September 2023 | Vol. 7

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 seventh 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

Ministerial Portfolio

There has been a change made to the UK government ministerial portfolios, so Minister Quince MP, the Minister of State for Health and Secondary Care, now holds responsibility for rare diseases.

NHS Wales Executive

The NHS Wales Executive has now been launched (1st April) and will support NHS organisations in Wales to plan and deliver the expectations set out in the quality statements/Rare Disease action plan to improve the delivery of equitable and access quality care for all. It is proposed that the Rare Diseases Implementation Group will transition into an implementation network, with enhanced workforce resources. This will continue to work closely with Genomic Partnership Wales, to ensure the voices of those impacted with a Rare Disease are considered across Wales.

UK Rare Disease Research Platform

In July 2023 the Medical Research Council (MRC) and the National Institute for Health and Care Research (NIHR) announced a new £14 million joint investment to target rare diseases. The UK Rare Disease Research Platform aspires to have a major impact on the Rare Disease Research landscape and improve the lives of those directly or indirectly impacted by rare disease conditions over the next 5 years.

The platform is made up of 11 UK-wide research nodes and a coordinating hub. The nodes based at universities across the UK, will be responsible for specific research projects. The hub, which is led by Newcastle University in partnership with The Newcastle upon Tyne Hospitals NHS Foundation Trust and Genetic Alliance UK, is responsible for coordination of the platform, supporting the nodes with their research, providing dedicated project management, engagement, and communications for the platform as a whole and designing and delivering training and a range of tailored events to bring together the wider rare diseases community.

Continued overleaf

NEWS

The inaugural Platform Executive Meeting has already taken place and work is already underway to establish infrastructure and principles of working to ensure strength of UK research into rare disease is amplified for increased international impact.

If you would like further information, please see the UKRI press release or email hub@rd-research.org.uk

LifeArc Translational Rare Disease Centres - £40M investment into the UK rare disease ecosystem

LifeArc is a self-funded, charitable medical research organisation specialising in translating early-stage scientific discoveries into the next generation of diagnostics, treatments, and cures. Their strategy commits to investing £1.3 billion into areas of unmet medical need by 2030.

Continued overleaf

NEWS

As part of their recently launched Rare Disease Translational Challenge, LifeArc has committed £40m to create around four Translational Rare Disease Centres, to tackle the major challenges in rare disease research many of which are outlined in the UK rare disease framework. LifeArc are working with UK academic institutions and the rare disease community to establish these centres of excellence, strengthening the UK's rare disease translational capability.

The Centres' main aim is to foster the collaborative, co-ordinated and multidisciplinary approach required to develop new diagnostics and therapeutics for rare diseases. The Centres will engage and involve the patient community, industry, and policy makers to overcome shared challenges and support skills development, including equipping early career researchers to establish their careers in translational rare disease research.

A separate call for a Co-ordinating hub, will provide support for administrative and cross cutting activities, including knowledge-sharing and engagement with the UK patient community.

For more information, visit Rare Disease – LifeArc

Funding bid for an all Wales virtual health hub

A funding bid has been developed and submitted to the Life Arc Rare Disease Translational Challenge call as a digital All Wales Rare Disease Centre. This was heavily supported by Medics 4 Rare Diseases and Wales are very grateful for their time and enthusiasm. This has been accepted through the first phase and following an application in partnership with the University of Oxford, the full application is to be submitted by 29th September and funding outcomes will be announced by 31st December 2023.

Digital Health Care Wales

Applications to Digital Health Care Wales have been submitted via the Bevan Commission Project for highlighting patients in secondary care with Rare Diseases and linking with Orphanet database information, similar to end of life planning flags.

NEWS

SWAN clinic

SWAN (Syndrome Without A Name) Clinic funding has been secured until March 2024. Ongoing discussions continue between Welsh Government and WHSSC (Welsh Health Specialised Services Committee) for long term funding are based on patient reported outcomes measures, specifically designed for the clinic by a CEDAR (Centre for Healthcare Evaluation, Device Assessment and Research) and RDIG (Rare Disease Implementation Group) collaboration. Members of the Genetic Alliance Cymru attended the Royal College of Paediatrics and Child Health annual Welsh conference to highlight the opportunities to refer into the SWAN clinic.

