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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 ninth 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

Rare Disease Day

Rare Disease Day is observed annually to raise awareness for Rare Diseases to improve the lives of individuals and families impacted by them around the world. This year Rare Diseases Day fell on 29 February, the rarest day of the year.

Rare Disease Day in Northern Ireland

Northern Ireland (NI) celebrated Rare Disease Day on 29 February, with Parliament Buildings at Stormont Estate, the seat of government in Northern Ireland, lighting up for Rare.

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Other buildings across NI were also illuminated, such as:

- Local Council HQs;
- Various historic buildings and landmarks;
- Queen's University.

Social media was used extensively to highlight Rare Disease Day across Health and Social Care (HSC).

All-Ireland Rare Disease Day

A successful all-Ireland Rare Disease Day event was held at Farmleigh House, in Dublin. This was jointly organised by Rare Diseases Ireland (RDI) and the NI Rare Diseases Partnership (NIRDP). The agenda included a recorded message from the NI Health Minister Robin Swann and an in-person address by the Minister for Health in the Republic of Ireland, Stephen Donnelly. Topics discussed included:

- Driving change through Rare Disease Policy;
- Evolving landscape of screening/diagnostics;
- Integrated health and social care;
- Advanced therapies for Rare Diseases.

The well-attended event provided an opportunity to celebrate the uniqueness of Rare.

All-Ireland Children and Young Adults Research Advisory Group

On Rare Disease Day, the All-Ireland Children and Young Adults Research Advisory Group (CRAG) was launched by the Ombudsman for Children, Dr Niall Muldoon, on behalf of the All-Ireland rare disease interdisciplinary research network (RAiN). This groundbreaking initiative addresses the unique needs of children and young people affected by rare diseases. By actively engaging with children and young people aged 6-17 years, the initiative will ensure that their voices are heard and valued at every stage, helping to facilitate collaborative research and advocacy and drive positive change in the rare disease landscape, to ultimately improve outcomes for affected individuals and their families. Outputs from the research network will be disseminated through open-access publications, regular presentations, and the network's website in easily accessible formats.

Continued overleaf

NEWS

Further information has been placed on the discussion board of the online RD xChange platform – see links below:

[RAiN CRAG — Rare Disease All-Ireland Interdisciplinary Research Network (rainrareresearch.org)]

[News | Rare Disease Focus: supporting rare disease throughout all communities (qub.ac.uk)])

Rare Disease Day in England and the NHS

On Rare Disease Day, Minister Stephenson spoke at Genetic Alliance UK's Westminster Reception, hosted at the House of Commons. The Minister spoke about addressing the challenges faced by people living with rare diseases and wore a pair of stripy socks to take part in Medics4RareDiseases #showyourstripes campaign to raise awareness.

For the first time this year rare disease day was coordinated across the NHS. Each trust was empowered to deliver their own activities and communication on the day through a toolkit shared from the central communications team.

The story of Charlotte Proud, a mental health nurse working in the NHS living with the rare condition osteogenesis imperfecta was shared across social media channels. NHS England also shared the announcement of the new treatment, belumosudil, for the rare condition graft vs host disease. This was picked up by national news outlets, and more details are included on page 7.

Continued overleaf

NEWS

Within the NHS, there was a drive to raise awareness and educate health professionals on rare diseases. This included publishing 5 new articles about rare disease on the GeNotes Knowledge Hub providing healthcare professionals with an introduction to rare disease and familiarising them with some key concepts to be aware of when working with people living with rare conditions. The NHS England Genomics Education programme developed a series of short films 'My genomics journey: Three perspectives', to shine a light on patients with different conditions and the ways in which their lives have been affected, both before and after diagnosis. The programme also ran its two-week online course: A clinician's guide to genomic testing for rare disease, offering NHS professionals a practical guide to the testing process. Those who enrolled on 4 March were joined by a team of expert mentors who provided support during the course.

Rare Disease Day 2024: harnessing genomic medicine and cutting-edge treatments to improve outcomes for patients with rare conditions

This year for Rare Disease Day, NHS England's Professor Dame Sue Hill and Fiona Marley wrote a blog post outlining the latest developments across genomics and rare conditions within the health service.

The article showcases collaborative efforts within NHS England and with partner organisations to provide the most effective diagnosis and treatment services for the more than 3.5 million people in the UK living with rare diseases. Examples of these services include the national Whole Genome Sequencing Service, supporting clinicians to make genomically-informed diagnoses, and the Inherited White Matter Disorders Service, which is at the forefront of providing remote care and support for patients from expert teams.

