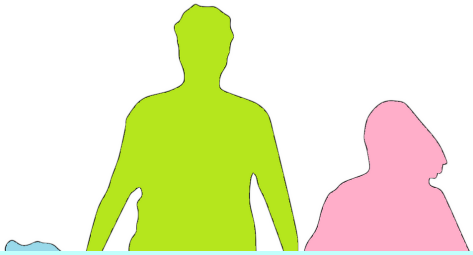
September 2021 | Vol. 1

QUARTERLY 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 first edition of the UK Rare Diseases Forum Newsletter. This is a chance to provide you – the rare disease community – with the most current 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. We will continue to post policy updates to the Forum, and we are very keen to seek feedback from the community, so do get involved and have your say – this is a key chance to feed into the implementation of the UK Rare Diseases Framework. We have already - and will continue to - feed your comments back at the strategic UK Rare Diseases Framework Board meetings and each nation will also use your input to help steer development of the nation specific action plans.

This newsletter covers all four nations of the UK and is published quarterly 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 (1)

The Medicines Repurposing Programme

The Medicines Repurposing Programme launched in March. This multi-agency initiative identifies and develops opportunities to repurpose medicines (that is, medicines used in ways not included in the original licence) in order to improve clinical outcomes, patient experience and value for money. The programme is currently considering how it might be used for the benefit of rare disease medicines. For more information please see the website or email england.repurposing@nhs.net.

Whole Genome Sequencing

The NHS Genomics Medicine Service (GMS) began the roll-out of whole genome sequencing to the NHS in England in November 2020. The roll-out is in the live clinical testing phase, which enables the service to be offered to a small number of rare disease and cancer patients and test the end-to-end pathway to support volumes being ramped-up. Whole Genome Sequencing (WGS) is currently available for seriously ill children likely to have a rare genetic disorder, people with one of 21 rare conditions where current evidence supports early adoption of WGS as a diagnostic test, and people with specific types of cancer for which there is likely to be the greatest patient benefit from using WGS (including children with cancer, sarcoma and acute myeloid leukaemia). NHS England and NHS Improvement will continue to expand the clinical indications delivered via WGS as the service continues to develop.

Monogenic Diabetes Programme

The NHS GMS is working in collaboration with the NHS England and NHS Improvement Diabetes Programme to roll out a new programme to support trusts to improve identification of monogenic diabetes. The initiative will ensure training and support for a designated nursing and medical lead in each Trust, and support provided by 15 specialist genetic diabetes nurses. For more information please see: www.england.nhs.uk/2021/08/nhs-to-diagnose-thousands-of-people-with-rare-diabetes/

NEWS (2)

Whole genome sequencing in newborn screening

Findings from the public dialogue on the use of whole genome sequencing in newborn screening were published on 8 July 2021: The report found members of the public were broadly supportive of the use of the whole genome sequencing (WGS) in newborn screening – providing that the right safeguards and resources are in place.

Innovative Medicines Fund

NHS patients are set to benefit from early access to potentially life-saving new medicines, including cutting-edge gene therapies, thanks to a new Innovative Medicines Fund and £680 million of ringfenced funding.

NICE Methods Review

The final consultation on the National Institute for Health and Care Excellence (NICE) methods, processes and topic selection for health technology evaluation is underway. Take part here.

Kuvan draft guidance

NICE has issued final draft guidance which now recommends sapropterin (also called Kuvan and made by BioMarin) as an option for treating phenylketonuria (PKU) in pregnant women until they give birth as well as for treating the condition in people until they turn 22.

National Congenital Anomaly and Rare Disease Registration Service

The National Disease Registration Service (NDRS), including NCARDRS, will be moving to NHS Digital from 1st October 2021. Other Public Health England functions will be moving to NHS England, the Department of Health & Social Care or the new UK Health Security Agency. NCARDRS is working to keep disruptions to output to a minimum and are looking forward to the opportunities that working within NHSD will bring.

"...the new Innovative Medicines Fund will mean all patients – not just those with cancer – will benefit from early access to the most promising and innovative treatments, backed by £680 million of ringfenced funds."