Code on Genetic Testing and Insurance

The UK government and the Association of British Insurers (ABI) launched a Call for Evidence on the Code on Genetic Testing and Insurance on 25th July 2023.

The Code is a voluntary agreement between the UK government and the ABI and provides guidance on the role of genetic testing in insurance across the UK. Following the 2022 review, we committed to holding a Call for Evidence in 2023 to assess whether the Code needed to be refreshed to better reflect the changing genomic landscape.

The Call for Evidence will be seeking views from individuals and organisations, including those with genetic conditions, family members and carers, healthcare professionals, the insurance industry, and the public, to ensure the Code remains fit for purpose.

It will look at two issues which may require the Code to be updated, these are:

- Whether the definitions for genetic tests used in the Code remain up to date.
- The need to outline a transparent and accessible approach to assessing which predictive genetic test results may need to be disclosed under the Code in the future.

The Call for Evidence will close on the 17th October. Results from the call for evidence will be reviewed and used to help determine whether the Code requires updating.

In addition to the Call for Evidence, the ABI have recently published an updated consumer guide of the Code following input from Genetic Alliance UK and the Huntington's Disease Association which we hope will empower people to understand the Code and respond to the Call for Evidence.

NEWS

NICE quality standard on transition from children's to adults' services

NICE (National Institute for Health and Care Excellence) quality standards set out priority areas for quality improvement in health and are developed independently in collaboration with health professionals and service users. In England's 2023 Rare Diseases Action Plan, DHSC committed to partnering with NICE to host a workshop to better understand how the NICE quality standard on transition could be adapted to ensure it is relevant to the needs of the rare diseases community. This workshop took place virtually on 29th June with attendees including those with lived experience of transition from paediatric to adult rare disease services as well as clinicians working in this area. The workshop took each of the quality statements in turn to consider whether these reflect the needs of those living with rare diseases and to discuss how they might be adapted. The workshop also covered areas of need not covered by the existing quality statements. The quality standard on transition will now be updated, taking into consideration the input gained at the workshop.

National Screening Committee

The UK National Screening Committee (NSC) published a blog article on 4th August, providing an update on the progress of its review of the evidence for newborn screening for spinal muscular atrophy.

UK NSC have also explored how newborn blood spot screening policy and practice in the UK compares with the patient-led European Organisation for Rare Diseases (EURORDIS) principles in a paper published in the Lancet Regional Health – Europe.

The paper is called Policy-making and implementation for newborn bloodspot screening in Europe: a comparison between EURORDIS principles and UK practice.

The UK NSC has also published a blog article summarising the paper.

NEWS

New NHS treatments helping extend survival for babies with rare muscle-wasting disease

On 7th August, NHS England shared the news that nearly three in four babies with spinal muscular atrophy (SMA) are now surviving for two years or more, thanks to advances in NHS treatment.

Historical studies of the rare condition have estimated that before the availability of treatments, fewer than one in ten (8%) children born with SMA type 1 – the most common form of the condition – survived to the age of 20 months without permanent ventilatory support.

But thanks to advances in treatment, 73% of children with SMA1 in the UK are now older than two years, according to new data from the national SMA Research and Clinical Hub (SMA REACH UK) database.

This follows the roll-out of three "transformative" new SMA medicines on the NHS from 2019 (nusinersen, Zolgensma® and risdiplam), ensuring access to treatment for patients with types 1, 2 and 3 SMA.

The news item included the stories of two children with SMA1 and SMA2, who have benefited from the new treatments, and was covered by ITV evening news, as well as in UK national media online and in print.