The authors also highlight the clinical resources of the NHS Genomics Education Programme's Rare Disease Education Hub, which contains information on the different types of rare diseases and their diagnosis, and GeNotes, which provides guidance on making the right genomic decisions at each stage of the clinical pathway.

NEWS

Rare Disease Day in Wales

The Rare Disease Day celebration events were widely supported in Wales, with the initiation of a chain of Rare Disease gardens highlighted in ITV bulletins, online and newspapers. The support and feedback from those that attended the Rare Disease celebration event in Cardiff Bay was very positive, and they were grateful to the Rare Disease Implementation Network (RDIN) Chair, Professor Iolo Doull for presenting at this event as well as the four nations event.

Rare Disease Day in Scotland

The Scottish Government supported a number of events to promote Rare Disease Day this year. The Minister for Public Health and Women's Health, also published an open letter to the rare disease community which reaffirms our commitment to improving the lives of those living with a rare condition in Scotland.

Two days prior to Rare Disease Day, the Minister also renewed the Scottish Government's commitment to the rare disease community speaking at a reception hosted by Genetic Alliance UK/Rare Disease UK. The event heard from those living with a rare condition and also showcased pioneering research into rare diseases.

New Rare Disease Data Release – National Disease Registration Service release first information on rare disease prevalence in England

On Rare Disease Day, the National Disease Registration Service (NDRS) released information on the prevalence of certain rare diseases and conditions in England. NDRS is committed to provide information to support the planning of services and improvement of treatments in England for people with a rare disease. We have taken information from our two registration services, the National Congenital Anomaly and Rare Disease Registration Service (NCARDS) and the Nation Cancer Registration and Analysis Service (NCRAS), to report the point prevalence of selected rare diseases, congenital anomalies and rare cancers. Prevalence tells how many people living with and beyond a diagnosis at a given point in time and can help determine the number of people who may have unmet needs that can benefit from new treatments or support services. We have brought together data on 46 individual conditions that are considered rare and calculated the prevalence as of 31 December 2020. For Some of these conditions, this is the first time national prevalence figures have been reported for England.

Some of these conditions are exceptionally rare with only 1 or 2 cases per million people. It is only through collecting data at a national scale that we can bring together sufficient intelligence on these conditions to support research and develop the best care for people with rare diseases or conditions. NDRS's population based disease registration service continues to expand the number of conditions registered, including rare genetic syndromes which cause an inherited predisposition to cancer.

NEWS

Cancer patients living with chronic graft versus host disease set to benefit from a pioneering new medicine on the NHS

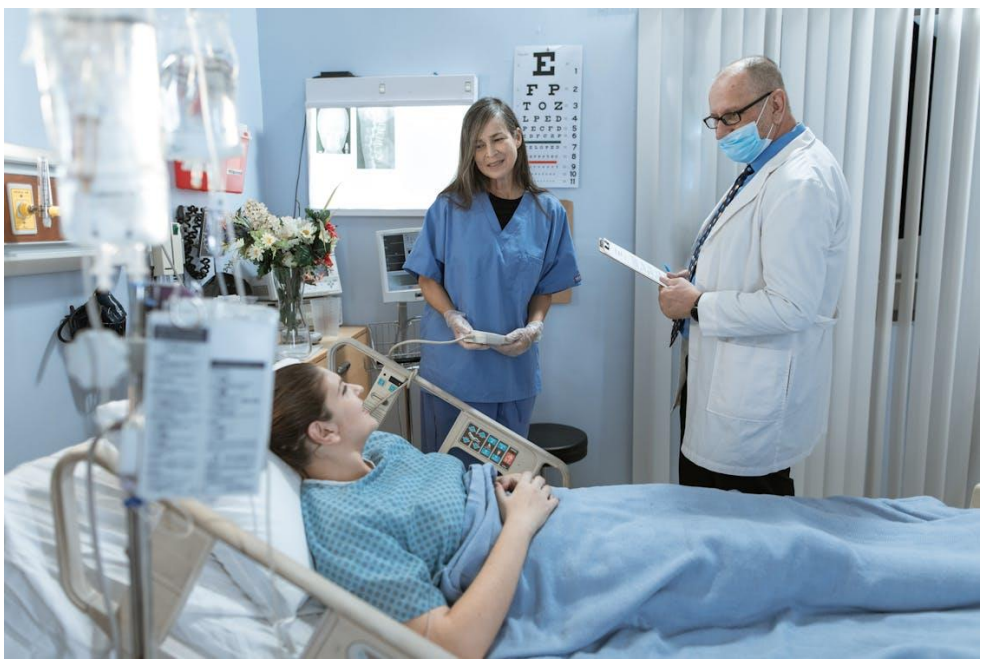
Belumosudil (Rezurock ®) is an innovative treatment for chronic graft versus host disease, which develops in around a third of patients who have undergone stem cell or bone marrow transplants for blood cancers such as leukaemia and lymphoma.