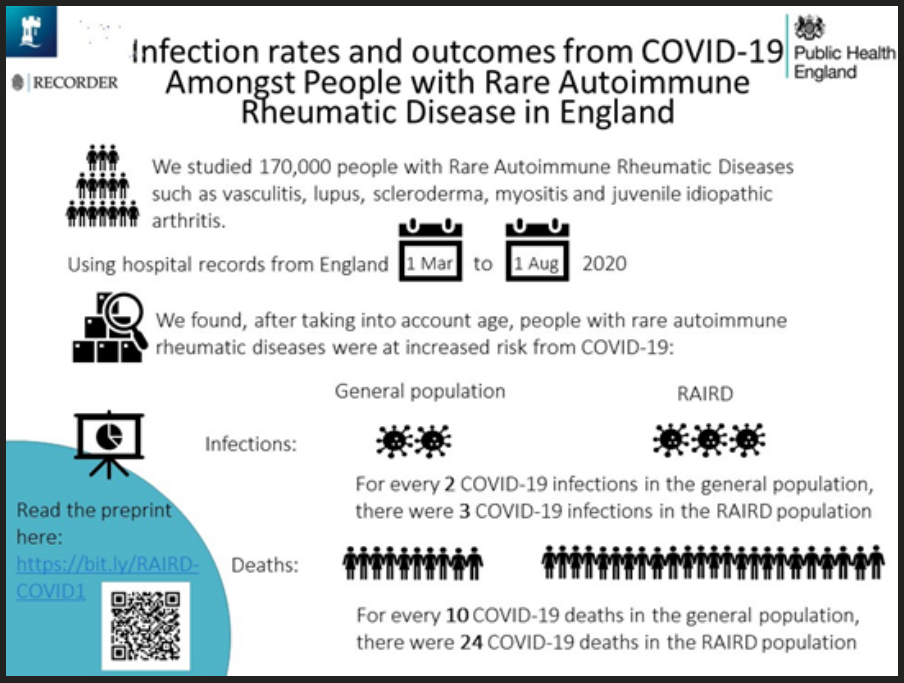
NHS Chief Executive, Simon Stevens

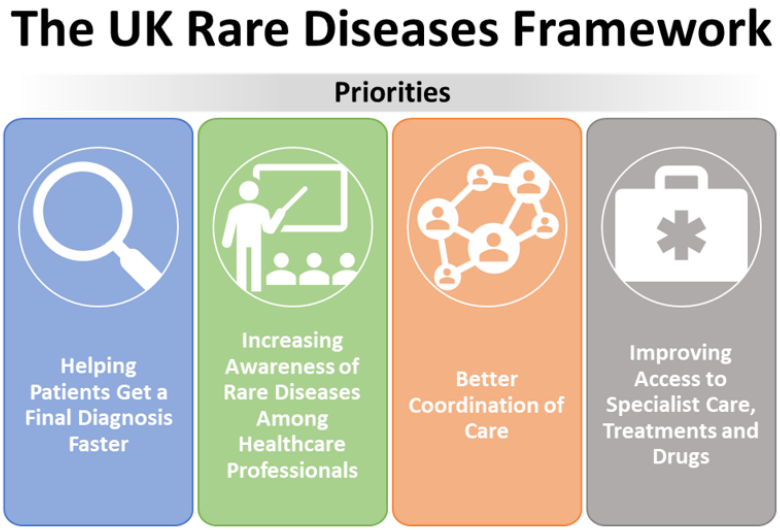
NEWS (3)

COVID-19 Infection, Admission and Death Amongst People with Rare Autoimmune Rheumatic Disease in England

The RECORDER Project, a collaboration between the National Congenital Anomaly and Rare Disease Registration Service (NCARDRS) and the University of Nottingham using electronic records for the whole of England, has found that during the first wave of the pandemic, people with rare autoimmune rheumatic diseases were 54% more likely to test positive for Covid-19 infection and 2.4 times more likely to die from it than people in the general population, when age and sex are taken into account. The research, published as a pre-print in medRxiv and currently under peer review, is the largest study of Covid-19 outcomes in people with rare diseases and highlights the unique capability of NCARDRS as a resource to support research to improve care, outcomes and inform health care policy for rare diseases.

RECORDER is developing important insights that help health services to better understand rare diseases and anticipate demand for potentially life-saving treatment,





POLICY UPDATES (1)

England

The England Rare Diseases Framework Delivery Group is the group responsible for developing and subsequently monitoring the action plan for England. This group has had two formal meetings to date (8th July and the 13th August), and the papers from the first meeting can be found in this folder within the 'Governance Structure Papers' folder on the UK Rare Diseases Forum. The most recent meeting in August focussed the discussion on priorities 2 and 4 ('increasing awareness of rare diseases among healthcare professionals' and 'improving access to specialist care, treatments and drugs') of the UK Rare Diseases Framework, examining the actions that had been put forward to meet these priorities. The next meeting of the Delivery Group will be on the 22nd of September, and this will focus on priorities 1 and 3 of the Framework ('helping patients get a final diagnosis faster' and 'better coordination of care'). Minutes of these meetings will be posted to the Exchange once approved.