Read more:

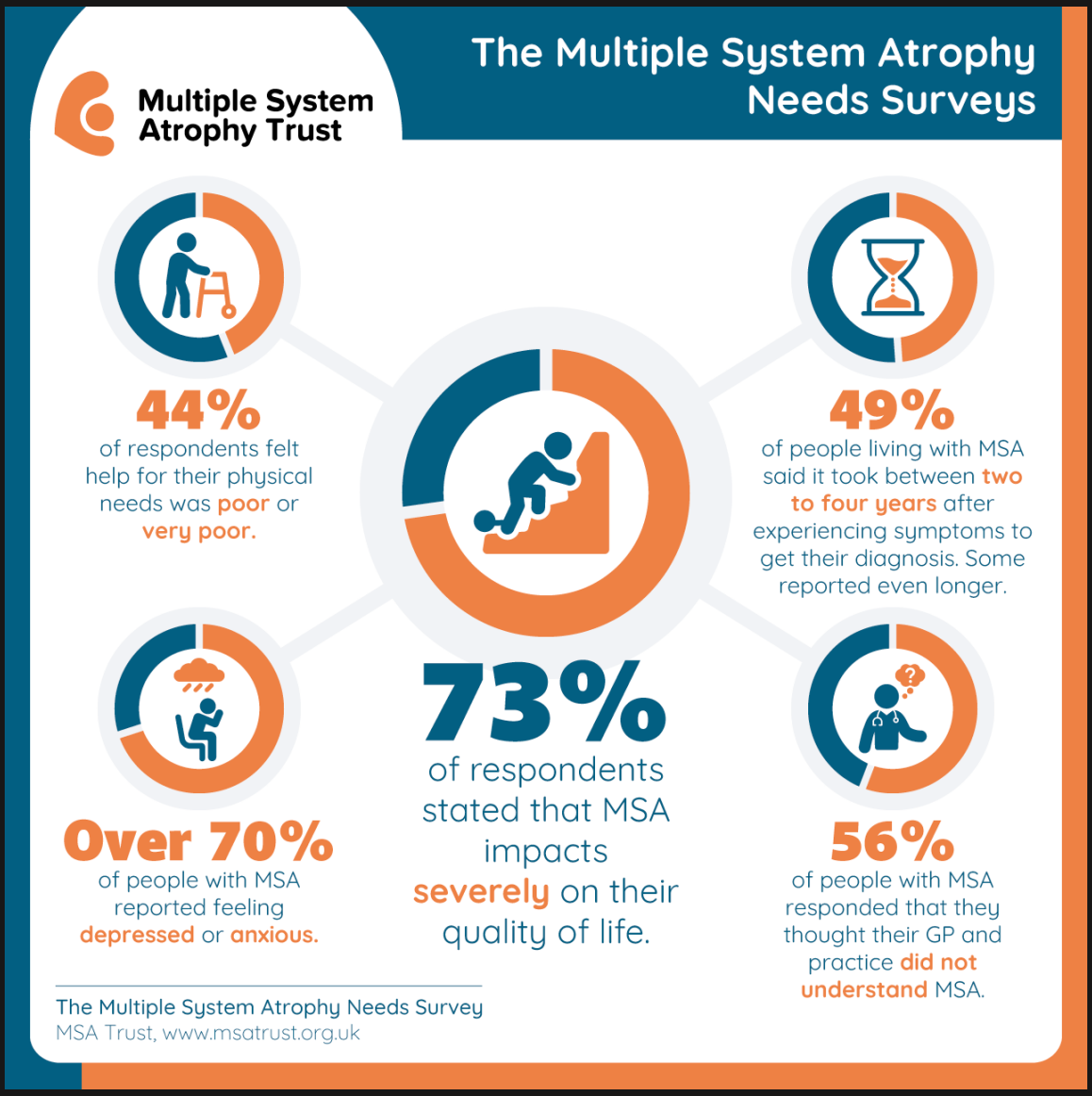
- Telegraph
- Itv news
- Express

NEWS

Multiple System Atrophy

The Multiple System Atrophy (MSA) Trust have recently completed their second survey of people affected by the condition. MSA is a rare, progressive neurological condition affecting adults, leading to premature death and has no cure. Completed by 520 people the findings indicate:

- Diagnosis is slow with 49% saying it took between 2-4 years with others reporting even longer
- ¾ of respondents said they had nobody to co-ordinate their care locally, despite their complex needs
- Awareness of MSA amongst GP's and other Healthcare Professionals was low
- The majority have never been offered any access or opportunities to be involved in clinical trials or other research opportunities.



Continued overleaf

NEWS

With MSA having significant impact on physical and mental health the report illustrates that much more could be done to improve support. Full survey results and a summary can be found here https://www.msatrust.org.uk/cause-and-cure/research-news-2/msa-needs-surveys-22/.