Around 200 patients living with the most severe cases of the disease and who have not had success with two previous therapies are now set to benefit from belumosudil over the next three years, after the NHS fast-tracked funding for the treatment following clinical guidance from National Institute for Health and Care Excellence (NICE). The drug is given once a day as a tablet and works by inhibiting the proteins which are responsible for the potentially life-threatening inflammatory response caused by chronic graft versus host disease. It can be taken at home, reducing the number of hospital appointments for some patients.

NHS England's Medical Director for Specialised Services, James Palmer, said: "This rollout is a real breakthrough for patients with graft versus host disease and it will help people living with this debilitating condition effectively manage their symptoms and enhance their quality of life and daily functioning."

The treatment is made available through NHS England's Innovative Medicines Fund, which fast-tracks the most promising non-cancer medicines to patients. Last September, the NHS used the fund to rapidly rollout Sebelipase alfa, a treatment for Wolman's Disease – a rare genetic and fatal condition which presents in babies and children.

Belumosudil is the latest innovative medicine that has been secured for NHS patients following recent rollout of wearable Parkinson's treatment, foslevodopa-foscarbidopa and preventative breast cancer drug, anastrozole.



NEWS

Improving patient advocacy

A paper has recently been published on how improving patient advocacy can lead to improvements in many aspects of healthcare delivery for both patients and medical practitioners. The National Ehlers-Danlos Syndromes (EDS) service, an NHS England highly specialised service, works closely with charities that support people living with EDS. This service worked with these charities to develop a 'self-advocacy survey' to better meet patient's needs through understanding how people advocate for themselves. The survey identified several key domains where patients can empower themselves to be effective advocates and healthcare providers can work with patients to ensure their needs are being met. This unique, collaborative, patient-focused model could be adapted by the rare disease community and other health care professionals to benefit those living with rare conditions.

English government endorses tyrosinaemia screening recommendation

The English government has endorsed the UK National Screening Committee (UK NSC's) recommendation to introduce newborn screening for tyrosinaemia type 1 (HT1). The rare genetic condition will be added to the conditions screen for by the NHS Newborn Blood Spot Screening Programme in England. This will potentially find an additional 3 babies a year who can be offered drug treatment and a special diet before they become symptomatic, reducing the chance of liver disease and the need for liver transplantation. See written ministerial statement from Minister of State Andrew Stephenson.

NEWS

Expert groups plan in-service evaluation of screening for spinal muscular atrophy

The UK NSC is progressing its work to review the case for newborn screening for spinal muscular atrophy (SMA).

This work has 2 main strands:

- developing a new comprehensive cost-effectiveness modelling study
- scoping an in-service evaluation (ISE) of newborn screening for SMA in real world NHS services

The SMA ISE is being overseen by a partnership board that includes screening experts from the 4 UK governments and NHS England's Vaccination and Screening Directorate, clinicians, academics, genomic experts and patient and public voice members.

SMA UK and prominent neurologists are actively involved in the design of the evaluation. They are an integral part of the team that is carefully evaluating screening for SMA in the UK.

The partnership board and clinical pathway group have both now held their first meetings. See Working in partnership to plan in-service evaluation of newborn screening for SMA.

Progress on blood spot task group workstreams

The UK NSC's blood spot task group (BSTG) was set up to identify practical and innovative approaches to help researchers and others develop evidence to help the UK NSC make robust screening recommendations for very rare conditions.