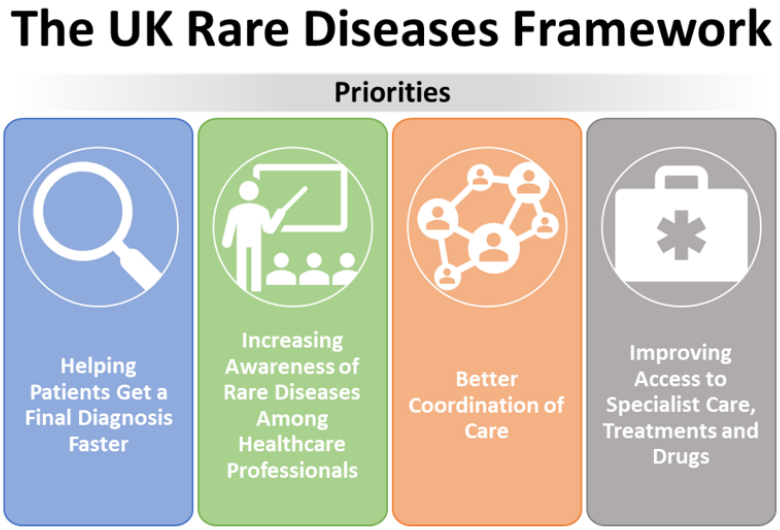
A number of draft actions have been developed by delivery partners with more to come. These are 'in progress' and details of draft actions were uploaded to this folder on the online platform as paper 3 for the UK Rare Diseases Framework Board. We plan to add a questionnaire to gather input on the draft actions proposed by delivery partners. Keep an eye out for this in the coming weeks. We also plan to hold two community roundtables later this year to gather feedback on England's action plan draft.

Scotland

The Scottish Rare Disease Implementation Board has been convened to develop and monitor the implementation of an action plan for Scotland. The group held its first meeting on 5 August, and the papers for the meeting can be found here.

As part of Scotland's new governance arrangements, a Patient Voices Advisory Group is also being formed to help inform the work of the Scottish Rare Disease Implementation Board.Membership recruitment is currently underway.

The group will provide a mechanism to support meaningful engagement and collaboration on the UK Framework Priorities and underpinning themes. This engagement will contribute to the development of Action Plan for Scotland.A programme of digital and virtual engagement that will be taken forward with the rare disease community in Scotland over the summer. The workshops and Patient Group will directly shape the Scotland Rare Diseases Action Plan, if you are from Scotland and want to participate, please email sarah.ogilvie@gov.scot.



POLICY UPDATES (2)

Wales

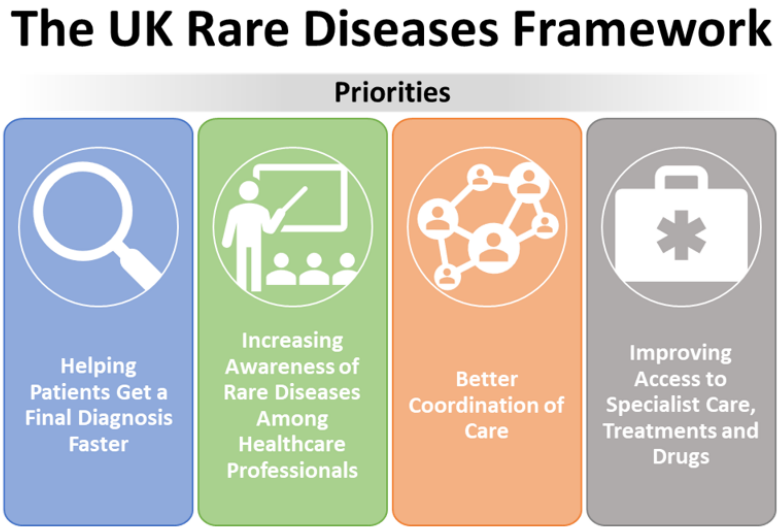
The Rare Diseases Implementation Group is responsible for the development and implementation of the Rare Diseases Action Plan for Wales. The RDIG will work with a dedicated programme manager funded by the Welsh Government, who will be appointed shortly. The RDIG last met on 25 June and reports to Welsh Government. It was established to implement the Welsh Implementation Plan for Rare Diseases (a 5-year plan until 2020). The RDIG's work has previously focused on three key actions:

- Identify and improve the pathway for patients with unknown or delayed diagnosis
- Ensure better use of patient feedback, best practice and evidence to improve pathways for primary, secondary and specialist services.
- Improve reporting of rare disease information including epidemiology, significant event analysis and shared learning.

