan has been received positively by the rare disease community, especially those actions which focus on coordination of care and health inequalities, but recognise that as always there is more to do. We are grateful to the rare disease community for their engagement and feedback to help shape the 2023 Action Plan. Going forwards, we will continue to engage with the community through the UK Rare Diseases Forum and online platform to gather feedback on implementation of actions. We will also continue to work with the England Rare Diseases Action Plan Patient Advisory Group to define remaining priority areas. The England Rare Diseases Framework Delivery Group will also continue to meet regularly Over the coming year, the group will focus on progressing the delivery of all published actions, including ongoing or extended actions from the 2022 Action Plan and new actions from the 2023 Action Plan. Interim progress reports will be provided verbally and via governance meeting papers, which are posted to the on-line forum and a detailed progress report will be given in England's 2024 Action Plan.

EVENTS

Community events

- Addison's Disease day – 29th May 2023
- Genomics England Research Summit – 19th September 2023

Governance events

- England Rare Disease Framework Delivery Group meeting: 18th May at 10am
- England Rare Disease Framework Delivery Group meeting: 13th July at 2.30pm
- UK Rare Diseases Forum: 20th July at 2.30pm
- UK Rare Diseases Framework Board: 8th November at 2.30pm