Progress on BSTG workstreams includes:

- publication of a manuscript comparing the key principles of newborn screening between the UK and European Organisation for Rare Diseases (EURORDIS)
- 2 international workshops held to help identify challenges, opportunities and practical approaches when developing modelling for rare diseases in newborn settings
- ongoing work to consider study design options for test accuracy studies
- a project focusing on how disease registries, data linkages and improved data coding could help provide evidence on rare disease outcomes

In relation to the last of these 4 workstreams, BSTG members agreed in January to commission a review of studies that explore outcomes from newborn screening, focusing on the mechanisms used in those studies to measure and monitor outcomes in the short and long term. See summary notes from January 2024 BSTG meeting.

NEWS

Edwards' syndrome consultation

The UK NSC has held a public consultation on modifying the antenatal screening pathway for Edwards' syndrome (trisomy 18).

Stakeholders were asked for their feedback on modelled data evidence that suggests the quadruple screening test, which uses maternal age and 4 biochemical markers, should be added to the Edwards' syndrome screening pathway.

The UK NSC will consider the evidence and consultation responses before making a formal recommendation on this proposed programme modification.

Evaluation of newborn screening for SCID

The practical evaluation of newborn screening for severe combined immunodeficiency (SCID) in English NHS services was launched in September 2021 by the Department of Health and Social Care (DHSC) and NHS England (NHSE).

The formal evaluation period is now ending. Data from the evaluation will help answer questions about the effectiveness of NHS screening for SCID in England compared to the way babies are detected without screening. Information is also being gathered on the impact of screening on the public and on healthcare professionals.

Following the completion of the evaluation, a report will be submitted to the UK NSC. The committee will review the report, take the advice of its newborn screening experts, and consult for 3 months before making a recommendation to ministers on whether SCID should become part of the NHS Newborn Blood Spot Screening Programme in the UK.

Potential benefits and harms of whole genome sequencing in newborn screening for rare diseases

The UK NSC continues to work closely with Genomics England to look very carefully at the implications of whole genome sequencing (WGS) for screening in the UK. These implications include very important questions as to which conditions it would be ethical and acceptable to test for, and how genetics could usefully add to existing newborn screening programmes.

Continued overleaf

The NHS-embedded Generation Study aims to use WGS to test at least 100,000 newborn babies for several hundred potentially treatable rare diseases that are likely to present by the age of 5 years.

This has huge potential for the future of screening in the UK. However, before making any recommendation to use WGS in screening, the UK NSC will need to consider all possible benefits, harms, practical and ethical challenges.

Challenges include assumptions that newborn genome sequencing will automatically lead to better outcomes, inconclusive results that could lead to families living in limbo after screening, and the potential for WGS to adversely affect uptake of existing newborn screening programmes if parents wrongly think it is a better test and that established screening is no longer needed.

The UK NSC therefore commissioned, via the NIHR Evidence Synthesis Programme, a review of the benefits and harms of WGS in newborn babies. This work, due to complete in summer 2024, will help make sure any future UK NSC recommendations on the use of WGS in newborn screening are clearly led by the evidence.

NHS Research Secure Data Environment

The NHS Research Secure Data Environment (SDE) Network is supporting the NHS to deliver the world's largest linked health datasets, which will improve care, support innovation and sustain the NHS.

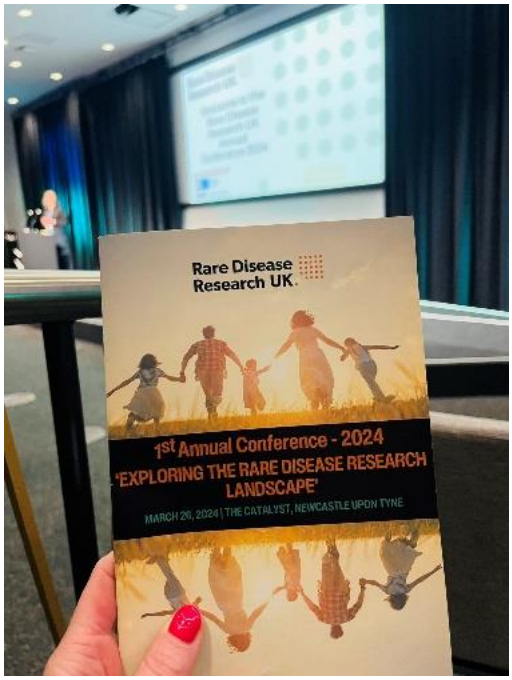
The Network comprises the NHS England SDE, which provides national scale coverage of highly curated, high value datasets including cancer and rare diseases; while the Sub National SDEs (Secure Data Environments) collectively cover all of England and provide access to granular, near real-time, multimodal data bolstered by clinical and data science expertise that support data curation and the translation of research into practice. There is a single front door to the SDE Network, provided by HDRUK, to streamline access for researchers.