The Welsh Minister for Health and Social Services has agreed to fund a 2 year pilot of a Syndrome Without A Name (SWAN) clinic, at a cost of £450,000. The SWAN clinic will offer an all Wales service with physical and virtual outreach services provided on a regional basis in south and north Wales.

The key objective of the SWAN clinic would be to shorten the time to diagnosis. Appropriate protocols will be developed with clear referral criteria, agreed in collaboration with the genetics service and colleagues in primary care. The clinic will offer a plan for further investigation and review of patients and will provide information and advice to the referring clinician. The clinic will also be responsible for providing ongoing expert advice to local clinicians if needed (although not direct case management) and increasing awareness and training of staff in the care of patients with rare diseases.

It is anticipated that the SWAN will consist of a paediatric and adult clinic, each running 2 clinics per week for paediatric and adult patients. This would allow 200 patients per year to be seen. At the conclusion of the 2 year pilot, there will be a rigorous assessment, which will inform future service delivery.



POLICY UPDATES (3)

Northern Ireland

The Northern Ireland Rare Diseases Implementation Group (NIRDIG) was established in April earlier this year and is working with stakeholders to develop a local Action Plan for Northern Ireland. The NIRDIG is chaired by our Chief Scientific Advisor Prof Ian Young and hosted by the Department of Health. Key stakeholders include Commissioners, the Public Health Agency, Universities, policy representatives from mental health, social services, primary and secondary care, as well as our local rare diseases patient organisation the Northern Ireland Rare Diseases Partnership (NIRDP). Some of the members participated in the previous UK Rare Diseases Strategy and this continued contribution is very welcome.

NIRDIG has had two meetings to date, the first in May provided an overview of the new Framework, governance structures, agreed Terms of Reference and the way forward. The second meeting was in July, where NIRDP presented its top three priorities for Northern Ireland:

1. Develop a national Registration Service for Rare Diseases in Northern Ireland
2. Dedicated online NI Rare Disease Information Hub
3. Lead Rare Disease Information Coordinator

NIRDIG agreed that further engagement with the wider public on actions for inclusion in the NI Action Plan is required and that a second round of engagement in the New Year, when the Action Plan is developed, would also be beneficial.

Currently NIRDIG is preparing for an online public engagement event for Northern Ireland on the 27th September. This event will consider proposed actions and if any additional actions should be included in the Northern Ireland Action Plan, to provide us with a firm, patient centred basis upon which to develop the Northern Ireland Action Plan. The event will be publicised across a wide range of platforms.
Ongoing engagement with healthcare professionals and clinicians is also planned and NIRDIG is considering how best to take this forward.

EVENTS

Community events

- Findings from the NCARDRS rare disease data collection discovery phase. 23rd September 12.00 – 13.00.
- Cambridge Rare Disease Network: Rare Summit 2021. 7th October.

Governance events

- Wales Rare Disease Implementation Group. 17 September 11.00.
- Public Engagement Event: Development of a Rare Diseases Framework Plan for Northern Ireland - Monday 27th September 12pm – 2pm via Zoom.

The UK Rare Diseases Framework was published on 9th January 2021, with a commitment that each UK nation will develop and publish its own plan detailing the steps to be taken to meet the priorities of the Framework in a way best suited to its population. This engagement event seeks the views of the Northern Ireland rare disease community, healthcare professionals, researchers, commissioners and other interested stakeholders, on priority actions to be included in the Northern Ireland Rare Diseases Framework Plan.

If you live in Northern Ireland and would like to participate, please register in advance:

PLEASE CLICK HERE TO REGISTER

After registering, you will receive a confirmation email containing information about joining the meeting.

- England Rare Diseases Framework Delivery Group Meeting #3. 22 September 09.00.
- UK Rare Diseases Forum Meeting #2. 21 October 10.00.
- England Rare Diseases Framework Delivery Group Meeting #4. 29 October 13.00

Thank you for reading, for any further questions or information, please contact gset@dhsc.gov.uk.