The MSA Trust would like to work with any other organisations who support people affected by MSA and are holding an MSA Study Day (both in person and virtual attendance) on 19th October details are here - https://www.msatrust.org.uk/event/2023-msa-trust-hybrid-study-day-for-health-and-care-professionals/

RISE study

The RISE study, developed by Medics 4 Rare Diseases to understand the knowledge base of undergraduate medical students, is nearing completion, following support from Cardiff and Vale UHB (University Health Board). This reached over 30 medical schools in UK, over 180 respondents, with work ongoing to modify the questionnaire to allied healthcare professionals and nurses.

GeNotes Paediatrics now live with 100+ resources!

GeNotes – the exciting new resource from the National Genomics Education Programme to support clinicians in accessing genomic testing – has welcomed another new specialty to its growing collection.

GeNotes Paediatrics has been developed in collaboration with clinical experts across the NHS and covers when, why and how to request genomic testing. Underpinning these In the Clinic scenarios is the Knowledge Hub, packed with bitesize resources on the fundamentals of genomics, conditions, therapies, technologies and more – perfect for busy clinicians who want to continue their learning. The collection launches with 48 In the Clinic scenarios and 57 Knowledge Hub resources. Be sure to check out GeNotes today!

NEWS

Care in the Digital Age

The Scottish Government's 2023-24 Delivery Plan for Care in the Digital Age was published on 4th August, with implications for improved co-ordination of care and access to services across health and social care.

Master's in Genomic Medicine: 2023/24 funding opens

The National Genomics Education Programme is pleased to announce that funding applications are now open for those wishing to further develop their knowledge of genomics through the popular Master's in Genomic Medicine framework.

Seven partner universities are delivering a continuing, co-ordinated programme of genomics education. NHS employees in England can now apply for funding to undertake up to four individual Continuous Personal and Professional Development (CPPD) modules that start before 31st March 2024.

Limited funding is also available for those wishing to extend a previously awarded qualification under the framework. This will be subject to meeting set criteria, as well as funding availability. Find out more on the Master's webpage.

POLICY UPDATES

Wales

The Welsh Rare Disease Implementation group (RDIG) meeting, held on 11th July, was well supported by clinicians and planners across Wales. Examples of innovation in Wales was also covered with discussion on the MRC grant application.

Kevin Francis has been appointed as the new Welsh Government policy lead for Rare Diseases in Wales, Kevin commenced his role on 3rd July 2023.

Reporting the progress of the Wales Rare Disease action plan is underway. Engagement activities are taking place with key stakeholders ready for the refresh of the plan at the UK Rare Disease forum meeting in October 2023.

Scotland

Scotland's Rare Disease Implementation Board (RDIB) resumed on 26th June following a period without a Chair. Dr Martina Rodie from the Office for Rare Conditions in Glasgow, and Dr Ruth McGowan have now been confirmed as Chair and Deputy Chair respectively. They have also been reviewing the membership and will continue to do so.

The RDIB meeting of 26 June gave members an overview of progress to date, up to the publication of Scotland's first Action Plan for Rare Disease in December 2022. Furthermore, members heard from guest speakers from across Health and Social Care on workstreams that align well with our objectives set out in the Action Plan. The next RDIB meeting is scheduled for 19th September 2023.

Collaborative work is ongoing between the Office for Rare Conditions and Genetic Alliance UK to understand the needs of healthcare professionals with regard to information about rare diseases. This will help to identify the best resources to use and signpost to increase awareness of rare diseases.

Scottish Government colleagues developing Getting It Right For Everyone (GIRFE) work have reported the first round of feedback from the 'pathfinders' working in each NHS Board. The initial feedback highlights the need for greater care co-ordination across health and social care and has many other interesting pointers (such as equalities) that the Rare Diseases team will continue to engage with as common interests.

POLICY UPDATES

Northern Ireland

The Northern Ireland Rare Diseases Implementation Group (NIRDIG) now meets on a quarterly basis to align with meetings of the UK RD Forum. At the most recent meeting, members of NIRDIG approved the Year 1 Progress Report and new actions for the Northern Ireland RD Action Plan for 2023/24.