The NHS is supporting innovative research of all types; from clinical trials, post market surveillance, epidemiology, translational research, and AI (Artificial Intelligence) development and validation. There are more than 61 active research studies in the SDE Network, tackling critical areas such as rare diseases and AI (Artificial Intelligence) development. A recent example of rare disease research is underway in the Thames Valley and Surrey Secure Data Environment where researchers are working with Cancer Research UK to research rare types of blood cancer.

NEWS

Rare Disease Research UK

Rare Disease Research UK (RDR UK) is a new £14million platform to advance research into rare diseases. Jointly funded by Medical Research Council (MRC) and NIHR, RDR UK is made up of 11 research nodes - involved in both disease-area-specific and cross-cutting research, engaging leading universities across the country- and a coordinating hub, hosted by Newcastle University, Newcastle upon Tyne Hospitals NHS Foundation Trust, and Genetic Alliance UK. The aim is to significantly impact the rare disease research landscape and improve the lives of those directly or indirectly affected by rare diseases.



The First Annual Rare Disease Research UK Conference took place on 26 March in Newcastle, bringing together around 120 individuals with an interest in rare disease research, including researchers, healthcare professionals, patients, and industry representatives, either in person or online. Keynote speakers included Prof Lucy Chappell - Chief Scientific Adviser to the Department of Health and Social Care (DHSC) and CEO of the NIHR- and Dr Kath Bainbridge, Head of Rare Diseases and Emerging Therapies at DHSC. Key topics covered included the UK Rare Diseases Framework, current Rare Disease Research landscape in the UK, RDR UK's research objectives, and reflections on the new landscape and the opportunities it presents.

The day also featured engaging panel discussions on Patient and Public Involvement and Engagement (PPIE) and industry collaboration for better trials, emphasizing the necessity of collaboration, investment, and patient involvement in addressing the complex challenges of rare disease research and treatment development.

Moving forward, Rare Disease Research UK plans to hold topic-specific workshops and a conference each year, each hosted in various locations across the UK. Each conference will be an opportunity to showcase the impact of RDR UK and the progress made in the nodes' research initiatives. These gatherings will bring together people from all parts of rare disease community to work together, share ideas, create awareness and find new ways to accelerate rare disease research and treatment.

Find out their news, research, activities and more on https://rd-research.org.uk. For networking and collaboration, follow them on LinkedIn (Rare Disease Research UK) and X (@RDRUKHub).

NEWS

LifeArc Translational Centres for Rare Diseases

On 23 April, LifeArc announced the news that £40m research centres which will unlock new tests, treatment and cures for people living with rare diseases.

The four new LifeArc Translational Centres which are spread across the four nations, will tackle barriers that prevent new tests and treatments reaching patients with rare diseases and speed up the delivery of rare disease treatment trials.

The four centres are:

- The £9.4m LifeArc Centre for Rare Respiratory Diseases at the University of Edinburgh. The centre is a partnership between universities and NHS Trusts across the UK.
- The £10.4m LifeArc Kidney Research UK Centre for Rare Kidney Diseases at the University of Liverpool.
- The £7.5m LifeArc Centre for Rare Mitochondrial Diseases at the University of Cambridge.
- The £12m LifeArc Centre for Acceleration of rare disease trials. This brings together a consortium of three universities from across the UK, Queen's University Belfast, Newcastle University and University of Birmingham, who are pooling their expertise in a partnership.

Patient and Carer survey

A Patient and Carer survey, developed by the Public Health Agency in partnership with Queen's University Belfast (QUB) and Ulster University (UU), has been issued to collect information on attitudes to, and experiences of, rare disease research. It is part of an action to improve opportunities and experiences of people with rare disease in Northern Ireland who have participated, or who wish to participate, in research studies. It will collect anonymised information that will be used to produce a report for the Northern Ireland Rare Diseases Implementation Group (NIRDIG) and a subsequent publicly available report.

NEWS

Genomics Training Academy

The Genomics Training Academy (GTAC) is a national initiative led by NHS England's Genomics Education Programme (GEP) and Genomics Unit, set to launch in spring/summer of 2024. The academy will provide training and education to the specialist genomics laboratory and clinical workforce in the UK via online courses, webinars, workshops, virtual reality and hybrid training, in partnership with the seven Genomic Laboratory Hubs.

Curious about what the GTAC team has been up to so far? Read this news release for the latest details, including the GTAC's teaching faculty and which courses are on the horizon. More information will be released in the coming weeks, such as launch dates for specific resources and how to enrol as a learner.

If you have a general query about the GTAC, please england.gtac@nhs.net and be sure to follow the GEP on X (Twitter), Facebook and LinkedIn.

POLICY UPDATES

Scotland

The first meeting for 2024 of Scotland's Rare Disease Implementation Board (RDIB) took place on 20 March. Members were updated on the progress of our Once-For-Scotland research sub-group; CARDRISS's completion of their first years' worth of registrations and comparisons made to linked datasets, and Genetic Alliance UK's 'Stats Behind the Stories' report. Members were asked to submit comment on a first draft of our Progress Report. Feedback returned on this draft was implemented and members are currently being asked for any further comment on the second draft ahead of wider sharing.

Genetic Alliance UK hosted patient involvement events in the last week of March 2024 to gauge their current views on Scotland's Action Plan and what they see as priorities for the coming year. Discussion from these meetings will inform the development of their progress report and priorities for the coming year.

In January, Scotland launched their first rare conditions page on the NHS Inform website. Rare conditions | NHS inform The webpage contains information to help those living with a rare condition and signposts to organisations such as Genetic Alliance UK and the Office of Rare Conditions for further support.

The Minister for Public Health gave a keynote speech at a Scottish Parliament reception on 30 April to mark Undiagnosed Children's Day in Scotland. Attendees heard from those living with a rare condition to give insight into the challenges young people face in their diagnostic odyssey and in the transition to adult care.

Northern Ireland

The Northern Ireland Executive returned on 3 February 2024, with Minister Robin Swann once again taking up the post of Northern Ireland's Health Minister. Minister Swann is a great supporter of rare diseases and was previously responsible for publishing Northern Ireland's Rare Diseases Action Plan in 2022.

Continued overleaf

POLICY UPDATES

The Northern Ireland Rare Diseases Implementation Group (NIRDIG) continues to meet quarterly, with the working groups taking forward actions. The meeting was held on 28 March.

Work on developing individual service specifications and care pathways in the Health and Social Care (HSC) in Northern Ireland is ongoing across the following adult services: Inherited Cardiac Conditions (ICC), Inherited Metabolic Disorders (IMD), Immunology, Haematology, Red Cell Disorders (RCD), and Cystic Fibrosis (CF).

England

On Rare Disease Day (29 February) we published England's 2024 Rare Diseases Action Plan which included 7 new actions as well as progress on previously defined actions, further information on this can be found here. This year's action plan included progress on designing and funding two pilots for Syndromes Without A Name (SWAN) clinics, networked models of care to ensure patients can be treated as close to home as possible, progress on the Generation study which will pilot whole genome sequencing (WGS) to screen for genetic conditions in 100,000 healthy newborns and the news that NICE have updated their quality standard on transition from children's to adults' services to ensure it is relevant to the needs of the rare disease community.

Following publication we met with Patient Advisory Group, run by Genetic Alliance UK to explain how their input helped shape the action plan and discuss how we will continue to work with them in development of next year's action plan. The England Rare Diseases Framework Delivery Group continues to meet regularly, in our March meeting we discussed how we will work over the coming year to effectively drive actions to improve the lives of people living with rare conditions.

Wales

The Wales Rare Disease Action Plan was updated and published in January and issued to the NHS in Wales under a Welsh Health Circular.

The Cabinet Secretary of Health and Social Care issued a written statement to the Welsh Parliament to mark Rare Disease Day, alongside a number of events and some related publicity.

The Wales Rare Disease Implementation Group has transitioned to an Implementation Network within the NHS Wales Executive and the group is currently reviewing which priorities it will take forward this year.

EVENTS

Governance Events

England Rare Disease Framework Delivery Group: 16 May and 2 July

Northern Ireland Rare Disease Implementation Group (NIRDIG): 27 June, 19 September and 19 December

UK Rare Diseases Framework Board: 6 November

Scotland's Rare Disease Implementation Board: 18 June

UK Rare Diseases Forum: 17 July and 23 October