The majority of the actions in the NI RD Action Plan are ongoing and have been taken forward into 2023/24. Two new actions were identified by the Working Groups during Year 1 – these are:

Action 15 – to establish a Rare Diseases Patient Advisory Group to provide input on all aspects of the NI RD Action Plan.

Action 16 – to improve understanding of special needs for children affected by rare diseases among educational professionals and to better link parents and these organisations.

Progress has been made in Year 1 across all areas of the Action Plan and a link to the Year 1 Progress report will be shared when it is publicly available.

England

Policy officials have continued to work with Delivery Partners to progress the delivery of actions in England's 2023 Rare Diseases Action Plan. Good progress is being made against published actions. In particular, significant progress has been made with the Rare Diseases Research Landscape project, and a report of this project is set to be published in autumn 2023.

The England Rare Diseases Framework Delivery Group continues to meet regularly. In the May meeting, the chair of the UK Cancer Genetics Group attended to lead a discussion about the inclusion of rare genetic conditions which cause an inherited predisposition to cancer within the scope of England's rare diseases action plans. Members noted that this is an important topic and are currently preparing written responses detailing work that is already being done in this area and opportunities for where further work could be done.

The deep dive discussion at the May meeting was led by the Workforce, Training and Education directorate in NHSE (formally Health Education England), who gave a detailed update on the progress made to deliver the actions they own. It was noted that good progress was being made, but delivery was against a background of organisational transition and associated challenges. A contract with Medics 4 Rare Diseases had recently finished, but work was underway to re-commission this activity. This partnership had enabled significant amounts of progress to be made.

Continued overleaf

POLICY UPDATES

continued

A deep dive update about the progress to deliver the virtual toolkit action, owned by the NHS England Highly Specialised Commissioning Team, was also given, where it was noted that the toolkit had been distributed to all acute NHS trusts in England and had received positive feedback. This had gone beyond the metric stated in the action plan (which was distribution to all highly specialised services clinical leads).

In the July meeting, the Highly Specialised Commissioning Team in NHS England led a deep dive discussion on progress against the remaining actions they own. Key themes of work for the team were highlighted, which included: access to drugs; approaches to advanced therapies; genomics; undiagnosed conditions. This led to a discussion about how individualised medicines were likely to become more commonplace and how appropriate access routes for these medicines were needed to be established.

The next delivery group meeting will be held on 13th September and will include deep dive progress updates on the actions owned by the DHSC screening team and by Genomics England.

Policy officials are currently in the process of organising three stakeholder engagement workshops. One workshop will be on mental health in children and young people with rare genetic conditions. Another will be on the topic of non-genetic rare conditions, and this will be a UK-wide workshop. And finally, one will be a follow-on from the workshop held in December 2022 with the National Congenital Anomaly and Rare Diseases Registration Service (NCARDRS) and will aim to start to identify solutions to challenges raised in the previous workshop, that build upon existing services.

EVENTS

Community events

- The George Pantziarka TP53 Trust (Li Fraumeni Syndrome). Annual conference – 9th September.
- Backbench Debate on PANS and PANDAS 12th September
- International Myotonic Dystrophy awareness day - 15th September
- Aortic Dissection awareness day – 19th September
- Genomics England Research Summit – 19th September
- Hybrid public Genomics Café – Cardiff - 21st September
- Neurosarcoidosis patient day – 23rd September
- Publishing Genomics papers of the future – 26th September
- Wolfram Syndrome UK annual conference - 30th September
- Genomics England speaking at International Consortium on Newborn Screening conference – 5th & 6th October
- Rare summit day 2023 – 12th October
- Huntington's disease community conference – 4 & 5th November
- Action Duchenne annual international conference – 10 & 11th November
- Metabolic support UK annual conference – 18th November
- London Rare Disease Showcase – 27th November

Governance events

- England Rare Diseases Framework Delivery Group: 13th September
- UK Rare Diseases Forum: 25th October
- England Rare Disease Framework Delivery Group: 31st October
- Genetic Alliance Annual meeting, Wales: 2nd November
- UK Rare Diseases Framework Board: 8th November